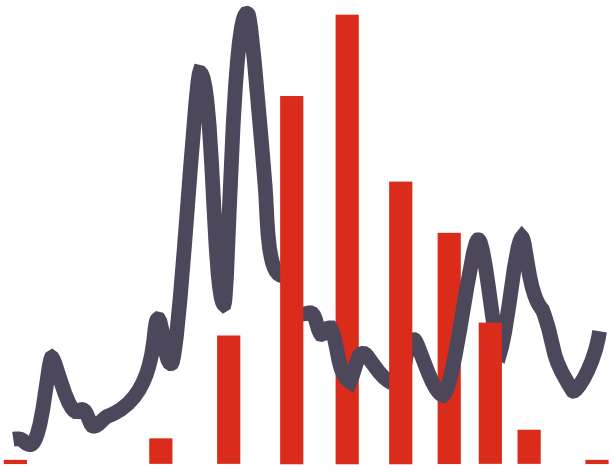
# DNA·VIEW®

# 2019

# User's Manual

revised June 15, 2015
program version 37.06



English

# Introductory Guide

**Installation:** To install use either the installation CD or installation file downloaded from the Internet. The CD should autostart. In either case the installation file has a name like `setupDNAVIEW37.06 ...exe`, which can be invoked by browsing with Windows' "My Computer" if necessary.

There may be a password necessary as part of the installation. Qualified users may obtain it by asking.

See §XV.A, page 295.

**Startup** Normally installation creates desktop icons for starting the program.

See §XV.E, page 303.

**Contact** for information, inquiries, or problems

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**Overview** of DNA·VIEW® — §I.E (page 22) and chapter II (page 29).

**Recent changes** in this manual and in DNA·VIEW — page 3.

**Updates** through the Internet:

Visit the Forensic Mathematics downloads page, `http://dna-view.com/downloads`.

Click on and save a file with a name like `setupDNAVIEW37.06update.exe`.

Request the password.

See **Installation** above.

Update **disks/CD's** available on request.

# The 2019 DNA·VIEW® manual and program

This manual is up to date as of program version 37.06. It is partly updated to 37.47.

The 2015 version added or substantially revised 30+ pages. The manual pdf is available to users at http://dna-view.com/downloads/manuals.

Some highlights include:

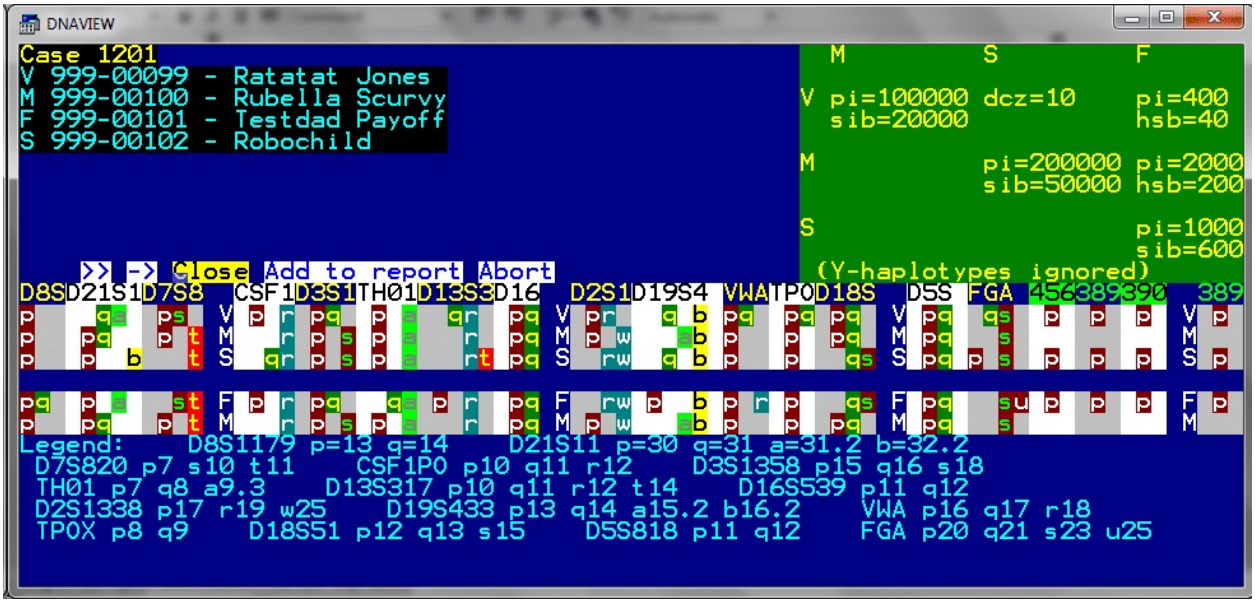
- **Commands & options**
  - 
  - **New** –
    - ☞ *Mixture Solution* (§V.F) is an advanced "continuous model" mixture analysis and computation program which considers peak heights, dropout, and other artifacts via stochastic modeling. Considerably enhanced between 2015 and 2019.
    - *Mixture explorer* (§V.E) – a very fast preliminary survey tool to suggest the likely contributors for mixtures when there are numerous possible reference.
    - *Open Case* (§V.A, V.A)
    - *Paternity Case*: *Calculate trio/duo* (§V.B.6), *Calculate avuncular* (§V.B.4.d), arbitrary prior probability (§V.B.18), θ (§V.B.19)
    - *Kinship*: θ (§VI.F.7.m)
  - **New (research)** – *Covert mutations, Mismatch expectation*
  - **Enhanced** – *Mixture Calculator Automatic Kinship* Y-haplotype calculations.
  - **Renamed** – *Worklist* (was *Membrane*), *Prototype Kinship* (was *Kinship*)
- **New features** mentioned in this manual and new material in the manual are generally indicated with marginal and/or superscript notation. For material that is (new – 2014) see pages 3, 25, 45, 59, 66, 69, 72, 73, 74, 77, 80, 92, 93, 96, 123, 215, 236, 240, 308.
- **New / enhanced 2019 –**
  - *Linked loci* – conservative calculation (per AABB recommendation) to use only one of possibl linked loci such as D5S818 and CSF1PO. Also useful for X haplotypes.
  - More versatile mechanism for *Set Default Database*.
  - *Import DNA profiles* in a custom format for Osiris compatibility.
- **Buttons** on menus (§XIV.A.1); consistent behavior of ***esc*** and ***tab*** among many interesting things.
- **Recent features** – new in the previous few years are marked (new – 2011) see pages 307.
- A few more RFLP bits have been excised. They can be found in old editions available on-line.

***New versions***

Please note the availability of DNA·VIEW updates through the Internet (page 2). If you already use the internet, you know that this is a very convenient way to obtain maintenance and improvements. For Luddite types who prefer the touch and feel of tangible objects, CD's are available on request.

Charles Brenner

June 15, 2015



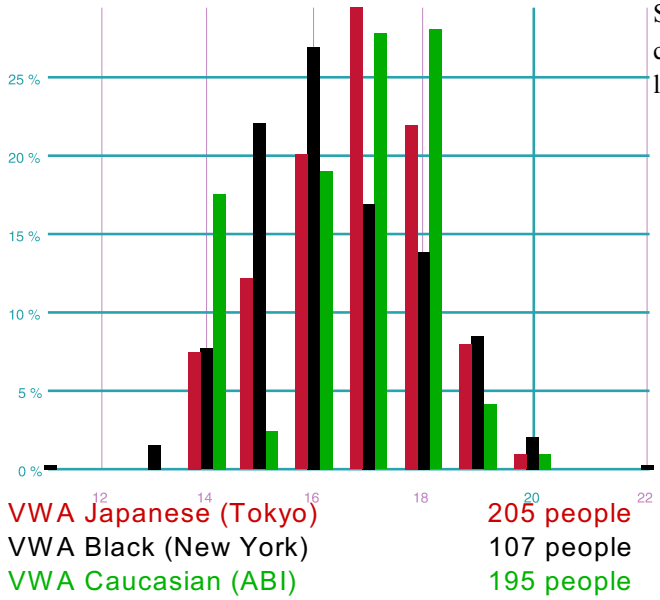

*Automatic Kinship*, *Immigration*, *Estimate likely relationships* (§VI.F.4.b – overview preliminary to kinship analysis) display includes symbolic genotypes.

**Name block** – Individuals in the case are shown with names ("Rubella", etc.) and role letters (V, M, F, S). The roles ("Victim" etc.) have no semantic significance to the kinship program.

**Estimated relationships** – plausibly related people are indicated with a large *pi* ("paternity index" – parent-child relationship) or *si* ("sibling index"). S & V seem to be closely related to M; less so to one another.

**Allele chart –** The letters are assigned to alleles (consistently within each locus). Colors and letter alignment in the allele chart are simply a visual aid. Horizonally scrolling permits viewing all the loci, even the Y loci although they aren't included in this particular calculation.

As you can see from the legend, consecutive letters indicate consecutive alleles (plausible mutation amounts).




Side-by-side *Plot* (§VIII.E, page 171) of VWA compares different populations and different laboratories.

# DNA·VIEW®

## DNA Identification Analysis Programs

# USER'S MANUAL

### Table of contents — overview

# Table of Contents

# List of Figures

# I. GENERAL INFORMATION

## I.A. What is DNA·VIEW?

### I.A.1. Software

DNA·VIEW™ is an integrated software package for DNA identification analysis. Facilities include

*Crime case* calculators – simple & mixed stains

*Parentage case* computation

*Kinship* analysis for any scenarios

Databases – provided & user configurable

Population analysis

Mass fatality identification

DNA·VIEW is written as a DOS program, but as such it of course can run under Windows (but only 32-bit versions).

### I.A.2. Hardware

DNA·VIEW runs on a PC (sorry, no Mac version). Adequate hardware is a Pentium computer with 20Mb of available RAM, color video, 40Mb of disk space, a disk backup device, and a Laserjet-compatible printer with installed fonts. The program may be used on a network by having the program files on a commonly accessible client machine.

DNA data is normally supplied to the program either by importing a text file such as a Genotyper™ allele table.

## I.B. Start Up

Normally installation creates desktop icons and/or Start Menu entries for starting the program.

See §XV.E for fuller instructions.

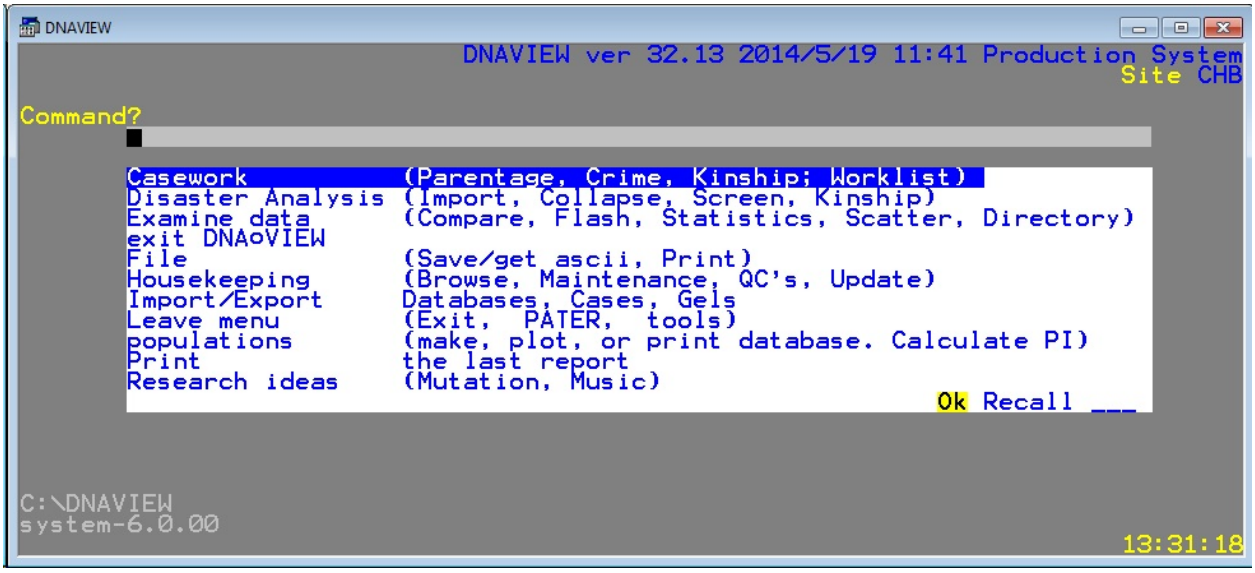



**Figure 1** DNA·VIEW top level command menu

## I.C. Learning DNA·VIEW

### I.C.1. Manual and release note

The information in this edition of the manual is up-to-date as of DNA·VIEW version 37.06. Later features may be described in supplementary documentation available on-line (page 3) and/or printed "DNA·VIEW release notes."

This is primarily a user's manual, and functions both as a reference manual and as a tutorial. Specific information can usually be found with the help of the index.

I.C.1.a. Outline of contents

For the regular user, Chapter I (General) and Chapter II (Operational Procedure) contain general information of an orientation nature that should be read entirely. Chapter XIV (Use of the Keyboard) should also be read entirely. It contains many vital facts, as well as detailed discussion of many shortcuts that make DNA·VIEW much more enjoyable to use. These three chapters are short.

Chapter IV (Tutorial — Casework) exhibits the normal routine for analyzing a paternity case and some other cases in keystroke-by-keystroke detail, includes reference to appropriate sections in the manual, and will be skipped only by the most confident and adventurous users. Chapter III (Tutorial — Various Operations) gives the exact keystroke sequence for initial installation, and for a sample *Kinship* session.

Please be aware of §XI.C (Program Errors).

The remaining chapters, while interesting and ultimately vital, can probably be assimilated as you gain experience.

I.C.1.b. Typographic conventions in this manual

The manual is reasonably consistent in following these rules for the use of various fonts:

`Who are you?` text typed by the program

**007** answer typed by the user

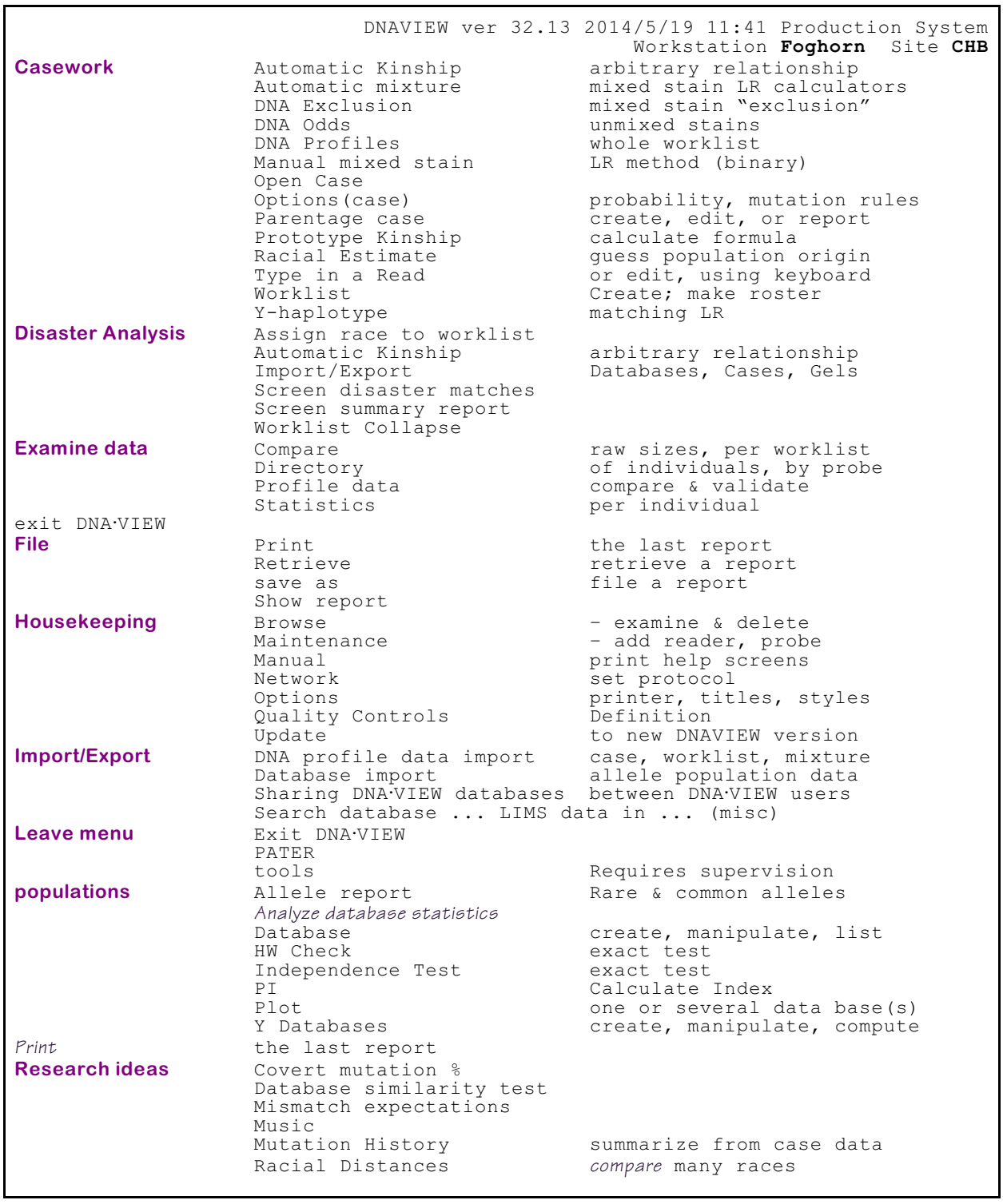



**Figure 2** DNA·VIEW top & second level menu commands

| | |
|---|---|
| ***space enter*** | words that name a key to strike |
| PROPERTIES | a screen button to click |
| *Kinship* | DNA·VIEW program name; a command menu option. (In older sections **Kinship**.) |

| | |
|---|---|
| **Housekeeping** | A command menu (group of commands). |
| *Edit the locus name* | lower-level menu option |
| **properties** | word on a Windows window |

### I.C.2. Program conventions

I.C.2.a. **Menus** in DNA·VIEW are always (usually the background) **green**, and a green background always is a menu, operated as described in §XIV.A.2, page 287.

I.C.2.b. ***Enter*** is usually the right response to any prompt. If a button is highlit (yellow) ***enter*** activates it.

I.C.2.c. ***Esc*** means back up, or abandon the current operation. ***Tab*** and ***shift-tab*** cycle the highlight among the buttons. These are the same as the Windows conventions.

I.C.2.d. A **yellow** banner on the bottom of the screen indicates that the program is waiting for a keystroke from the user to continue to the next step.

I.C.2.e. ***Ctrl-pause*** means **break**; abandon the current operation and return to the main menu. ***Ctrl-c*** usually has the same effect (and is a little easier to type).

### I.C.3. Telephone and fax assistance

are part of the DNA·VIEW license agreement. The numbers are listed on page 2.

### I.C.4. Select program option

Programs are selected from the main (COMMAND) menu shown in **1** and **Figure 2**. The method of selection is intuitive, and is explained in detail in §XIV.A.2. §IX.F.3 explains the method of choosing between the menu styles of the two figures.

## I.D. To Exit

### I.D.1. From a command

After the selected program finishes, the COMMAND menu will usually reappear.

Some of the programs end leaving information on the screen for you to read. In that case there is a yellow banner at the bottom right corner. Then you need to ***Enter*** (or any key) to release the display.

### I.D.2. From graphics mode

DNA·VIEW has some graphics capability mainly for drawing graphs. The graphics mode requires Windows XP or older (or an appropriate virtual system), and requires starting DNA·VIEW with the special DNA·VIEW PLOT icon.

The presence of curved lines or pastels on the video screen means that *graphics mode* is on. ***Enter*** should get you out of graphics mode and into the next program step. If any event ***ctrl-break*** will abort immediately to the COMMAND menu.

### I.D.3. From "special editing mode"

A few programs — especially tools — use the built in APL editor (§XIV.B). To abort editing, ***ctrl-q***.

### I.D.4. From `tools`

From the special state called **`tools`**, strike ***F3*** to return to the main menu.

### I.D.5. To Windows

The menu choice *exit DNA·VIEW* closes DNA·VIEW.

You don't need to close DNA·VIEW to use another Windows process. Certain keystrokes that are recognized by Windows are well worth knowing.

- ***Alt-tab*** switches to the next process in sequence. Hold ***alt*** and tap the ***tab*** key to see the ring of sequential processes and to select the desired one.
- ***Alt-enter*** toggles the DNA·VIEW screen between full-screen and windowed appearance.

### I.D.6. (Avoiding) program delays

In various circumstances the program pauses for several seconds to let the user read a message on the screen. For example, there is a 2-second delay to show the initial banner when DNA·VIEW is started, and a 4-second delay for any (temporary) errors that are encountered during startup when a new system is first installed.

If you do not feel like being patient, you can hurry the program past these delays by hitting ***space***.

See §XV.E.3.a about avoiding delays in *other* Windows operations while DNA·VIEW is running.

## I.E. The world of DNA·VIEW

### I.E.1. Data structures

To understand DNA·VIEW it is helpful to be aware of the various data structures in it.

I.E.1.a. **Cases** (§V.B, page 41 and §XVI.B.2)

The commands *Paternity case*, *Automatic mixture*, and *Automatic Kinship* (§V.B) group individuals into cases. *Screen disaster matches* (§VII.D) expects the references of a family unit to be grouped as a case.

I.E.1.a.i.) Case numbers

Cases are known by case numbers of digits only. Details in §XIV.A.10.

I.E.1.a.ii.) Roles (**Figure 154**, page 311)

Each person in a case occupies a *role* within that case. Roles are single capital letters. Each role letter has a descriptive word associated with it – "mother" associated with the role "M" for example – but for most purposes the roles can be used arbitrarily. The person occupying role "M" need not be a mother.

Only one person may occupy any one role in a case. The role-letter + case-number thus represents an alternative way – besides the sample number – to refer to a sample.

A person may belong to more than one case and/or simultaneously occupy different roles in a single case.

I.E.1.a.iii.) Language

The set of available roles, and the associated descriptive words, depend on the *language* or genre of a case. The possible genres are paternity, maternity, kinship, and crime (**Figure 154**).

I.E.1.a.iv.) Miscellaneous other data associated with a case include:

» case comment (arbitrary and searchable)

» comment per role – typically the name of a person or description of a unknown sample

» computation race(s)

I.E.1.a.v.) NOT part of a case: DNA profiles. The case links to DNA profiles through the individuals (§I.E.1.b.iii) in the case, but the profile data is not stored as part of the case.

I.E.1.b. **Individuals/samples** (§XIV.A.9, §XVI.B.1)

People or samples can be found as a role in some case, or can exist independently of any case as an *orphan*. For that matter, the same person can be in several different cases or even occupy more than one role in one case.

I.E.1.b.i.) Two ways to identify a sample

» If a sample occupies a role in a case, then it can also be designated by using the case number + the role letter (§I.E.1.b.ii).

» Its *sample* (or *accession*) *number* (§I.E.1.b.iii) can be used to identify any sample, including orphans.

I.E.1.b.ii.) Case + role (§X.A.8.b and §XIV.A.9.a.iii)

The notation `61234 M` refers to role `M` of case number `61234`. The number is clearly a case number and not a sample number because it doesn't have a dash in it. This method of identifying an

individual is convenient for preparation of data input files (§X.A.8.c), for user-selection on-screen of a profile for calculation (§XIV.A.9.a.iii), and is preferred by the DNA·VIEW programs for display.

I.E.1.b.iii.) Sample/accession number (§XIV.A.9.a)

The accession number is number with a dash ( – ) in it that is formatted like 2009–01234. There may be up to 4 digits before the dash (a year for example) and up to five digits after. (For data entry purposes leading 0's in either part are not significant.)

Some laboratories like to assign accession numbers that reflect the case number. Hence the mother, child, and alleged father in paternity case 444 might be 2000–04441, 2000–04442, and 2000–04449.

Other laboratories assign consecutive numbers without regard to case. The people accessioned in 2000 might be 2000–00001 through 2000–02468. **Recommendation:** If sequential numbers are used, I advise continuing with 2001–02469 in the next year, etc., so that the sequence part of the number won't repeat for several years. This helps to avoid mistakes.

I.E.1.c. **Worklist**

A *worklist* or *roster* (formerly called *membrane*) consists of a number of *lanes*, which typically correspond to the lanes or capillaries of an electrophoresis run in the laboratory. A worklist may have only a few lanes (minimum of one), or may be defined with thousands of lanes.

Each lane may be labeled with a *sample* (or *accession* or *person*) *number*, as described in §I.E.1.b.iii, or with an *allelic ladder* (in case of GeneScan™ data), or may be unlabeled.

The roster is thus an arbitrary list of people. Each person may incidentally be defined as part of some case (paternity, crime, or kinship), or of several different cases, or of no case at all.

The people in a case do not need to be grouped together on the same worklist. A person may even be assayed on several different lanes or worklists, in which case the user has flexibility in analysis whether to use a particular assay (§V.B.26) or to check for a consensus among all assays.

I.E.1.c.i.) Worklist maintenance

The *Worklist* command (§V.J, page 92) can create or delete or edit a worklist, including editing the roster of sample numbers.

**Import/Export** options such as *Genotyper import* (§X.A.1 page 226) or *Hitachi STRCall import* (§X.A.6, page 232) also can create a worklist and populate the roster.

I.E.1.d. **Read** (DNA genotypes for 1 locus, multiple samples)

Each lane of a worklist potentially has a DNA profile associated with it. The DNA data is stored as *reads*. Each read is the data for one locus and all the lanes of a worklist. Thus if for example the CODIS loci are used a worklist might have 13 or 14 loci associated with it. There is no limit, though, and it is also permitted to have multiple reads for the same locus.

The data for each read is, like the worklist, organized into *lanes* or capillaries. Each lane may have zero or more *bands* or alleles. Bands associated with unlabeled lanes are not accessible by or used by any analysis program, but a lane label can always be added.

Each read has a 3-digit *reader number* associated with it, which identifies the technician responsible for creating it.

I.E.1.d.i.) Read maintenance

The command *Type in a Read* (§IV.D.4) permits manual creation or editing of reads. But usually, if you do need to enter data manually, it's easier to use Bill Gates' fine Excel program and ...

**Import/Export** options such as *Genotyper import*(§X.A.1 page 226) or *Hitachi STRCall import* (§X.A.6, page 232) import allele sizes and save them as reads.

I.E.1.e. **Loci** (§IX.B, page 201)

DNA·VIEW has an extensible list of loci. (Usually users don't need to add new loci though, as all the known ones are already included or are automatically added along with software updates.)

Associated with each locus is a variety of parameters such as repeat amount (e.g. 4 for tetramers), spelling of the name (e.g. vWA or VWA) and notation rules (e.g. STR alleles are usually numbered; SNP alleles are nucleotide letters).

I.E.1.e.i.) Locus maintenance is via the *Probe/locus parameters* option of the *Maintenance* command in the  menu, or by using the ***** key to select a locus from the locus menu in any context.

I.E.1.f. **Races** (§XIV.A.9.a.iii, page 293)

DNA·VIEW accommodates 52 races, corresponding to the letters **a**-**z**, **A**-**Z**, plus one "unknown/child" category denoted by a dash (**-**). The user may associate any race name desired with each letter.

I.E.1.f.i.) Maintenance of the race list is via the *codings for races* option of the *Maintenance* command in the **Housekeeping** menu, or by striking **?** whenever a race letter is to be provided in response to a prompt.

I.E.1.g. Allele probability *database*s

A large number of reference allele population databases are delivered with DNA·VIEW, and more may be added in various ways. There may be several databases even for a particular locus and racial/ethnic group, but only one among them may carry at any given time a special tag denoting that it is the "default" database to be used for probability computations for that race/locus combination.

I.E.1.g.i.) Database maintenance

The *Database* command (§VIII.C, page 158, in the **Populations** menu) has options (among others) to

» *compile a database from DNA·VIEW data* (i.e. compile it from Reads)

» *Edit* or *Type in data to create a database*

» *rename, delete,* or *set default* status of a database

» perform various statistical analyses

The *Import/Export* command has options to import a database in various formats.

*Plot* creates graphs and histograms of allele frequencies.

### I.E.2. Casework Analysis

DNA·VIEW has several commands to analyze cases of different kinds and in different ways. Each of them operates on a selected *case* (§I.E.1.a) that has a collection of roles with sample numbers attached. Assuming that the sample numbers also appear in some worklist roster (§I.E.1.c), the DNA profiles for the various individuals in the case are brought together for the analysis

I.E.2.a. **Paternity/maternity case**

The typical paternity trio case, or common variations of it such as missing mother or maternity, are most easily and thoroughly analyzed by the *Paternity case* command (§V.B, page41). It operates on a has

roles designating mother (usually), child or children, and alleged man or men. Per trio it performs the analysis locus by locus and prepares an informal and a formal report of the results.

I.E.2.a.i.) PATER interface

Additionally, the results of the analysis are written to an external table (documentation available) which can be conveniently interpreted by another program. The PATER program is one option for creating a suitable lay-person/court friendly report.

I.E.2.b. **Kinship**

The paternity trio is one example of an infinitude of *kinship* problem – deciding how strongly the DNA evidence favors one or another arbitrary alternate hypotheses as to how a collection of people might be related.

The general problem is analyzed by the *immigration/kinship* (§VI.E, page 109) sub-option of *Automatic kinship* (or for that matter of *Parentage case* or *Automatic mixture*).

I.E.2.c. **Mixed stain** (revised – 2014)

*Automatic mixture* includes several tools for mixture analysis:

I.E.2.c.i.) *Mixture calculator* (§V.D, page 65) computes a likelihood ratio for a mixed stain under the "binary model" (each allele is present or not).

I.E.2.c.ii.) *Mixture Solution* (under development) is a full-fledged "continuous model" (dropout, intensity, stutter, drop-in aware) mixture calculator.

I.E.2.c.iii.) *Mixture explorer* (experimental) very quickly guesses which references are associated with a mixture.

### I.E.3. Disaster Screening

*Screen disaster matches* (§VII.D) and other programs including *Automatic Kinship* are useful for mass fatality investigations.

### I.E.4. Single-profile or manual crime stain analysis

There are also several options for crime stain analysis that operate either on a single individual named by sample number, or on a DNA profile typed immediately in.

I.E.4.a. **profile probability – calculate a simple stain**

The *DNA Odds* command (§V.G, page 84) computes profile odds for a single profile at a time, and allows a variety of computation options such as those recommended in the NRC II report, for autosomal loci.

*Y-haplotype matching LR* (§V.H, page 87) performs a similar computation for Y-haplotypes.

I.E.4.b. **mixtures**

*DNA exclusion* (§V.I, page 89) computes the "probability of exclusion" for a specified mixture of alleles at a collection of autosomal loci.

The stand-alone *Manual mixed stain*, like the *symbolic mixture calculator* option of *Automatic mixture*, computes a likelihood ratio for a mixture. However, it works strictly on typed-in data.

### I.E.5. Prototype *Kinship*

*Kinship* (§VI.J, page 134) is a stand-alone command that is the original, prototype version of the automatic version (§I.E.2.b).

### I.E.6. Database analysis

Several commands do population or population-genetic analyses.

I.E.6.a. *Racial Distance*

I.E.6.b. *HW Check* and *Independence test*

These commands test population studies for evidence of non-independence. The input is in from a text file.

I.E.6.c. **Database statistics**

The command (also *Database* option) *Analyze database statistics* and the command *Allele report* compute various statistics and summaries based on DNA·VIEW databases.

*Y-haplotype database statistics* (§VIII.F.5) computes useful statistics for the Y databases.

### I.E.7. Reports in DNA·VIEW

Many DNA·VIEW operations produce a report. The report resides in the "report buffer" until it is supplanted by another report. In the meantime, the report can be (§V.K) printed (*Print*), viewed (*Show report*), or saved as a file (*Save as*).

### I.E.8. Where to Find It — Sub-menus

Many commands do a specific thing. Others do a number of related operations, as indicated in these sub-menus:

*Browse* – (examine & delete) *Reads, Lanes, Standards Ladders, Configurations*

*Options* – control printing, report formats, national *Keyboard* & *Language*. Set *Case Options*.

*Paternity/Automatic Kinship/Automatic mixture* – retrieve, modify, analyze cases. Set *Case Options*. Kinship and simulations.

*Maintenance* – lists etc. Examine & modify loci, races, network preferences. Paradox info, maintenance. Record/compare "switch" settings.

*Database* – allele population samples. Compile, recompile, *Analyze database statistics, Delete or rename, Edit a database, Set default database, per race, Type in data to create a database*

*Plot* – data base(s); *DISPLAY* (screen), *METAFIL* (file) printer, *more plots, fewer plots, probability of paternity*

*Quality Control* – *add control, disenroll control, edit stats, range report, recompute*

*Import/export* ▸ DNA profile data import

ABI import: *GeneMapper, Genotyper*

*Hitachi STRCall import*

*Import/export* ▸ Database import from table

import publication format: Allele frequency (or count) population database import

import reference genotypes and build tables

*Import/export* ▸ Sharing DNA·VIEW databases

transfer between users: *DNA·VIEW Frequency Database import* and *export*

*Import/export* ▸ miscellaneous

*List and search database profiles*

*Case and Accession data import*

**I.E.9. Where to Find It — Guide to DNA·VIEW**

| SUBJECT | ACTION | COMMAND |
|---|---|---|
| Reports | print<br>save<br>draw | *Print*<br>*Save as*<br>*Plot* |
| Readers | add, delete, modify | *Maintenance* |
| Probes | add, modify | *Maintenance* |
| Standard ladders Configura-tions | add<br>delete<br>examine<br>define alleles | *Maintenance, Worklist, Read, PCR Read*<br>*Browse*<br>*Browse, Maintenance*<br>*Maintenance* |
| Worklists | add<br>modify<br>delete<br>examine | *Import/Export*, *Genotyper/GeneMapper import*<br>*Worklist, Type in a Read*<br>*Worklist*, *Browse*<br>*Worklist Sizes* |
| Reads | create<br><br>delete<br>edit | *Type in Read*, *Import/export Genotyper/GeneMap-per import*<br>*Worklist*<br>*Type in Read* |
| Lanes | create<br>delete | *Type in a Read*, *Worklist*<br>*Browse* |
| Data Base | create<br>examine<br>delete<br>exchange | *Import/Export*, *Database*<br>*Plot, Database*<br>*Database*<br>*Import/Export* |
| Computa-tions | Sizing<br>paternity index<br>set parameters<br><br>change data base defaults<br>independence of data<br>stains & mixed stains | *Import/export ABD GeneScan import*, *Read*<br>*Paternity Case*, *Kinship*<br>*Maintenance* (also *Paternity/Crime Case, Auto-matic Kinship*)<br>*Database*<br>*HW exact test, Independence test*<br>*Automatic mixture, Manual mixed stain* |
| Case | define, modify<br>list, compute | *Case, Import/Export*<br>*Case, Kinship* |
| Individuals | define, modify<br>list<br>monitor | *Case, Worklist, Statistics, Import*<br>*Case, Directory, Quality Control*<br>*Quality Control* |

# II. OPERATIONAL PROCEDURE

## II.A. Outline of Operations

Suggested orders of DNA·VIEW operations for casework:

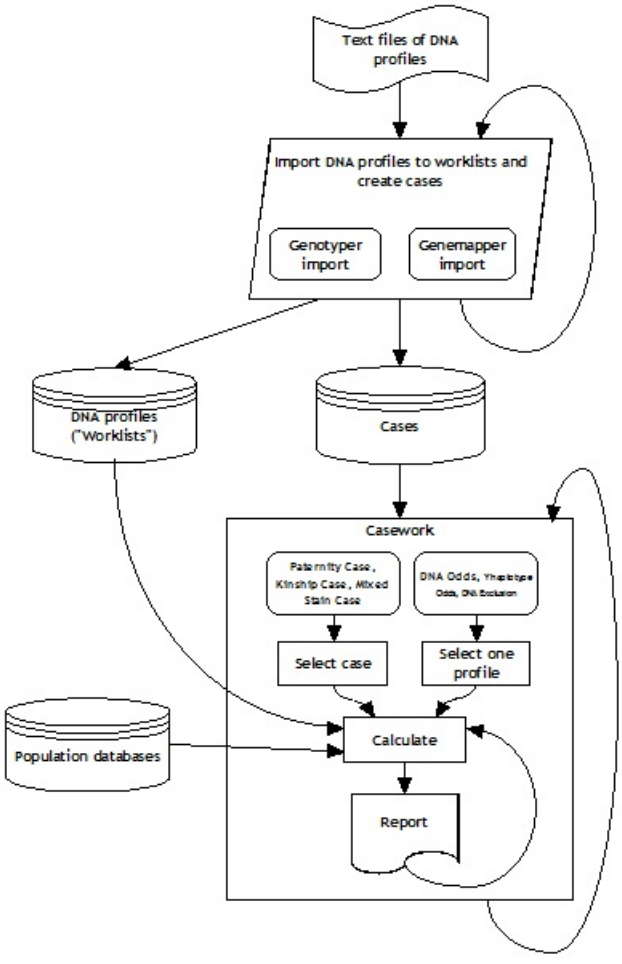



**Figure 3** Schematic workflow for casework

### II.A.1. Routine for casework – very efficient method

II.A.1.a. Use *Import/export* ▸ *Genotyper* (*GeneMapper*, etc) *import* to import the DNA profiles and to defined the people and cases at the same time.

II.A.1.b. For paternity cases, go to *Paternity case*

II.A.1.b.i.) *batch select, add the cases from a worklist* to prepare a batch run for all the imported cases,

II.A.1.b.ii.) *batch run* to make all the calculations and create report files.

II.A.1.c. For other cases, use *Automatic mixture* or *Automatic kinship* for each case to retrieve all the data, summarize it, and make appropriate kinship or matching index calculations.

### II.A.2. Routine for casework – second best method

II.A.2.a. Use *Paternity/Automatic mixture* or *Automatic Kinship* (§V.B/§V.D, §VI.F) to define one or several cases, and the people in them.

II.A.2.b. Use *Worklist* (§V.J) to define a roster of assignments of people to lanes.

II.A.2.c. Use *Type in a Read* (§IV.D.4) to enter DNA profiles.

See tutorial §IV.A for examples.

### II.A.3. Occasional operations

II.A.3.a. **Maintaining allele frequencies** includes some of the following tasks:

II.A.3.a.i.) Compiling the accumulated casework (or other) data into sets of allele population frequencies is done using **Database** (§VIII.C). **Compare** (§IX.H), **Statistics** (§IX.D ), and **Browse** (§IX.E) are used to analyze problems and clean up the data, as explained in §VIII.C.8.a, "Analyzing exception reports."

II.A.3.a.ii.) *Import/Export* (automatic) and *Database* (manual) are used to install databases from other sources.

II.A.3.a.iii.) Various analysis and statistical capabilities are in *Database*, *HW Check*, and *Allele report*.

II.A.3.a.iv.) *Plot* (§VIII.E) is a very useful tool for gaining knowledge and insight.

## II.B. Preliminaries — details

More details of the steps discussed below are given in the Tutorial (Chapter IV). These setup operations have been pre-configured as part of system customization insofar as possible, and the remaining ones can more or less be postponed.

### II.B.1. List of loci

The locus list is probably adequately preconfigured as part of system customization. It can also be configured "on-the-fly" – i.e., as needed. §IX.B explains loci in DNA·VIEW. The critical issues are

II.B.1.a. Locus list arranged as desired. The loci are most conveniently and easily activated and arranged in order with the REORDER: MPLX button (§IX.B.1.d.i). See also §IX.B.1.e.

II.B.1.b. Set parameters

Key parameters for PCR systems include: repeat amount, notation style (i.e. numbered alleles, SNP notation, Amelogenin, Polymarkers), mutation rate.

The parameters can also be defined or modified any time the locus menu appears (via PROPERTIES).

### II.B.2. List of races

The race list can be accessed via the EDIT RACES button in response to a race-letter request by the program, so pre-configuration is unnecessary. You can also use *Maintenance*, *codings for races* to make sure that race names exist for each race letter that you will use.

### II.B.3. Default data base

Whenever allele probabilities are needed as part of some computation in DNA·VIEW, the program checks to see if a "default" has been established for the locus and race at issue. The facilities for default databases are explained in §VIII.C.2.

### II.B.4. List of readers

"Readers" are technicians who use the program. Readers can be added as needed on the fly (§IX.A.5.b), or a list of them can be built as a separate operation using the *Maintenance* command in **Housekeeping**, option *readers* – §IX.A.5.

### II.B.5. Define Quality Controls

To take advantage of quality control monitoring, use **Quality Control** (§IX.J) to enroll accession numbers for quality control samples.

### II.B.6. Miscellaneous options

Several of the *Options* §IX.F (**Housekeeping** menu), may be useful.

II.B.6.a.i.) They include formatting of dates, case numbers, numbers (i.e. 1.35 vs. 1,35) and alleles; keyboard configuration, language for prompts, printer options.

II.B.6.a.ii. Especially check the *Case* options, §V.C. Select *Options*, *Options from "Case"*, and make sure that all the values are acceptable.

# III. TUTORIAL — VARIOUS OPERATIONS

This chapter gives the keystroke sequences for various operations.

## III.A. Quick installation

Insert PROGRAM CD or download the Setup file from the internet. The CD is supposed to autostart. The same effect can be obtained by double-clicking, in the Explorer, on the `SetupDV... .exe` file..

| keystroke(s) | reason |
|---|---|
| ***Enter*** | =Next, to continue |
| **`C:\dnaview`** ***enter*** | location to install (or other desired choice) |
| (click **Install**) ***enter*** | Original installation (as opposed to Update) |
| **`DnaView`** ***enter*** | folder in the Start Menu for shortcut |
| ***enter*** | Create DNAVIEW ☑ desktop icon, ☑ start menu entry |
| ***enter*** | Install |

---

## III.B. First execution of DNA·VIEW

| keystroke(s) | reason |
|---|---|
| ***enter*** | = Yes; allow the "update" to proceed (§XI.B) |
| ***enter*** | = ok to continue |
| ***enter*** | = **Y**es |
| ***enter*** | to see news/help |
| ***esc*** | escape from news |

## III.C. Practice session with (stand-alone) *Kinship*

| keystroke(s) | reason |
|---|---|
| **Cas** *enter* | **Casework** |
| **Pr** *enter* | *Prototype Kinship* (§VI) |
| **R** *enter* | read a `*.KIN` file |
| **examples** *enter* | `EXAMPLE` sub-directory |
| **Tr** *enter* | Trio example case |
| **F** *enter* | *Find formula* (which is 1/2b) |
| *enter* | continue |
| *enter* | *Evaluate* |
| **0.1** *Esc* | b=0.1; LR=5 |
| *enter* | continue |
| *enter* | *Post this locus* |
| *enter* | *Locus* |
| **V** *enter* | VWA |
| **E** *enter* | *Edit* |

change genotypes on line [7] to read

**Child a : Mommy a + Daddy a / ?**

| | |
|---|---|
| *Esc* | finished changing |
| *enter* | continue |
| *enter* | *Find formula* (which is 1/a) |
| *enter* | continue |
| *enter* | *Evaluate* |
| **.1** *space* **0** *esc* | a=0.1; LR=10 |
| *enter* | continue |
| *enter* | *Post this locus* |
| **L** *enter* | *Locus* |
| **Tp** *enter* | TPOX |
| **D** *space* **c** *enter* | *Display case summary* |
| *enter* | continue |
| **F** *down enter* | *File (or fetch) complete case* |
| *enter* | *(a new case)* |
| **test case** *tab* | name, and LR; save the analysis |
| *enter* | continue |
| **Pr** *enter* | *print the case summary* |

---

## III.D. Finished!

*Ctrl-pause* break; return to menu

**Exit** *enter* Leave DNA·VIEW

# IV. TUTORIAL — CASEWORK

## IV.A. Paternity and Kinship Casework

The following sections demonstrate – keystroke by keystroke – the various common DNA·VIEW operations for running paternity and kinship cases.

The shaded sections indicate a few less-common operations that are included.

### IV.A.1. Preview

IV.A.1.a. Preliminaries

There are various operations that may need to be done infrequently, once, or even not at all because of pre-configuration of the software. These include:

IV.A.1.a.i.) Defining a locus

Probably the loci are already defined. Nonetheless directions for defining a locus are included.

IV.A.1.a.ii.) Defining a "reader" (technician who uses the program)

IV.A.1.b. Data entry

Two approaches are shown for data entry – manual (§IV.D) and imported (§IV.B.1).

IV.A.1.c. Case analysis

Two approaches are shown for analysis – the paternity-specific method using the *calculate paternity/maternity* of *Paternity Case*, and the generalizable method using the *immigration/kinship* option.

### IV.A.2. Sample case

For purposes of the example, assume the following data:

| Sample Info | Comment | D3S1358 | | VWA | | FGA | | D8S1179 | | D21S11 | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| 101 M Hilda | investigation of | 15 | 17 | 15 | 18 | 25 | 25 | 12 | 14 | 30 | 30 |
| 101 C Sonny | paternity | 15 | 17 | 15 | 19 | 24 | 25 | 12 | 14 | 30 | 30.2 |
| 101 D Cher | [lang=p races=cb] | 15 | 15 | 18 | 18 | 22 | 25 | 12 | 15 | 30 | 30.2 |
| 101 F Mr. Z | | 15 | 17 | 15 | 19 | 22 | 24 | 14 | 15 | 29 | 30.2 |

## IV.B. Fast, typical method

The Sample Case (§IV.A.2, page 35) data may be found as part of the installed DNA·VIEW system as `\dnaview\examples\101.txt` (but with a full set of 13 loci rather than the handful pictured above). If not, then prepare it as follows:

1. Type the data into an Excel spreadsheet.
2. Save as `\dnaview\examples\101.txt`

   Save as type: tab-delimited text

The flowchart **Figure 3**, page 29 illustrates the steps below.

### IV.B.1. Import the data

| keystroke(s) | reason |
|---|---|
| **I** *Enter* | *Import/Export* command (Chapter X) |
| **D** *Enter* | *DNA profile data import* |
| **G** *Enter* | *Genotyper import* (§X.A.1) |
| **examples** *Enter* | `Genotyper import from what folder?` (§X.A.2.b) |
| **101.txt** *Enter* | select the file `101.txt` (which has the data for case 101) |
| *Enter* | to add a NEW worklist |
| *Enter* | today's date is OK (***Space*** through month, day, if you want to edit the date.) |
| *Enter* | "`101.txt`" is OK as a comment |
| *Enter* | `Ok so far?` NEXT>> |
| *Enter* | User number `099` is OK |
| *Enter*=NEXT>> | assuming the menu of column heading-locus correspondences is ok. Otherwise, |
| *enter* | to CHOOSE LOCUS for any item needing resolution (§X.A.2.g) |
| **y** | YES `Proceed with import?` |
| *Esc* | through viewing the import report |
| *Ctrl-c* | ABORT – return to top menu. |

The above importing simultane

ously creates a case numbered 101, and creates a roster and imports DNA profiles. To finish this exercise,

### IV.B.2. Compute paternity indices

| | |
|---|---|
| **C** *enter* **Pat** *enter enter* | **Casework** ▸ *Paternity Case* ▸ *select case* (§V.B) |
| **101** *enter* | select case 101 (§V.B.28) |
| *enter* | *calculate paternity/maternity*. (Independent trio evaluations are made for the two children.) (§V.B.7)<br>(database selection may be requested per §VIII.C.2.b.ii)<br>(file creation name/confirmation may be requested per §V.C.5) |
| *Enter* | response to the yellow "wait" banner |
| *Enter* | *print report* (but see §XV.D.2.a.v re USB printer) |
| *Ctrl-break* (or *Esc*) | Revert to main menu. |

## IV.C. Practice (automatic) Kinship Case

See §VI.E, page 109.

For this exercise assume that case 101 has been imported per §IV.B.1.

DNA profiles have been defined for four people: M, C, D, and F. As a practice problem, we consider the full- vs. half-sibling question:

> Assuming that C's mother and father are M and F, and that D's mother is M, compare the scenarios:
>
> $H_1$: D's father is also F  $H_0$: D's father is some unrelated person

| keystroke(s) | reason |
|---|---|
| **C** *enter* **Pat** *enter enter* | **Casework** ▸ *Paternity Case* ▸ *select case* (§V.B, §V.B.28) |
| **101** *enter* | select case 101, OK |
| **/** *Enter* | *immigration/kinship* (§VI.F), OK |
| *Enter* | *Estimate likely relationships* is interesting (§VI.F.4.b) |
| *Enter* | Accept the default genotype display ordering. |
| *Tab Enter* | ADD TO REPORT – save the estimated relationship analysis |
| *Enter* | *Type in scenario 1* (§VI.F.1) |
| **C : M + F** *Enter* | C is child of M and F (See §VI.C) |
| **D : M + F/Joe** | D is child of M, and F or some Joe (See §VI.C) |
| *Ctrl-enter* | OK; finished entering scenario |
| **Prior** *Enter* **.5** *Enter* | Let prior probability = ½. (§VI.C.6) |
| **C** *Enter* | *Calculate & report LRs, 3 races* (§VI.F.2) |

```
Caucasian cumulative LR       71800
        Posterior probability=0.999986, assuming prior=0.5
Hispanic  cumulative LR      174000
        Posterior probability=0.9999942, assuming prior=0.5
Black    cumulative LR       79900
        Posterior probability=0.999987, assuming prior=0.5
```

| | |
|---|---|
| **reports/Kin101.txt** *enter* | Filename for posting [as a formatted report] (per §V.C.5.c.i) |
| | Note: The LR attributed to VWA comes from an ad-hoc "mutation calculation" that may not be realistic (§VI.F.7.l). |
| *Enter* | finished with yellow waiting banner |
| **log** *Enter* | *Show (log) report* |
| *left-click* PRINT button | Print the "log" report, (not the formatted one per §VI.F.2.a) |
| *Ctrl-c* (or ABORT button) | Revert to main menu. |

## IV.D. Manual methods

### IV.D.1. Practice session – modify case definition

IV.D.1.a. Open the case

| keystroke(s) | reason |
|---|---|
| **C** *enter* | **Casework** menu |
| **Pat** *enter* | *Paternity Case* command (§V.B) |
| *click* OPTIONS | enable the *option* menu choices |
| **Name** | view the menu item `Name tag style: NAME/DATE` (§V.B.21) **(rare operation)** |
| *Enter* (=OK) or *Bksp* (=___) | change it to `NAME`, or leave it if already `NAME` |
| **s** *enter* | *select case* (§V.B.28) OK |
| **101** *enter* | open case 101 OK |

IV.D.1.b. Add another person

| | |
|---|---|
| *alt-e* **a** *enter* | edit menu; *add role* (§V.B.3) OK |
| **G** *enter* | new person is role G – man #2 (§XIV.A.9.a.iii) |
| **2003135** *space* | Define G's accession number as `2003-00135`. |
| **c** | his race=c (note: instead of **c**, you can hit **?** to see and to edit the race list) |
| **Herman** *enter* | his name; finished editing |

IV.D.1.c. Delete a person

| | |
|---|---|
| **pe** *enter* | *edit people* (§V.B.12) |
| *down-arrow down-arrow* | move to third person (Cher, child #2) |
| *del* | DEL, away with her |
| *end* | DONE editing |

IV.D.1.d. Define computation races for the case

| | |
|---|---|
| **ra** *enter* | *edit race list* (§V.B.13) |
| **chb** *enter* | Choose Caucasian, Hispanic, and Black for possible computations |
| *Ctrl-c* | revert to top level menu |

### IV.D.2. Practice session – create empty case #14

I find it useful to have an empty case – no people, no DNA – available for simulations (§VI.G). Create it with the following steps.

| | |
|---|---|
| **C** *enter* | select **Casework** |
| **au** *enter* | *Automatic Kinship* |
| *enter* | *select case* |
| **14** *enter* | case #=14 |
| **c** *enter* | define computation race(s) as Caucasian |
| *end* | DONE; don't create any roles (§V.B.3). |
| *Ctrl-c* | back to top menu |

### IV.D.3. Practice session – modify a roster

| | |
|---|---|
| **C** enter **W** enter | **Casework** ▸ *Worklist* command (§V.J) |
| **101.t** *enter* | Select the "101.txt" worklist (§V.J.3) |
| **5** | move the red bounce-bar to lane 5. |
| *Click* CASE | We'll add a person who is already in a case. |
| **101** *enter* | from case **101** OK |
| **G** *enter* | select *G Herman* OK (§XIV.A.9.a.iii) |
| *click* NEXT>> | man is added. |
| *Click* FILE & QUIT | save changes |
| *Ctrl-c* (ABORT) | Done defining; revert to main menu. |

### IV.D.4. Practice session – modify DNA data

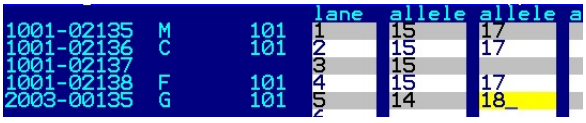
| keystroke(s) | reason |
|---|---|
| **C** *enter* **T** *enter* | **Casework** ▸ *Type in a Read* command (§IV.D.4) |

IV.D.4.a. Add a reader **(infrequent operation)**

| | |
|---|---|
| NEW | new reader |
| **17** *enter* **F. Bacon** *enter* | Add F. Bacon as "reader" (technician) 017. |
| **17** *enter* | Sign in as reader 17. |
| **101** *enter* | Select worklist. |

IV.D.4.b. Type in alleles

| | |
|---|---|
| **D3** *enter* | select D3S1358 |
| *left-click* allele column of lane 5 | |
| **14** *space* **18** | G is 14, 18 |
| *Esc* | Done with DNA screen. |
| **F** *enter* | *File this image* |
| **n** | `Another locus for this worklist?` **no** |




## IV.E. Crime stain practice

### IV.E.1. profile computation – *DNA odds*

See *DNA odds*, §V.G.

| keystroke(s) | reason | |
|---|---|---|
| **C** *Enter* **DNA od** *Enter* | *DNA odds* command | |
| *Ctrl-enter* | Options (§V.G.2.a.v) | |
| *Enter* | *person* | |
| *click* COPY FROM A CASE | about to select a person/sample | |
| **101** *Enter* | `... (M) of case #` | |
| **M** *enter* | `Copy whom?` (§XIV.A.9.a.iii) | |
| **c** | `Computation race?` **c** | profile computation is displayed |
| *Ctrl-enter* | Options (§V.G.2.a.v) | |

| | |
|---|---|
| ***click*** PRINT REPRT | Print report |

To modify the computation, use the mouse (**single** click to navigate) to select fields such as race, modality, theta, etc.

| | |
|---|---|
| ***Ctrl-c*** | Revert to top menu |

## IV.F. Some maintenance operations

IV.F.1.a. Establish locus multiplex **(rare operation)**

| | |
|---|---|
| **H** ***enter*** | Select the **Housekeeping** menu |
| **M** ***enter*** | *Maintenance* command (§IX.A) |
| **l** ***enter*** | *locus parameters & preferences* (§IX.B) |
| REORDER: MPLX | Look at menu of multiplexes. (§IX.B.1.d.i) |
| **Ident** ***enter*** | Ensure Identifiler loci are active and PUT THESE AT TOP of the menu |
| **D3S** | Highlight the locus D3S1358 |

IV.F.1.b. Define parameters for a locus **(rare operation)**

| | |
|---|---|
| PROPERTIES | properties screen for the locus D3S1358 |

IV.F.1.b.i.) Define the pseudo-enzyme for the locus as "STR"(probably redundant)

| | |
|---|---|
| **Ass** ***enter*** | *Assign enzyme (or PCR status) to this locus* |
| **STR** ***enter*** | Define the locus as an STR |

IV.F.1.b.ii.) Establish the repeat amount and the (often irrelevant) amplicon size

| | |
|---|---|
| **P** ***enter*** | *PCR parameters* |
| **66** ***enter*** **4** ***enter*** | 66=size of "allele #0", 4=tetrameric STR |
| **0** ***enter*** **0** ***enter*** **0** ***enter*** **0** ***enter*** | choose notation style=STR, plus some often irrelevant parameters |
| **1** ***enter*** | Genetic type is "autosomal" |

IV.F.1.b.iii.) Discrete allele system (i.e. measurement uncertainty=0)

| | |
|---|---|
| **W** ***enter*** **0** ***enter*** **0** ***enter*** **0** ***enter*** **0** ***enter*** **0** ***enter*** | *Window size* parameters (measurement uncertainty etc.) |
| **C** ***enter*** | *Copy window-type params to similar loci* (i.e. other STR's) |
| ***Esc esc esc*** | finish with locus properties, with exercise |

# V. CASEWORK ROUTINE

The programs discussed in this chapter are those used for every case. *PI* (§XII.B) is also part of the casework routine for DNA·VIEW Abridged system users.

## V.A. *Open Case*

**Casework** ▸ *Open Case* presents a menu with a unified list of all DNA·VIEW cases – paternity, maternity, kinship, and crime – from which you may select to open the case. The same selection menu can also be obtained by invoking any of the commands *Paternity/Automatic Kinship/Automatic mixture*, then *select case* and click the SEARCH button. It is often a useful convenience to select a case using the case menu because the menu display includes case comments. Therefore because of the way DNA·VIEW menus work, you can often find a case of interest by typing a key word (such as **incest**) in the comment.

## V.B. *Paternity/Automatic Kinship Case*

The *Paternity/Automatic Kinship Case* commands let you create, modify, or calculate a paternity or any kinship case. Therefore these (very similar) commands embody possibly the most important DNA·VIEW function.


**Figure 5** *Case* nametag matrix and subcommand menu

### V.B.1. What is a Case?

V.B.1.a. Components of a Case

See §I.E.1.a for a description of a Case as a collection of individuals – people, samples, or DNA profiles – each of whom occupies a slot called a *role* (§I.E.1.a.ii). In addition, the case includes:

V.B.1.a.i.) an optional *name* or description associated with each individual/role

V.B.1.a.ii.) an optional name or comment string associated with the case

V.B.1.a.iii.) a list of one or more computation race letters.

V.B.1.b. Purpose of a Case

A case is a way of grouping together a collection of individuals who may be considered together for purposes of relationship or mixed stain analysis.

### V.B.2. Operation of the *Case* commands

MENU: BASIC:
§V.B.7 *calculate paternity/maternity*. . . . 47
§V.C *case options*. . . . 58
§V.B.10 *delete case*. . . . 50
§VI.F *immigration/kinship*. . . . 110
§V.B.15 *language is:*. . . . 52
*quit*
§V.B.28 *select case*. . . . 56
§VI.G *simulation*. . . . 124
§V.D *mixture calculator*. . . . 65
MENU: CALCULATE includes *immigration/kinship* & *simulation* & *mixture calculator* above and also
§V.B.4 *batch run*. . . . 44
§V.B.4.d *calculate avuncular*. . . . 45
§V.B.6 *calculate trio/duo*. . . . 45
§V.B.8 *calculate family trio*. . . . 49
§V.B.9 *compute one locus*. . . . 49
§V.B.30 *parentage test examples*. . . . 57
§V.D.9 *mixture examples*. . . . 76
MENU: EDIT includes several of the above and also
§V.B.3 *add role*. . . . 43
§V.B.10 *delete case*. . . . 50
§V.B.11 *edit case comment*. . . . 50
§V.B.12 *edit people*. . . . 50
§V.B.13 *edit race list*. . . . 50
MENU: REPORT includes *batch run* above and also
§VI.F.7.c *display raw sizes (=Eyeball check the raw sizes)*. . . . 120
§V.B.14 *export case*. . . . 51
§V.B.17 *list cases*. . . . 52
§V.B.24 *recap*. . . . 54
§V.K.3 *review/print report*. . . . 96
MENU: SELECT includes several of the above and also
§V.B.4 *batch select*. . . . 44
§IX.B.1.d *reorder loci*. . . . 201
§V.B.26 *restrict data*. . . . 55
MENU: OPTIONS includes *Case options*, *language is:*, *reorder loci* above and also
§V.B.20 *locus parameters*. . . . 53
§V.B.21 *Name Tag style: NAME/DATE*. . . . 53
*Null frequency: AUTO/Manual*

MENU: ALL includes everything above

**Figure 6** *Case* menu options

The various choices are selected from the *Case* menu, explained below. The usual first step is *select case* and that choice from the sub-menu is therefore highlighted so that you can immediately press ***enter***. Typical operation is as follows:

V.B.2.a. to define a case

V.B.2.a.i.) manually (see tutorial, §IV.D.1)

» Choose a *language* (§V.B.15). *Language is: kinship* is the most flexible choice because then *add role* (§V.B.3) will present all 26 letters in the menu of roles that you can add. *Language is: paternity* is suitable for actual paternity cases (trio, duo).

» *select case* — type in the case number, and type in the accession numbers

» If there are more than the usual 1 man, 1 child, use *add role* (from the EDIT menu)

» If multiple race computations are desired, or if the "computation race" somehow appears to be incorrect, *edit race list* (from the EDIT menu).

V.B.2.a.ii.) automatically

» Use *Import/Export*, *Genotyper/GeneMapper Import* (§X.A.1, also see tutorial, §IV.B.1).

V.B.2.b. to compute a paternity case

*select case* — type in the case number. Then, either press ***enter*** a total of three times, or (shortcut), simply press ***tab*** one time to complete these operations:

*calculate paternity/maternity* performs all analysis and computation across all loci, several races.

*print report* to produce a paper copy of all results, including explanations of any unusual circumstances, raw data (the band sizes), databases used for computations.

The sub-menu also includes many other options. See **Figure 5**. Some are for special control of the computation for a particular case, some make special reports, others fall under the rubric of maintenance operations.

The following pages describe them in alphabetical order, as they appear in the sub-menu.

V.B.2.c. to compute a kinship or immigration case

(Typically it is slightly more convenient to access *immigration/kinship* via *Automatic Kinship* rather than *Paternity Case*, but the operation is nearly identical.)

*select case* – as §V.B.2.b above

*immigration/kinship* – for arbitrary relationship analysis. See Chapter VI.

*print report*, export it, etc.

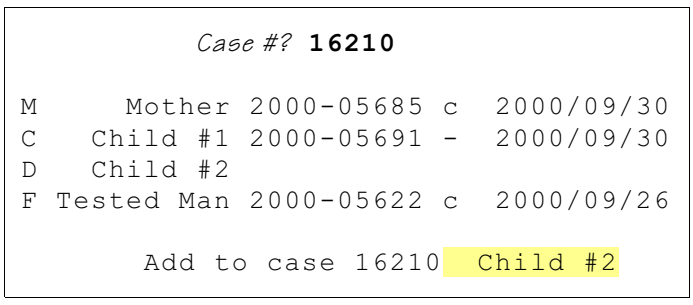



**Figure 7** adding a second child

**V.B.3.** *add role*

A paternity case can have up to ten children, up to three possible fathers, and/or zero to three mothers. Other cases (crime, automatic kinship) have slightly different limitations, shown in **Figure 154** in Chapter XVI, page 311. To make a case with extra people, first select or create the case in the usual way, then select *add role*. You will be presented with a

menu from which you can choose an (additional) *Mother*, *Child*, or *Man* (or whatever roles are appropriate to the type of case – **Figure 154**) to add to the case.

DNA·VIEW then presents the case definition block with the cursor in position to define the new person.

To delete a role from a case, set the year part of the accession number to **0** or use ***Delete*** to exit from the year field.

The report from *Paternity Case*, *calculate paternity/maternity* will include a computation for every possible selection of a mother (if any), a child, and a man.

### V.B.4. *batch run, batch select*

The *batch* facility is to let you initiate computation of several cases, then walk away from the machine and let it finish.

First select *batch select*. You then create a list of case numbers to run, in any of several ways according to the choices offered by a popup menu (**Figure 8**).

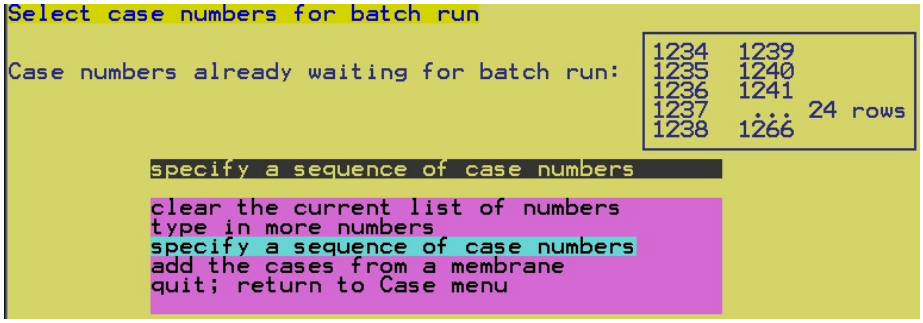



**Figure 8** *batch select* options

V.B.4.a. *clear the current list of numbers* to start over defining a batch of case numbers.

V.B.4.b. *type in more numbers* – (outmoded method) You type the (additional) case numbers to be run into a box. ***Esc*** when done. (However, if the previously entered case numbers use up too much room, the "special editing screen", §XIV.B, is used in which case type ***Ctrl-e*** when finished.)

V.B.4.c. *specify a sequence of case numbers* (recommended). Respond to the prompts to give a starting case number and the number of consecutive numbers. It won't be a problem if some of the numbers in the sequence don't exist; they will be skipped over fairly quickly (but not instantly – don't specify an interval that includes a million non-existent cases!).

V.B.4.d. *add the cases from a worklist* (recommended) – especially convenient if you have imported one or several worklists with cases to analyze.

Then invoke *batch run*. The cases will be run sequentially. After each case is run its number is removed from the batch list, so if you happen to interrupt the process and restart it later every case will be run. In fact, the list of (remaining) cases to analyze persists even between DNA·VIEW sessions.

**V.B.5.** *calculate avuncular*

( – 2014)

*Calculate avuncular* considers the hypothesis that the tested man (or woman if maternity) is alleged to be the *sibling* of the alleged parent, i.e the uncle or aunt of the child, versus the alternative hypothesis of being unrelated to the child.

V.B.5.a. Generalized avuncular calculation

More generally, the program allows the user to specify an *avuncular coefficient*. The choice *avuncular coefficient*=½ gives the avuncular calculation described above testing whether the tested (wo)man is an uncle (aunt). Choosing *avuncular coefficient*>½ tests whether the tested person is some more distant unilineal relative of the father (mother), e.g. 3/4 corresponds to an alleged cousin of the child.

V.B.5.b. θ>0

Setting θ>0 modifies the alternative ("unrelated") hypothesis in the same way as for *calculate paternity/maternity*. See §V.B.19.

**V.B.6.** *calculate trio/duo*

(new – 2014)

This option allows a parentage computation designating any desired role letters for parents and child, not restricted to M, C, F, etc as in §V.B.7.b. All variations of "parentage" are allowed including (generalized) avuncular, maternity, duo, prior, and theta. The calculation uses the best mutation model and therefore is probably preferable to the kinship program (*immigration/kinship*, §VI.F) when there is a choice. Results can be exported ready-for-Excel per *Case options* §V.C.5.c.

**Operation**: Open the case ▸ MENUS: CALCULATE ▸ *calculate trio/duo*

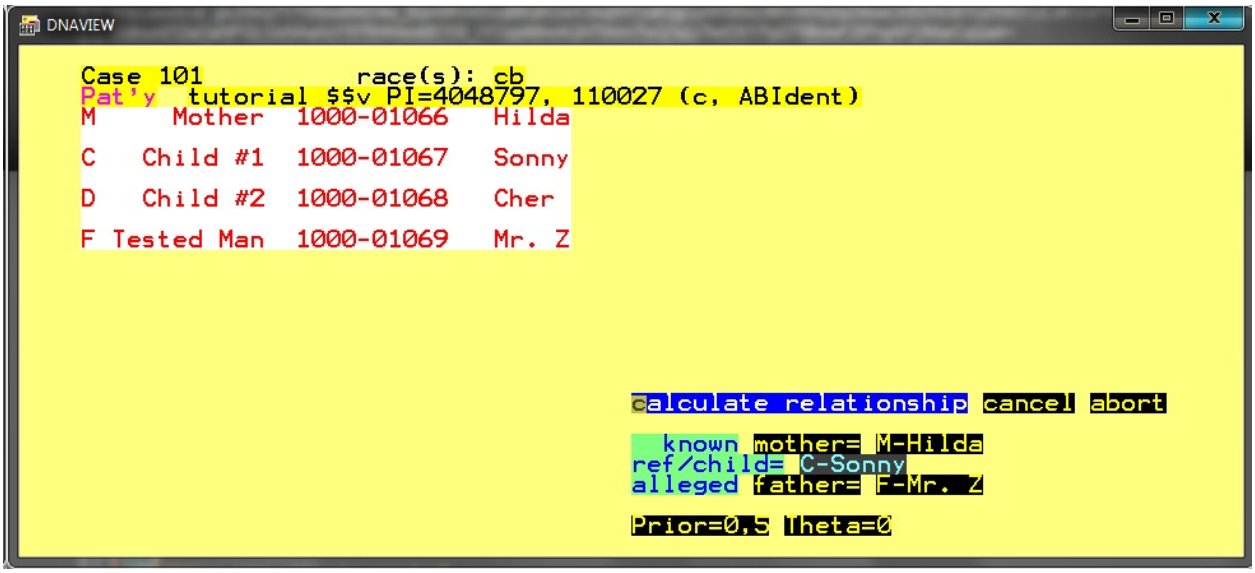



**Figure 9** *Calculate trio/duo* panel

Some examples using the tutorial case 101 will illustrate the use.

V.B.6.a. Clicking CALCULATE RELATIONSHIP from the initial button status shown does the avuncular computation for child C, assuming mother M, and testing whether F is uncle vs. unrelated.

V.B.6.b. Question: Assuming father unknown, is M the mother of D, vs. M unrelated to D?

Solution: Click MOTHER= and from the popup menu select *language: maternity*. (The case switches to **Mat'y** and the button changes to FATHER=. Also, the last button changes to F-HILDA.)

Click the button after FATHER= and from the popup click (NONE).

Click REF/CHILD=... and select *D-Cher*

Click ALLEGED UNCLE/AUNT/GPARENT= and choose *mother*.

Leave the last button as F-HILDA.

***Enter*** to CALCULATE RELATIONSHIP.

V.B.6.c. Question: Can C be the father of D? Assume prior probability=2% and compare with the alternative of a random father slightly (θ=0.01) related to D.

Solution: Restore (if necessary) KNOWN FATHER= to MOTHER=. Set the next button to (NONE).

After REF/CHILD=, put D-CHER.

After ALLEGED, put FATHER=, then C=SONNY.

Click PRIOR and set to 0.02. Click THETA and set to 0.01.

***Enter*** to CALCULATE RELATIONSHIP.

**V.B.7.** *calculate paternity/maternity*

This options calculates the paternity index computation and creates reports for it. The computation includes

```
Case 16080
Data for case 16080  1998/5/31 13:38
          M     Mother  91-05307 b  95/09/26
          C      Child  91-05309 -  91/09/10
          F Tested Man  91-05306 b  91/09/10
Computation per race(s): hbac
```

```
     Paternity Index Summary for case 16080
D3S1358 3p       PCR X/Y = 0.5  / 0.392  = 1.27 (h)
FGA     4q       PCR X/Y = 0.25 / 0.155  = 1.62 (c)
D8S1179          PCR X/Y = 0.25 / 0.195  = 1.28 (h)
D21S11           PCR X/Y = 0.25 / 0.0536 = 4.66 (c)
D5S818           PCR X/Y = 0.5  / 0.394  = 1.27 (c)
D13S317          PCR X/Y = 0.5  / 0.0892 = 5.61 (h)
D7S820  7q11     PCR X/Y = 0.25 / 0.111  = 2.26 (h)
TH01    11p15.5  PCR X/Y = 0.5  / 0.258  = 1.94 (h)
CSF1PO  5q33-34  PCR X/Y = 0.5  / 0.149  = 3.36 (h)
                              combined PI = 1290
```

```
     A (exclusion probability) =  99.87%
  W (at 50% prior probability) =  99.923%
```

```
Analysis for D3S1358 3p PCR
 14                  Mother
 14  15               Child
     15  14      Tested Man

***  D3S1358 3p PCR probabilities from 412 observations
   Promega Hispanic

PI = 1.27±6%    X/Y=0.5/0.392
     P{14} = 0.0847 and P{15} = 0.392.  RMNE = 0.631
(PI=1.76 ABI Caucasian)
```

```
Analysis for FGA 4q PCR
 23  22              Mother
 23  22               Child
     22  24      Tested Man    etc.
```

**Figure 10** *Case* report

all loci and caters to a number of variations and elaborations that might arise. In particular:

V.B.7.a. variations on the paternity case

- » Depending on the setting of the "*Language is*" parameter (§V.B.15) – *paternity* or *maternity*, either the father or the mother may be unknown.
- » two party cases – The mother (father if maternity case) need not be tested, or may have missing types in some loci.
- » multiple children – If several children are included in the case (role letter C for the first one, then D, then E, back to A (because F means tested man/woman), etc, maximum of 10), then each one

is computed as a separate trio. If you would like to make a computation that takes the several children into account simultaneously, use *Kinship*, Chapter VI).

» multiple tested men/women – Similarly, more than one suspected father (or suspected mother if maternity, or for that matter, though it is not very likely, more than one presumed mother/father) may be included in the case – using role letters F for the first one, then G, then H – and every different man-woman-child combination is computed as a separate trio.

V.B.7.a.i.) **Mutation possibilities** (§XIII.C) are computed automatically (§V.C.3). A mutation calculation is indicated by a code on the *Case* report (**Figure 10**). The symbol μ (*mu*tation) indicates that a mutation calculation has rescued a nominally inconsistent pattern such as child=18, tested man=15 19 from what would otherwise be PI=0:

```
VWA  X/Y = 1/2350 / 0.113  = 1/266   (c) μ
```

A * symbol means that the pattern is consistent, but the parameter of §V.C.3.f is set so small that a mutation explanation is also included for accuracy. For example if the paternal allele is rare the random man computation might take into account not only a random sperm with the rare allele but also a random sperm mutated from a neighboring common allele.

» Null allele (or inheritable primer hybridization failure) possibilities are computed automatically (§XIII.B.5).

V.B.7.b. Role letters and paternity cases

For the purpose of this option only, the role letters – M, C, F, etc – have special computational significance. In the simple trio paternity case, the M is the mother, F the *alleged* father, and C the child.

» For maternity cases or those with extra people, please look at the role letter chart, **Figure 154**, page 311.

» For **maternity** cases specifically be sure to note that **M means father!** (§XVI.A.1.a)

V.B.7.c. multiple or various race computation (§V.C.6.a)

The menu option *calculate paternity/maternity* collects all the available measurements relevant to the case. The data is condensed into one size measurement per individual per locus by the averaging algorithm described in §XIII.F.

Analysis then proceeds

V.B.7.d. trio-by-trio. If there are extra men and/or children then each combination of man and child is treated as an independent case. For example a case with two men and three children is calculated as 6 trio cases.

It may be that there is no maternal data; the man-child duo is then considered as a "trio."

For each trio, the analysis is:

V.B.7.e. locus-by-locus. For each locus, the program decides on the pattern of possibly matching alleles among the parties of the trio, then makes a PI calculation.

V.B.7.e.i.) Matching patterns are shown in boxes for the example case of **Figure 10** The allele measurements (averaged over multiple assays in the case of RFLP) are presented in staggered rows, as shown in the boxes in **Figure 10**. The rows labeled *mother*, *child*, *tested man*, and *man & child*, and show the allele sizes of each person or combination. The rows are staggered to indicate the apparent sharing of alleles through inheritance.

V.B.7.e.ii.) **Race-by-race**

V.B.7.e.iii.) **Choice of race**

If there is more than one race in the race list for the case, then calculations for all of them are shown in the per-locus computation block.

The summary report for the trio is either based uniformly on the race that gives the overall minimum PI, or, depending on the setting of the "ultra-conservative" *Case Option* setting §V.C.6.a, the race for which the PI is smallest may be chosen locus-by-locus.

V.B.7.e.iv.) **Choice of database**

The database employed for each calculation is determined by linking the race code to the "default for" designation of the database list. For example if the race code is **c** and the locus is VWA, if there is a VWA database which has the race code **c** included among its "default for" letters, then that will the chosen database for computing a **c** probability.

V.B.7.e.v.) **database on-the-fly**

If there is no appropriate default database designated for a certain race/locus combination, the program will pause and query the user to select a database to be used. When that happens you may either

1. *supply no database* by hitting ***enter***, in which case no computation is made; or
2. *choose a database*, in which case the program opens the dialogue box for defining default databases (see §VIII.C.2.b), and you can if you like take the opportunity to designate the chosen database to be a default choice for future casework.

   In order to avoid the need for the user to repeat the same database selection task for every locus, the program searches for similarly named databases for other loci and lists them on the screen along with instructions. That screen lets you make a consistent choice of default database for all or some of the other loci (§VIII.C.2.b.ii).

The parameters — δ, σ and (for the Gjertson case) $\sigma_g$ — in effect for the calculations are established via *window size* (§VII.B.2) menu option.

### V.B.8. *calculate family trio*

This is a custom calculation designed for the special case of animals such as horses where both maternity and paternity might be in doubt. Both the trio index (paternity) and duo maternity index are calculated, then they are combined assuming a prior probability of 50% to obtain a posterior probability that both relationships are correct..

### V.B.9. *compute one locus* (paternity analysis detail)

This *Paternity Case* option allows convenient analysis of a specified pattern. It uses data from the presently selected case as a starting point, but permits arbitrary modification.

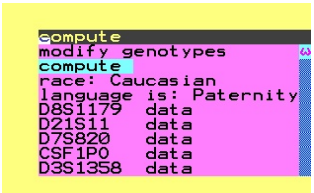


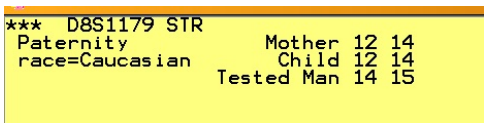

**Figure 11** *compute one locus*

V.B.9.a. Typical operation

V.B.9.a.i.) Select a locus – e.g. `D8S1179 data` from the menu

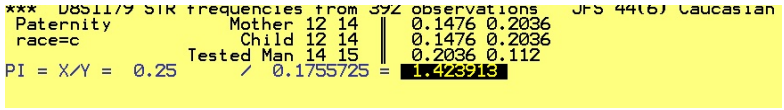
V.B.9.a.ii.) select `compute`

V.B.9.a.iii.) select `modify genotypes` from the menu and type in different genotypes; `compute`. Try a different `race` if you like.



With this tool you can examine step-by-step the effect on the calculation of examples involving possible mutation and/or null alleles.

V.B.9.a.iv.) Switching between `language is: Paternity` *or* `Maternity` makes a difference when the analysis involves possible mutation.




V.B.9.a.v.) The `locus:` menu choice lets you select and enter data for an arbitrary locus, including one for which the case has no actual data.

**V.B.10.** *delete case*

deletes all the table references in DNA·VIEW to the currently selected case. The people in the case are not deleted; they are still defined, with associated race codes for example, and they still occupy their places on worklist rosters.

If the case also exists in PATER, the PATER parts are not deleted. In particular any serological data that PATER holds for the case will remain.

**V.B.11.** *edit case comment*

The option *edit case comment* in the *Case* menu lets you add a description or comment to a case (such as **`Hawthorne`** in **Figure 5**). The comment is free-form and up 60 characters long. The comment can include a name such as the name of a victim, family name, or client associated with the case, and personal codes for classifying the case. The comment is useful because it

V.B.11.a. appears on the screen when you retrieve (*select case*, §V.B.28) a case;

V.B.11.b. can be searched against to find a specific case or a category of cases; see §V.B.28.a.ii;

V.B.11.c. is printed on the log version (§VI.F.2.a) of an *immigration/kinship* report.

**V.B.12.** *edit people*

Choose *edit people* to change or fill in the accession codes, races (the race of the child must be –), and dates for the people in a case.

There are few special facilities, mentioned on the screen, that can help you to edit faster:

V.B.12.a. ***Shift Tab*** backs up to the previous person. This action is cyclic, the last person preceding the first.

V.B.12.b. ***End*** to end editing.

**V.B.13.** *edit race list*

V.B.13.a. paternity

The race list tells which race or races are to be used for computation – that is, for allele probabilities.

In the case of paternity this means that the race list is the list of race code letters for "random men" who are to be considered as potential alternate fathers in computing the paternity index. Traditionally the race of the alleged father was used but that is not really logical. DNA·VIEW takes the specified race of the

alleged father to seed the race list, i.e. as a default for the computation race, but since the user can edit the race list there are conceptually two different roles that race can play in a case:

» The race list specifies the "computation race" or races.

» The race of the alleged father is merely his "databasing race" – in case his DNA is eventually included in allele frequency databases.

By using *edit race list*, that letter may be changed to another letter, or to a list of several letters. Note that once this is done, the race of the alleged father no longer plays any role in the paternity index computation.

When there are several race list letters, the *calculate paternity/maternity* option will attempt to make a computation for each letter for each probe/locus. The smallest PI thus obtained, locus by locus, is the one used for the PI summary and the cumulative PI in the case.

V.B.13.b. kinship

The first race on the list is the one initially selected for kinship (*immigration/kinship*, Chapter VI) computations and simulations.

The race list is the list of alternate computations that are made by the *Calculate & report LRs, [all] races* (§VI.F.2) option and the list of races included in the *Next race from:* (§VI.F.7.h) option.

### **V.B.14.** *export case*

This option creates a text file that contains the essential data of the case – names, roles, and genotypes. The text file format is like the ABI Genotyper "allele table," so that it can be imported into DNA·VIEW using the *Import/export* ▸ *Genotyper import* option (§X.A.1).

This is therefore a convenient way to communicate a case to another person, especially another user of DNA·VIEW, who can simply use *Import* the file and recreate the same case. The people are designated in the "Sample Info" column using the role-within-case format (see **Figure 123**, page236) By default the exported case number is the original case number, but you are given an opportunity to specify a different case number instead.

V.B.14.a. Creating a case from a simulation

The *Kinship simulation* (§VI.G) menu includes an option to export in the same way. Therefore it is possible to generate simulated profiles according to a specified relationship scenario, export the simulated types, then import them in order to create a practice case.

### **V.B.15.** *immigration/kinship*

**Use:** Relationship calculations for any non-standard situation.

This sub-option of *Automatic Kinship/Paternity /Automatic mixture* is a link to **Kinship** (Chapter VI). Immigration cases is just one application — it can equally be used for any kinship problem including paternity, inheritance, missing person, etc.

For instance, you can type the kinship scenario definition

```
C : M + F/?
```

to *immigration/kinship* and thereby obtain approximately the same result as the normal trio analysis via *calculate paternity/maternity* §V.B.7). However, you can also enter any other kinship formula, and thereby quickly and automatically investigate other kinds of relationship.

*Immigration* is an involved and important option; see Chapter VI, Kinship, for full documentation.

**V.B.16.** *language is [paternity | maternity | crime | kinship]*

This option tells you, and instructs DNA·VIEW, whether the present case uses the language of paternity ("Mother", "Tested Man"), or a different language from among those listed in **Figure 154**.

V.B.16.a. Changing the "language" between maternity and paternity exchanges the roles of the male and female parent (if any) or alleged parent, as most clearly shown by examples:

| example & comment | before | | after | |
|---|---|---|---|---|
| trio paternity → maternity<br>The role letters for Mary and Frank are changed, because for DNA·VIEW "M" designates *known parent*, not *female parent*, etc. | pat'y<br>M Mother<br>C Child<br>F Tested man | <br>Mary<br>Charles<br>Frank | mat'y<br>M Father<br>C Child<br>F Tstd Woman | <br>Frank<br>Charles<br>Mary |
| duo maternity → paternity<br>The paternity case lacks a Tested Man so cannot be calculated. | mat'y<br>C Child<br>F Tstd Woman | <br>Charles<br>Mary | pat'y<br>M Mother<br>C Child | <br>Mary<br>Charles |
| Multiple men, paternity→maternity | pat'y<br>M Mother<br>C Child<br>F Man #1<br>G Man #2 | <br>Mary<br>Charles<br>Frank<br>George | mat'y<br>M Father #1<br>N Father #2<br>C Child<br>F Tstd Woman | <br>Frank<br>George<br>Charles<br>Mary |

V.B.16.b. Changing the "language" between any other pair of the possibilities (crime, kinship, paternity, maternity), *only* changes the words ("Mother", "Suspect" etc. per the role letter table in §XVI.A) that are sometimes (§V.C.5.a) attached to the role letters. Each person retains the same role letter.

» Note that therefore if you do two switches paternity→kinship→maternity, the role letters are *not* swapped. This might be a useful trick if you accidentally imported an intended maternity case as paternity.

V.B.16.c. To **create a maternity case** you may:

V.B.16.c.i.) Import case data defined as a maternity case.

V.B.16.c.ii.) Define the case initially as a paternity case, open the case, then switch *language is:* ▸ *maternity*.

**V.B.17.** *list cases*

The *list cases* option in the *Case* menu gives a summary of cases in a specified range, or that include a specific person/sample number.

Data shown includes the case number, the type of case, (the race list), the comment, people in the case, the number of loci available, and the number (if more than 1) of worklists with relevant data.

Select the *Case* option *list cases* to find a previously entered case.

Now choose letter **C** or **P** for Case or Person.

```
List by Case or by Person (C or P)? C
          List people between 1217  and 1230

     Cases from 1217 to 1230
1217 pat (abc) FMB-xx
 M 1000-10075 x 10 loc    Mommie
 F 1000-10076 - 10 loc    Alleged Dad
 C 1000-10077 - 10 loc    Caleb
1218 kin (c) Body id (Bro & Uncle)
 A 1000-10020   13 loc(5) son of Sylvie
 B 1000-10021   13 loc(5) brother of Sylvie
 Y 1000-10022   13 loc(5)
```

**Figure 14** Case list by case

The **C** report is a row-per-case summary (case #, accession #'s & races) for a specified range of case numbers. See **Figure 13**.

The **P** report is a list of people (accession #, race, accession date) within a specified accession number range, followed by a C report listing every case that contains any of the listed people.

**V.B.18.** *Let PRIOR=* **(0≤p≤1)**

(new – 2011)

Instead of the traditional prior probability of ½ for paternity, you may choose a different value, or *none*.

**V.B.19.** *Let THETA=* **(0≤θ<1)**

(new – 2011)

Setting θ>0 (*experimental feature*) modifies the denominator of the traditional *Calculate paternity/maternity/avuncular* likelihood ratio to consider that the alternative parent (or uncle) is related to degree θ.

**V.B.20.** *locus parameters*

Same as §IX.B.3.

**V.B.21.** *Name tag style: NAME/DATE*

Select this option to toggle between two styles of displaying the name tag block (**Figure 14**). Recommended style is "name", since the date not very useful, and by displaying the names (which are also the names used by PATER) you can also edit them, greatly enhancing the documentation for the case.

V.B.21.a. Editing dates

However, if you do want to change the draw date (because you want the correct date to appear on the PATER report), then you must – at least temporarily – toggle to *Name tag style: DATE*. Then you can edit the dates.

**V.B.22.** *Null allele frequency: AUTO/ASK*

This option affects the way that the probability ***o*** of occurrence of a null allele (see §XIII.B.5) is determined during a paternity trio calculation (*Paternity case*, *calculate paternity/maternity*). Select to toggle between the *AUTO* and *ASK* treatment. *ASK* means that the program pauses to allow the user to modify the nominal value obtained from the relevant database. *AUTO* means that the program should just use the value from the database without pausing.

**V.B.23.** *(case) options*

There are a dozen or so options that govern some details of case computation. Since the description is several pages, and some of the options have relevance beyond *Paternity Case*, they are explained in a separate section, §V.C, page 58.

**V.B.24.** *recap*

After computing each case, DNA·VIEW posts a one line summary to a special "recapitulation" list. The *recap* option lets you examine and maintain that list, illustrated in **Figure 14**.

From the *recap* menu, you can immediately:

| | |
|---|---|
| *clear* | throw away all the information in the recap file |
| *create & display report* | show the recap report on the screen. You can examine it by scrolling. Exit *Case* then *File* to print, save, etc. |
| *1-letter codes* | define the codes (e.g. **y** above) used to designate each probe. |
| *back to case* | |

```
 Case       When             Tests performed     PI (among
            calculated       incls   excls       inclusions)
11663-MCF   1993/8/5 10:04   te                  116
16001- CF   1993/8/5 10:05   ye      t            30.2
```

The coding `CF` indicates a motherless case. A second child or man would be coded `MDF` or `MCG`, etc. In case 16001, the man was included with loci designated **y** and **e**, but excluded in **t**. Considering only the inclusions the PI was 30.2.

**Figure 15** The Recapitulation report

**V.B.25. Reorder loci**

Same as §IX.B.1.d.

**V.B.26.** *restrict data*

is used to specify a temporary subset set of the available genotype data for subsequent calculations (*calculate paternity/maternity* (in *Paternity Case*), *immigration/kinship* calculations, or *mixture calculation* (in *Automatic mixture*)). In particular, you can choose which people and/or which loci to include or to exclude from the computation. If there are multiple assays for a locus-specimen, you can choose only one of them.[1]

After the restricted selection is complete, the program proceeds with the subsequent calculation.

V.B.26.a. *restrict data* selection process

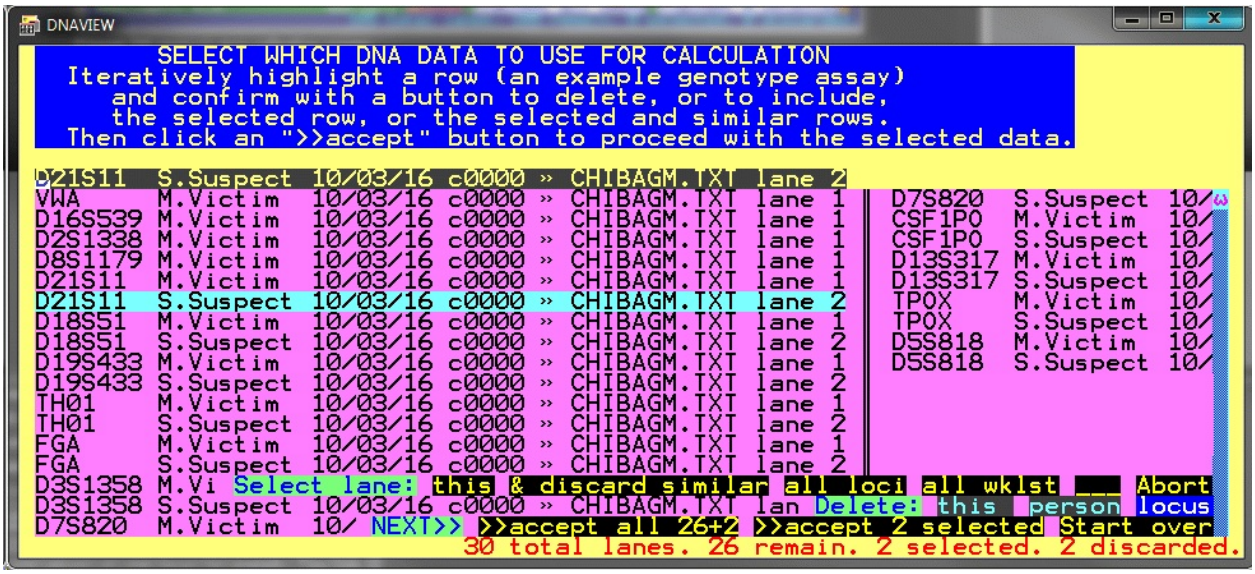



**Figure 16** *restrict data* selection screen

All available data is presented in a menu (**Figure 16**). Each line represents the data for one assay of one person in one locus. By a successive menu selections and using various buttons (or corresponding short-cut keys) representing a variety of selection rules, you can choose any desired subset of the data.

Selection is by including desired data (SELECT LANE: row of buttons) or by removing undesired data (DELETE: row of buttons). The bottom button and screen lines summarize selection progress.

The selection process can be either negative (delete undesired data) or positive (select desired data).

V.B.26.a.i.) Three buttons govern over-all rules:

» START OVER – Forget all selections and start selecting over again. This is to recover from a mistake.

» >>ACCEPT ALL r+s – Finished selecting. Accept all lines except those specifically rejected (methods of §V.B.26.a.ii). This approach is the easiest way to eliminate one person or one locus for example. (r+s refers to r remaining genotypes neither selected nor discarded plus s selected genotypes.)

» >>ACCEPT s SELECTED – Finished selecting. Accept those lines specifically nominated for inclusion (methods of §V.B.26.a.iii). This approach is convenient in order to use only one of multiple assays, or to analyze only one locus and a few people.

[1] Recall, the default DNA·VIEW procedure (§XIII.F) would be an average or consensus of both.

The remaining buttons apply after first moving the bounce-bar highlight to a particular menu item. They are logically divided into two categories:

V.B.26.a.ii.) Rejection buttons, preliminary to >>ACCEPT ALL . . .

» DELETE: THIS – omit this assay of a person/locus.

» DELETE: PERSON – omit this person (i.e. this line and also other lines with the same role letter).

» DELETE: LOCUS – omit this locus (i.e. this line and also other lines with the same locus).

V.B.26.a.iii.) Selecting buttons, preliminary to >>ACCEPT . . SELECTED

» SELECT LANE: THIS – include this person-locus genotype

» SELECT LANE: THIS & DISCARD SIMILAR – include only the highlighted assay of a person/locus (removing any other assays for the same person/locus).

» SELECT LANE: ALL LOCI – include this line and also all other loci for the same person/lane/roster.

» SELECT LANE: ALL WKLST – include this line and also all lines (genotypes) from the same worklist so long as the genotype occurs just once. (For example do not include a person-locus combination which occurs in two different lanes).

### **V.B.27.** *review/print report*

Open the *Case* report long version (In addition to the key information that is shown in **Figure 10**, it also contains information about inconsistent readings and ignored data.) in a print-preview-like window (as §V.K.3) for printing or saving.

### **V.B.28.** *select case*


Choose *select case* to change the current default case number, or to initiate a new case. The case number selection dialogue may also occur in other contexts (e.g. §V.J.3.d).

You may simply type in a number. Or, there are various tricks (§V.B.28.a) to find a case or help type. If the case number is already on file, identification data is presented for mother, child, and tested man as shown in **Figure 5**, page 41.

If the case number is not already on file, an empty case matrix is presented on the screen and you will be asked to enter (per §V.B.12) the people of the new case.

V.B.28.a. Selecting tricks (revised – 2011)

A number of shortcuts and other aids are available to make it easier to find the right case number.

V.B.28.a.i.) RECENT for recently visited or popular cases.

This key pops up case-selection-menu of recently-used or often-used case numbers.

The parenthetical numbers following the case number are the number of times that the case has been selected, among the most recent 500 selections.

» The *Recent* box includes WORKLIST button, which allows selection of a worklist whose cases are then added to the *recent* list.

Notes:

» The cases from a just-used worklist will already be included in the list per §V.B.28.a.i. In particular, if you have just done a *Genotyper Import* for example, then the new cases are automatically in the recent or popular case list.

» Dashes instead of a parenthetical number after the case means zero previous selections, and the case is from the selected worklist.

» Such dash-denoted cases are put at the top of the list. This makes it convenient to successively analyze all the cases of a worklist. You can simply pick the top one, analyze it, then select the new top one, and continue in this manner until there are no more dashes.

V.B.28.a.ii.) SEARCH – and optionally open a case – by case number or case comments (§V.B.11) which are displayed in a popup window. This facility becomes a powerful open-ended feature by judicious use of case comments thanks to the *context searching* (§XIV.A.2.a.iii) property of DNA·VIEW menus.

If the comment contains the name, typing the name at this menu automatically finds the corresponding case.

Further it is convenient to search for all the cases with a particular code or set of codes embedded in the comment. Recommended and suggested codes include

| | |
|---|---|
| `>>` | *Screen disaster matches* can recognize this as a completed or otherwise presently inactive case and sort it to the end. |
| `$v` | Case used for validation |
| `$r` or `>>$r` | Case currently under review |
| `{C:M+Fa/?{Fa,M: ?+?}` | kinship scenario for the case (example) |

V.B.28.a.iii.) prefixing a year number. Type the sequence part of the case such as **987** then click one of the YEAR: buttons to prefix digits representing THIS, LAST, or NEXT year. The number of year digits, and whether the sequence part is padded with leading 0s, depends on the *Case # format* (§IX.F.2).

V.B.28.a.iv.) NEXT AVAILABLE NUMBER – First type a positive integer (a possible case number), then click to obtain an unused case number.

**V.B.29.** *simulation*

Same as §VI.G.

**V.B.30.** *parentage test examples*

This is a validation option to run a variety of paternity calculations, comparing the results against canned results. The main purpose is to test DNA·VIEW versions before release.

## V.C. (*Case*) *options*

### V.C.1. Where *Case Options* are

The [*Paternity/Crime/Automatic Kinship*] *Case* option *options* gives you some choices about details of the case reporting. Your selections will remain effective until you change them.

(*Case Options* are not to be confused with the DNA·VIEW *Options* – §IX.F – command in the **Housekeeping** menu. However, the *... Case Options* are also available as a sub-menu item under *Options*.)

### V.C.2. Changing *Case Options*

The items listed below are presented in a selection menu. The procedure to change is as follows:

i) Select the item to change.

ii) If it is a numeric parameter, fill in the desired new value. If a yes/no parameter, this step is skipped.

iii) *Ok to change?* Answer **y**.

### V.C.3. Mutation *Case Options*

The first five options below (*Use the model, Ratio, Ratio, Fraction, Lower bound*) comprise the *STR Mutation model*, discussed in more detail at §XIII.C.3.



**Figure 18** Index of *Case Options*

V.C.3.a. `Use the model below for mutation in STR systems?` **y**

See §XIII.C page 281 for a discussion of mutation in paternity casework. The next four options are relevant only if this one is set to "yes" (recommended).

V.C.3.b. `Ratio of 1-step to 2-step mutations? (probably 10-50)` **10**

V.C.3.c. `Ratio of paternal to maternal mutation rate (about 4)` **3.5**

V.C.3.d. `Fraction of mutations that increase allele size (0.5-0.6)` **0.5**

V.C.3.e. `Lower bound, probability of any mutation (usu 0, or e.g. 0.0001)` **0**

V.C.3.f. `Include mutation possibilities if effect > x% (e.g. 1-30%)` **10**

Lets the user control to what extent mutation possibilities are considered. Consider an example such as

| | | | |
|---|---|---|---|
| Mother | 12 | 14 | |
| Child | 12 | | 15 |
| Man | 12 | | 16. |

Obviously we must consider the possibility of a 16➛15 (paternal) mutation as part of the computation of X=Pr(given DNA types|paternity). Another possible explanation, not far-fetched, would be a maternal

14➛15 mutation with the man contributing the 12. A computation of X should probably include both of these possibilities.

There are further theoretically possible explanations as well, such as with two mutations and mutation of 12➛15. Including all of these in the computation would introduce complexity – difficulty in verifying the computation – with no worthwhile gain in accuracy. It seems better to leave trivial terms off.

The mechanism implemented is to give the user the choice of omitting any terms that would affect the answer by less than a user-chosen percentage.

V.C.3.g. `Default STR mutation rate (if no locus-specific) (%; recommend 0.28%).` (new – 2014) The typical mutation rate for a polymorphic forensic STR locus is 1/350 or 0.28% hence the recommendation which is used for any STR locus whose locus-specific rate (§IX.B.3) is set to 0. (**Note –** if this rate is 0 the non-STR value is substituted!)

V.C.3.h. `Default non-STR mutation rate, e.g. SNP (logarithm, i.e. -8 for 10^-8).` (new – 2014) A mutation rate often cited for single nucleotides is $10^{-8}$, hence the recommendation.

V.C.3.i. `Minimum PI - report PI as 0 if less than (0-1, reasonably 0.01)`

See §XIII.C.1.c, page 281.

V.C.3.j. `Max # inconsistencies for a parent (rec: 999; prefer "Min PI")` **`999`**

See §XIII.C.1.c, page 281.

V.C.3.k. `Use motherless calculation for loci with maternal exclusion? (Recommend: no)` **`n`**

Using STR systems and the STR mutation model (§V.C.3.a), **`no`** results in an excellent computation properly taking into account the possibility of a maternal mutation. **`Yes`** means, if the mother is excluded, ignore her and make the motherless calculation as if she were not typed in this locus.

### V.C.4. Allele probability *Case Options*

(revised – 2010)

There are various situations requiring an evidential matching probability, namely

*the probability than a randomly selected allele (or haplotype) would match a given observed type.*

The options in this section allows the user to select among various methods for making such a calculation.

V.C.4.a. *Count observed allele in database how many times (recommend: 1)?* **`1`**

This option offers a coherent approach to the vexing problem of matching probabilities for rare alleles, such as alleles that occur in a case but don't exist in the database.

The recommendation is: any allele probability is calculated by adding the critical allele *one extra time* to the database. If the present allele was observed $k$ times in a database of $N$ alleles, the probability is $(k+1)/(N+1)$. In particular for a never-before observed allele, the matching probability is $1/(N+1)$. See §XIII.A.1.e for further discussion.

Why toss the new allele into the database? That's easy — because we have seen it. The database should be a representative sample; to ignore this most important observation would be to create bias. See http://dna-view.com/AlleleProbability.htm for further discussion of this recommendation.

By setting the parameter (call it $t$) in this option to various values, the user can achieve various policies:

$t=0$ to retain the old (and industry standard) behavior, probability=$k/N$.

Also there may be circumstances where the suspect may be presumed already to be counted in the database — a convicted offender database, for example.

***t*=1** recommended choice, following the above argument, probability=($k$+1)/($N$+1).

***t*=2** or more to give probabilities that are (conservatively) large.

***Note a:*** This facility is therefore a clear improvement on and a preferred substitute for option §V.C.4.b.

***Note b:*** See also the option §V.C.4.e below.

V.C.4.b. *Minimum # of matching alleles to assume for probability calculations* **1**

This indirectly puts a cap (maximum value) on PI's and other likelihood ratio computations. The default and smallest allowable value is 1, so that PI is never infinite. Some statisticians like to use a larger value. For example, a value of 3 guarantees than an allele probability will never be reported by DNA·VIEW as rarer than 3/N (N=number of alleles in the database), which is 95% confident to be a conservative estimate for the true frequency of an allele that was never before observed. (The astute reader will notice a disconnect in confusing frequency and probability.)

The actual limit on PI implied by this parameter is proportional to the database size, and also depends on the matching pattern (whether the man has 2 eligible alleles or one) in a given case.

V.C.4.c. *Minimum frequency (recommend 0. prefer minimum count)*

If you insist, you may set a minimum value for allele probabilities. Entering a probability $f$>1 is interpreted as 1/$f$. That is, for a minimum probability of 1%, either enter **0.01**, or enter **100**.

If the motive for setting a floor is to take into account the effect of sampling variation, then the appropriate frequency to choose should depend on sample size. In fact, it is very nearly proportional to sample size, so a more sensible floor for frequencies is obtained by using option §V.C.4.b.

V.C.4.d. *Y-haplotype LR model (recommend:* κ *if rare)*

There are three options for computation of Y-haplotype matching probability.

V.C.4.d.i.) *count* – the (augmented) sample frequency per §V.C.4.a is taken as the matching probability.

V.C.4.d.ii.) κ *if rare* (recommended) – For haplotypes never before seen, use the κ rule of Brenner (2010), and see §VIII.F.5. For haplotypes already seen in the database, use the *count* rule.

V.C.4.d.iii.) κ *always* – Use the κ rule per Brenner (2010).

V.C.4.e. *Calculate crime allele probabilities simultaneously?* **n**

This option modifies the "($k$+1)/($N$+1)" allele probability computation behavior discussed above in §V.C.4.a *Count observed allele in database how many times*.

I recommend *not* using this option (answer **n**) because the effect is complicated. It is offered only for compatibility.

It applies only to crime casework — *Mixture Calculator*, *DNA odds*, *DNA exclusion*. For paternity and immigration calculations, the probability of each allele is always computed independently of the others, as described in §V.C.4.a.

If there are several alleles — $s$ alleles — specified at a locus, a "($k$+1)/($N$+$s$)" formula is used, which represents including the alleles as part of the database for purposes of calculating the probability of each one. See §XIII.A.1.e for the precise rules, and §XIII.A.1.e.iii for examples.

### V.C.5. Reports and filing *Case Options*

Both *Paternity calculate paternity/maternity* option and *Immigration/kinship -- Calculate & report LRs,* [n] *races* will (optionally – per certain *Case Options*) produce a file of results in standard format. This report is in addition to the printable "log" report that is displayed on-screen during the computation. The original intention of the file is for automatic further processing by a LIMS system (§X.G), or it can be included as part of a manually generated report.

V.C.5.a. *Omit role descriptions for kinship/crime cases*

Select **y** if you would rather not see the name assignments name assignments associated with the role letters (**Figure 154**) for the people in kinship and crime cases. The reason would be that in some cases those names are misleading – for example, you have put a person into the kinship case with role letter D but the associated word "Daughter" has nothing to do with what the person is actually considered to be. Therefore if the word "Daughter" appears on a report – or perhaps even on the screen – it will be confusing.

V.C.5.b. *Ask confirmation before posting DNA calculations?* **n**

Turn this toggle to **yes** as protection against accidentally re-posting experimental paternity calculations to the DNAAPPRO (="approved DNA results", §XVI.B.3) queue. The purpose of the queue is twofold:

V.C.5.b.i.) to save a record of all paternity computations for later (semi-automatic) analysis (§VIII.K) in order to estimate mutation rates.;

V.C.5.b.ii.) If you are using the PATER program (to produce paternity reports), DNA data from DNA·VIEW is passed to PATER for reporting through this queue.

If the option is set to **yes** then you will be prompted, once (even if there are multiple children) for every case, to confirm that the results are valid and should be posted. In response to the prompt

```
Do you want to post case nnnnn?
```

» type your initials (or anything), then ***enter*** in order to post, or

» ***enter*** in order *not* to post.

If the option is set to **no** then posting will normally happen automatically and invisibly. However, if the case has previously been calculated, then the prompt will occur anyway to give you a chance to avoid re-posting.

Re-posting replaces the original record. It does not create a duplicate record.

V.C.5.c. *Export paternity calculations to a text file?* **y**

This is a way to post the essential computational results from the case calculation to a file that can be imported into a database or word processing program. Thus it can be used as a hook to transfer data from DNA·VIEW into a paternity report formatting program of your own devising. (This file is *in addition* to the computation log that can be printed with *Print*.)

If the export file already exists, you will be given a choice whether to add to it or to replace it:

```
reports\PA101217.txt has 1322 bytes of old data.
Shall I clear the old data before posting? y
```

V.C.5.c.i.) Setting the filename for posting

If you want to specify the filename, toggle the option from **yes** to **no** (if necessary) and then back to **yes**. to obtain the prompt:

`Filename for posting?` **reports\Gotham.txt**

» Note that if the file name begins with **** it will be a "fully-qualified path" and filename.

» If not, then it will be interpreted relative to the directory (folder) in which DNA·VIEW resides.

V.C.5.c.ii.) Numbered files

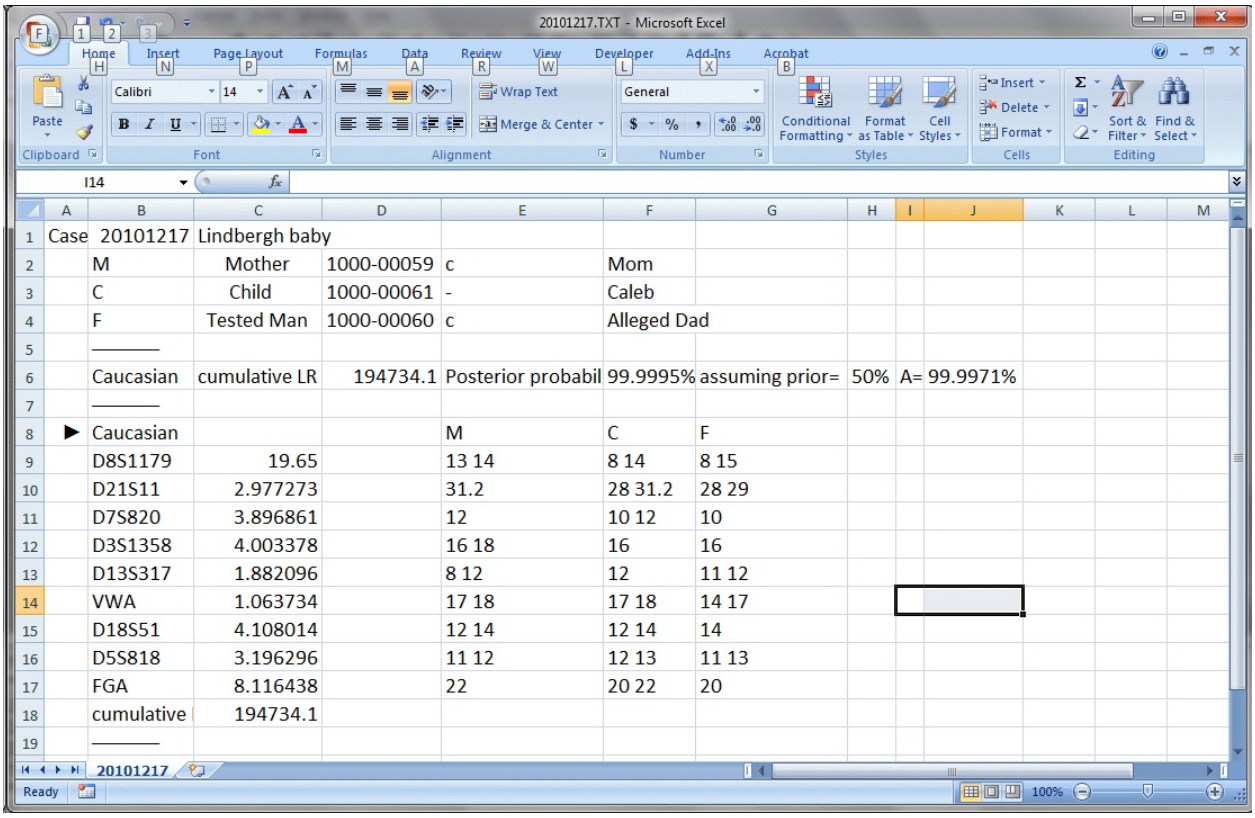
If you choose a filename that has a number as the file name before the extension, or the terminal part thereof

`Filename for posting:` **reports\PA000.txt**

then the actual case number will be substituted for the number each time data is posted.

» However, in order to enforce conformity with the "8.3" file naming convention, if the case number is too long the leading digits will be dropped.

» Example: Assuming the template above suppose the case number is `20101217`. The file name would be **reports\PA101217.txt**.




| | A | B | C | D | E | F | G | H | I | J | K | L | M |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1 | Case | 20101217 | Lindbergh baby | | | | | | | | | | |
| 2 | | M | Mother | 1000-00059 | c | Mom | | | | | | | |
| 3 | | C | Child | 1000-00061 | - | Caleb | | | | | | | |
| 4 | | F | Tested Man | 1000-00060 | c | Alleged Dad | | | | | | | |
| 5 | | ——— | | | | | | | | | | | |
| 6 | | Caucasian | cumulative LR | 194734.1 | Posterior probabil | 99.9995% | assuming prior= | 50% | A= | 99.9971% | | | |
| 7 | | ——— | | | | | | | | | | | |
| 8 | ▶ | Caucasian | | | M | C | F | | | | | | |
| 9 | | D8S1179 | 19.65 | | 13 14 | 8 14 | 8 15 | | | | | | |
| 10 | | D21S11 | 2.977273 | | 31.2 | 28 31.2 | 28 29 | | | | | | |
| 11 | | D7S820 | 3.896861 | | 12 | 10 12 | 10 | | | | | | |
| 12 | | D3S1358 | 4.003378 | | 16 18 | 16 | 16 | | | | | | |
| 13 | | D13S317 | 1.882096 | | 8 12 | 12 | 11 12 | | | | | | |
| 14 | | VWA | 1.063734 | | 17 18 | 17 18 | 14 17 | | | | | | |
| 15 | | D18S51 | 4.108014 | | 12 14 | 12 14 | 14 | | | | | | |
| 16 | | D5S818 | 3.196296 | | 11 12 | 12 13 | 11 13 | | | | | | |
| 17 | | FGA | 8.116438 | | 22 | 20 22 | 20 | | | | | | |
| 18 | | cumulative | 194734.1 | | | | | | | | | | |
| 19 | | ——— | | | | | | | | | | | |

**Figure 19** Formatted paternity report ("standard/LIMS")

V.C.5.d. *If so, use tab-delimited text format ("standard"/LIMS) format?*

V.C.5.d.i.) **yes** (standard/LIMS format) is illustrated in **Figure 19**. Use the DNA·VIEW Report Viewer (§XV.D.2.a.iv) to view the report in Excel.

V.C.5.d.ii.) **no** (machine-readable format) is the same as for Paradox DNAAPPRO table (§XVI.B.3), but comma-delimited text.

V.C.5.e. *If so, post Consensus (rather than exact) sizes?* **y** (RFLP feature; text redacted)

V.C.5.f. *Export (optionally) Kinship all-races computation?* **y**

This option pertains to a standardized Kinship report. It is produced as a side effect of the *Immigration* option *Calculate & report LR's, all races*. The user will *always* be asked to confirm creation of the output file, through being given the opportunity to specify a different file:

```
Filename for posting: reports\case1234.txt
```

The format is illustrated in **Figure 41**, page 113. It is intentionally similar to the standard/LIMS paternity report format above, **Figure 19**.

Establishing the filename follows the same procedure as described §V.C.5.c.i and §V.C.5.c.ii.

V.C.5.g. *Customize "standard" pat'y & kinship files for Excel viewing?* **y**

Answer **y**es if you plan to view the standard reports with Excel (recommendation: use the DNA·VIEW Report Viewer to open the report in Excel). This option will condition the file so that Excel won't do its strange tricks such as treating the fraction `1/2` as January 2 (or 1 February).

DNA·VIEW will include a dynamic posterior probability calculator in the Excel worksheet – permitting you to adjust the prior probability(ies) and see the posterior probability(ies) change accordingly – if you accept the recommended answer of **y**es.

If you will import the standard report into a text editor such as Word, then answer **n**o to simplify the output to plain text.

V.C.5.h. *Make consensus size the same for multiple children, etc (recommend: y)?* **y**

Assuming that you use consensus sizes (in order to report the same size for an allele in the child, as the size of the apparently matching allele in the parent), you might as well let the program try to report a consistent set of sizes across multiple children. It is ultimately an impossible goal, however, and the program's heuristic may not be the best possible, so you should always check its results.

V.C.5.i. *Create "Stat Sheet" of sizes, ranges, PI's?* **n**

The **Stat Sheet** format is a specialized paternity report, summarizing sizes and PI's.

V.C.5.j. *Include QC information on Case report?* **n**

(not yet implemented)

### V.C.6. Calculation *Case Options* (other than allele probability)

V.C.6.a. *Ultra-conservative jumbled race PI computation?*

When the list of "computation races" includes two or more races, this option controls which of two modes of computation are used. In either mode, multiple computations are made for each locus, one for each computation race.

**yes** produces the traditional behavior for *Paternity case*, *calculate paternity/maternity*. Locus by locus, the more conservative result is reported. The cumulative PI therefore reflects the ultra-conservative point of view that the random man may belong to a different race at each locus.

**no** produces a report based on only one race, namely the single race that, overall, gives the smallest cumulative PI.

Description of the following facilities written for RFLP work have been redacted as obsolete. If you are interested consult an earlier manual.

V.C.6.b. *Use NML mm units (and not %) for window, probability calcs?* **n**

V.C.6.c. *Use the Gjertson method as standard?* **n**

V.C.6.d. *Minimum Gjertson PI -- report as 0 when less than 10 to the minus ...?* **5**

V.C.6.e. *Include the Empirical PI computation (can be slow)?* **n**

## V.D. *Automatic mixture* and *Manual mixed stain*

### V.D.1. Purpose

DNA·VIEW offers various methods for mixture computations – the "likelihood ratio"[2] methods which are discussed in this section, and "exclusion" (implemented by *DNA Exclusion*, §V.I.).

### V.D.2. Overview

V.D.2.a. Likelihood ratio mixture programs

The capabilities of the likelihood ratio programs include analysis of mixed stains, multiple contributors, and victim contributions. The *Mixture ("LR") calculator* implements the likelihood ratio methods

V.D.2.a.i.) (binary) as discussed by Weir *et al* (1997). The result is given both numerically and symbolically as an algebraic expression. The *mixture ("LR") calculator* is designed for maximum ease of use and complete flexibility. It is available both

» (manual version) – as a top-level command (**Casework** ▸ *Manual mixed stain*) which operates simply as a high-powered spreadsheet calculator into which the user must type the alleles (although scenarios may be saved and later retrieved and reworked),

» (automatic version) – and also as a sub-command of *Automatic mixture* (**Casework** ▸ *Automatic mixture* ▸ *select case*, then *mixture ("LR") calculator*). This version begins with imported (e.g. *Import/Export* ▸ *Genotyper import*) case profile data including stain mixture and references. The user designates which individual or combination of individuals (roles) is to be regarded as stain, as suspect, and as (optional) victim.

Both versions allow modifying, adding, or deleting data in spreadsheet fashion, and saving your work to retrieve and resume at a later date.

V.D.2.a.ii.) (continuous) with analysis approach similar to Puch-Solis *et al* (2013) incorporating intensity (i.e. rfu) information and awareness of dropout, stutter, and other artifacts. Like the automatic binary version, it uses imported profiles and the user designates the roles. The analysis is then automatic.

V.D.2.b. Theory

The mixture analysis paradigm considers four types of contribution (per Weir et al, 1997):

1 **Mixture** (aka *stain* aka *unknown*) is the set of alleles to be explained – typically a DNA mixture from a crime scene.

2 **Suspect** (reference profile) is a person whose contribution is disputed. According to the first hypothesis (numerator, usually the prosecution) the suspect contributed to the stain; according to the second hypothesis (denominator, i.e. defense), the suspect did not contribute.

3 **Victim** (reference profile) is alleles whose source is not disputed. Typically "victim" is a rape victim whose types are included in the vaginal mixture. The types of an acknowledged boyfriend or, in some cases, a convicted or presumed accomplice rapist, are logically also coded as "victim."

4 **Unknown contributors**. In addition to the explicitly specified alleles, the analysis model allows that each of the prosecution and defense may stipulate 0 or more additional unknown, untyped, and unrelated contributors to the mixture.

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[2] so-called, but this standard term isn't well chosen since in reality the exclusion method also in effect takes a likelihood ratio approach.

V.D.2.c. Practice examples (added – 2014)

There are two examples in the `dnaview\examples` folder. The example illustrations in this chapter are from the first example, case 128.

`NPmix.txt` Import as "GeneMapper" format. This big example has many mixtures, suspects, references, and includes rfu information. It will be case 128.

`Hoffmix.txt` Import as "Genotyper" format. This is an old small example with no rfu information. It will be case 10089.

V.D.2.d. Operation (see §V.D.3 for details) (rewritten – 2014)

Note – prompts and report wording can be (partly) in Spanish or German according to the setting per **Housekeeping** ▸ *Options* ▸ *Language* (§V.B.15).

Invoke either the stand-alone **Case** ▸ *Manual mixed stain*, or the automatic version (e.g. via *Open Case*).

(NOTE - restrict data [3])

The binary *Mixture ("LR") calculator* operation involves three main screens:

V.D.2.d.i.) Select mixture components

The screen shown in **Figure 19** allows the user to define a scenario in which the victim, unknown, and suspect profiles in terms of the roles in the case.

Operation then continues with the other screens alternately and iteratively.

V.D.2.d.ii.) Scenario calculation screen

From the scenario calculation screen (?) the user may select a locus to analyze or do various other operations such as view a calculation summary.

V.D.2.d.iii.) Mixture computation screen

The mixture computation screen is a worksheet that computes the likelihood ratio for a single locus. The screen is initially populated with any available data for the locus which may be either data from the case (in case of automatic use) and/or any data previously entered and saved for the locus. The user may modify the data in various ways, update the calculation, and exit back to the scenario calculation screen (§V.D.2.d.ii).

**V.D.3.** *Mixture ("LR") calculator* **operational details**

*Import* and *Open Case* 128.

V.D.3.a. Define a scenario: select mixture components (revised – 2014)

V.D.3.a.i.) Type **Z** in the first line as the mixture. Hit ***enter*** which activates the down arrow (since it's the highlit button).

V.D.3.a.ii.) Put **C** for the suspect and **G** for the reference. Note that the SCROLL button becomes visible when the input area is more leftward, so the scrolling trick (§V.D.3.a.v) is revealed.

[3] Exceptionally, *restrict data* (§V.B.26) first – useful if you want to selective use some loci but not others, or to use only one of repeated assays for the same individual and locus.

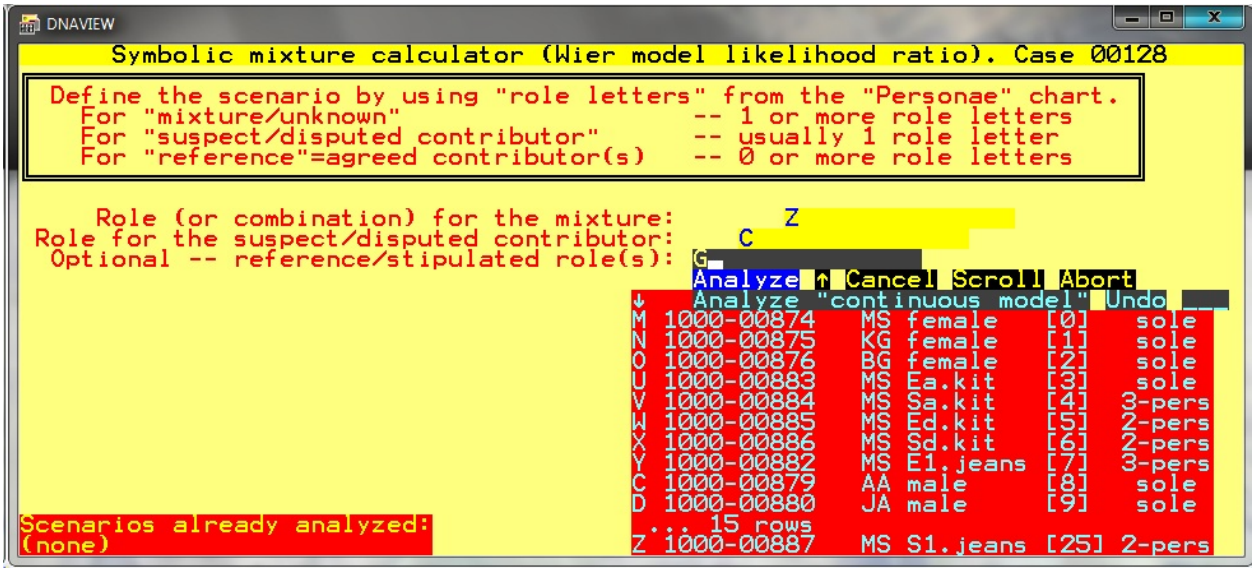



**Figure 20** Define a mixture scenario

V.D.3.a.iii.) **Note:** In case of the stand-alone *Manual mixed stain* there are no role letters; enter names.

V.D.3.a.iv.) **Note:** If you only want to resume the analysis of a saved calculation (per §V.D.4.d), it isn't necessary to specify any constituents. Just click ANALYZE, then RETRIEVE CALC at the subsequent screen.

V.D.3.a.v.) **Note:** Scrolling *Personae* – scrolling. The *Personae* cheat-sheet list has elided rows if there are too many to fit on the screen. The (semi-secret) hotkey ***page-down*** will scroll the list.

V.D.3.a.vi.) **Note:** It is permissible to enter multiple roles in a cell, or none. Comments:

| | zero roles (leave blank) | one role letter | ≥2 role letters |
|---|---|---|---|
| Mixture | not allowed | normal | synthetic mixture for demo or experiment |
| "Suspect" | rare, but allowed to permit comparing hypotheses differing only in number of contributors | normal | allowed but illogical (§V.D.3.a.vi) |
| Reference | common – whenever there are no agreed contributors | common – agreed contributor may be rape victim or boyfriend; may be the accused (§V.D.3.a.viii) | logical (but unusual) |

V.D.3.a.vii.) *Multiple suspects* are never logical for a casework analysis.

Suppose the prosecution hypothesis is that the mixture is DNA of semen from rapists **C** and **G** (and possibly other contributors). Then to evaluate the evidence against **C**, make a computation with **C** as the "suspect" and **G** as a reference. For the evidence against **G**, make a new computation with switched roles. If we calculate with **CG** as a combined suspect, the program will produce a (very large) number, but the number will be useless because we will not know how much of the number represents evidence against **C** (or **G**) in particular.

V.D.3.a.viii.) *Victim as suspect*. The logic of the mixture calculation is that "suspect" means a disputed contributor, which is not necessarily the same as the accused person.

For example, suppose t-shirt has a mixture that looks like a combination of the accused assailant **C** and the victim **V** of a violent crime. If the t-shirt belongs to the accused and is found in his home, then his own DNA isn't particularly incriminating but the presence of the victim DNA would be. Therefore for analysis in this situation **C** is an agreed contributor – just a reference, while **V** plays the role of "suspect" – disputed contributor – because it is the presence of **V** that is suspicious.

After specifying the constituents for each constituent category, ANALYZE (or – experimental work in progress – DNA·VIEW MIXTURE SOLUTION – see §V.F) to proceed with scenario analysis.

### V.D.4. Master calculation screen for a scenario

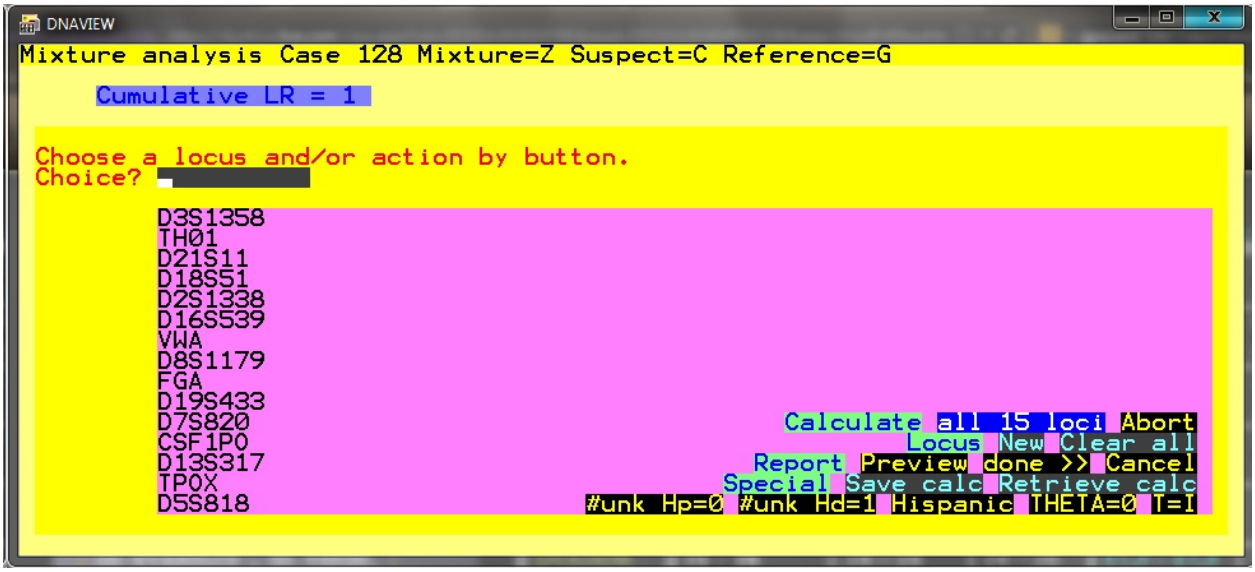



**Figure 21** Master calculation screen ("binary" mixture analysis)

From this screen you can calculate or recalculate all (§V.D.4.a.ii), some (§V.D.4.a.iii), or a single locus (§V.D.4.a.i), change calculation parameters, save a modified version of the data and later retrieve and continue working with it, view a report.

V.D.4.a. Calculation buttons

V.D.4.a.i.) To calculate and perhaps edit a single locus such as D3, arrow down to it and the CALCULATE: LOCUS button appears. ***Enter*** to trigger that (highlit) button opening the mixture calculation screen (§V.D.5) for that locus. Upon return from that screen, the **Figure 21** screen will be updated with the calculation for the locus.

Note that the locus menu includes LR= and parenthetical codes for loci that have been calculated.

The codes in parentheses — `(Cauc; 0, 1 unknowns; Th=0)` for example — mean:

`Cauc` racial origin of the samples in the database used for computation

`0` number of unknown contributors under prosecution hypothesis $H_p$

`1` number of unknown contributors under defense hypothesis $H_d$.

**Note:** Ideally the number of contributors is the same at every locus and if not, check the data and rework the computation carefully. However differing numbers of contributors and even half-contributors can sometimes be a way to approximate dropout or to calculate a conservative number for the benefit of the defense.

Th=0 shows whether and what $\theta$ value the computation uses.

LOCUS: NEW allows selection of a new locus for which the user may type in alleles. This is necessary when using the *Manual mixed stain* version.

V.D.4.a.ii.) CALCULATE: ALL 15 LOCI automatically calculates or recalculates all loci. This computation should be considered as only preliminary; almost for sure it will be necessary to review locus by locus such as

» to resolve issues of dropout and interpretation

» to review the automatic assumptions about the numbers of unknown individuals who also contributed to the profile, under each of prosecution and defense hypotheses.

V.D.4.a.iii.) CALCULATE: 7 LOCI automatically calculates remaining uncalculated (or cleared) loci but does not change calculations already done.

V.D.4.a.iv.) LOCUS: CLEAR CALC a selected locus to remove the *calculation* for the locus. LOCUS: CLEAR ALL deletes calculations from all loci. Any manual adjustments to alleles or interpretation (§?) are retained.

V.D.4.a.v.) LOCUS: REMOVE deletes a locus including all the alleles and calculations entirely from the scenario. (new – 2011)

V.D.4.b. Parameter settings

V.D.4.b.i.) # UNK HP and # UNK HD set the default number of unknown untyped persons to assume in the mixture for hypotheses of prosecution and defense respectively. The actual number will be larger if the default is insufficient to explain the observed alleles, or can be smaller if the user overrides on a per-locus basis.

V.D.4.b.ii.) The current computation race is indicated by a button e.g. HISPANIC. Click it to respecify the computation race. Recomputation requires an additional step – either revisiting individual loci or CALCULATE: ALL ...

V.D.4.b.iii.) THETA= allows setting the inbreeding/relatedness parameter $\theta$, $0 \le \theta < 1$, for mixture calculation. If $\theta=0$ the program produces a $\theta$-insensitive formula. If $\theta>0$, a $\theta$-sensitive formula per Curran *et al* is derived and used.

V.D.4.b.iv.) The calculation rules for an allele marked s**T**utter are toggled with the T=I or T=N button. See §V.D.5.e.ii.

V.D.4.c. Reporting (improved – 2014)

» REPORT: PREVIEW shows the report for the scenario calculation which can be printed or saved as a text file. The report includes a summary paragraph with the overall LR and a line per locus, as well as a paragraph per locus which is similar to the calculations screen for that locus.

» REPORT: DONE>> when through analyzing the present scenario. It bring you to a screen from which you can initiate a new analysis scenario, and view reports for all scenarios so far computed either per scenario (like that per PREVIEW or all together.

**Note**: "DONE" means that it will not be possible to resume and modify the scenario analysis (unless you saved it per §V.D.4.d).

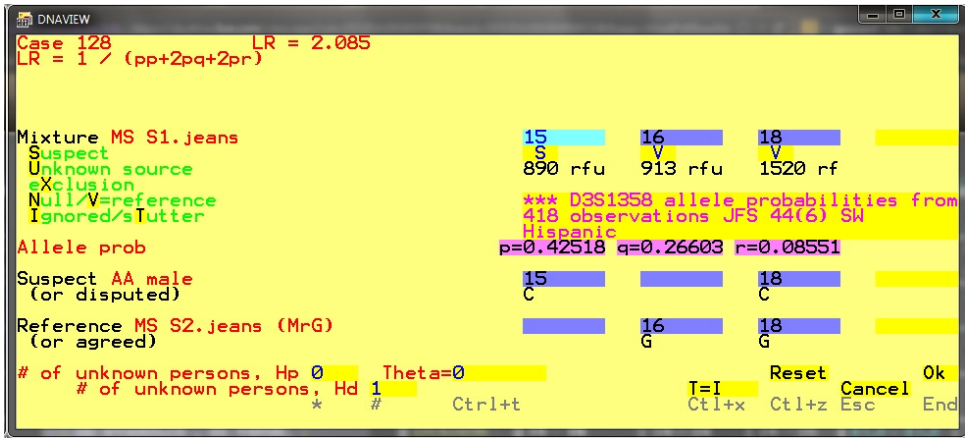
V.D.4.d. Interrupting your analysis (improved – 2014)

» SAVE CALC to save the calculation to a special file from which it is possible to later resume the same analysis.

» RETRIEVE CALC to retrieve and resume an analysis from the special file. **Caution:** If you open *Automatic mixture*, *select case* (some number), and then retrieve an analysis saved from a different case number, there may be confusion.

To resume analysis at a later date, note §V.D.3.a.iv.

### V.D.5. Mixture computation screen for one locus




A likelihood ratio, computed from the data shown in the screen, is shown both numerically and algebraically. You may be able to deduce from the terms in the formula what genotypes the program "thinks" could be contributed.

The screen shows various kinds of data, most of which the user is allowed to manually alter. The computation is updated when the data changes.

Each allele has a column. The rows are explained as

V.D.5.a.i.) The top group of three rows defines the mixture – allele, "interpretation", and (optionally) intensity.

V.D.5.a.ii.) "Interpretation" is a one-letter code which controls how the computation views this allele. These codes are automatically updated as alleles are added or removed, but also may be edited independently.

**V** marks a **mixture** allele that is (sufficiently) *explained* by its presence also in a **victim**, i.e. stipulated reference contributor. The use of V is

(a) to mark a stain band that is not incriminating to the suspect, even if he has it

(b) but doesn't need to be explained by an additional unknown party

(c) and to permit the flexibility of overlapping with bands of additional unknown parties.

**S** marks a **mixture** allele *explained* by the **suspect** and probative against him. It is

(a) an allele shared by **suspect** and **mixture** and

(b) either *not observed* in the **victim**, or (manual override) of insufficient intensity in the victim to completely explain its appearance in the **mixture**.

**X** means eXclusion. It marks a suspect allele not found in the stain. If there is an X the likelihood ratio is 0.

**N** marks a "null" allele, that is, either

(a) an allele present in the **victim** that does not appear and is not required to appear in the **mixture**, or

(b) an allele in the **mixture** that doesn't require an *explanation* as having a source, such as an artifact.

N is logically and computationally no different than V.

**U** marks an allele in the **mixture** that is explained by neither **victim** nor **suspect** and hence must be attributed to an unknown (and unrelated) person.

**I** means the column is ignored for computation, just as if it were deleted. (new – 2014)

**T** marks a probable stutter allele. See §V.D.5.e.ii. (new – 2014)

V.D.5.a.iii.) intensity. This information is optional and little used by the binary mixture program. It is mainly displayed, if available, as a guide to the analyst. (new – 2014)

V.D.5.a.iv.) The allele probability used for computation. The exact value depends on the allele probability *Case Options* (§V.C.4).

V.D.5.a.v.) "Suspect" alleles, or more precisely the reference type of a contributor assumed by the prosecutor and disputed by the defense. Role letters under the alleles help visualization.

V.D.5.a.vi.) (optional) Reference alleles for any agreed contributors, such as a sexual assault victim.

Notes:

» (non-typical situation) When a **mixture** found in the suspect's car includes the victim, probably the roles of suspect and victim are reversed for purposed of calculation, the victim profile taking the role of "**suspect**", the suspect profile being an **agreed reference**.

» If the prosecution alleges an *accomplice assailant*, then the accomplice types should normally be listed on the **victim** line, not **suspect**.

» If the **mixture** from a rape looks like a combination of suspect, victim, and the victim's consensual sexual partner, the victim and consual partner may both be considered as "victim".

» In the case of a one-person "mixture" it is easier to use *DNA Odds* instead of *Mixture ("LR") calculator*.

To remove an allele, use ***delete*** or replace it with 0.

If you enter an allele that exactly matches an existing allele in another row, the new allele will be moved to the matching column if the slot is open.

The alleles can be entered in a variety of notations. In case of an STR system, the usual method of entry is ***9*** or ***9.3*** or ***9+3*** or ***10-1*** indicating allele number and base pair offset. However 100 or more is interpreted as base pairs, except that ***#123*** again indicates allele number. RFLP alleles are interpreted as base pairs, or as kilobases (e.g. ***3.245***) if less than 100.

V.D.5.b. Add an allele: Click in the box to the right of **18**, and type **`17`** ***down-arrow***.

V.D.5.b.i.) Automatic sorting – Note that the columns automatically sort and a new blank column for allele 17 appears. (new – 2014)

V.D.5.b.ii.) Automatic LR recalculation: The LR calculation is updated. The # of unknowns for Hp had to be incremented. (new – 2014)

V.D.5.c. Ignored allele. (new – 2014)

Type **`I`** ***down-arrow*** which marks allele **17** as ignored and positions entry in the space below.

V.D.5.c.i.) Automatic recalculation includes reducing # of unknowns Hp to the default value of 0, so the LR is now the same as originally. Ignoring alleles is nicer than deleting them because it leaves them documented.

V.D.5.d. Manual edit of rfu value (new – 2014)

Type **140** ***up-arrow***.

V.D.5.e. Stutter allele (new – 2014)

Type **T** ***enter*** which marks allele **17** as stutter. Two effects:

V.D.5.e.i.) Stutter allele heights are shown as % of the parent allele – in this case 9% of the +4 parent **18** – rather than rfu. (new – 2014)

V.D.5.e.ii.) For calculation, depending on user toggle button choice, stutter alleles are either

» treated as null (**N** – same as **V**="no explanation needed") if the toggle is T=N. The computation then allows for the possibility that the allele might be a combination of stutter and an actual (scant) contributor.

This treatment is usually conservative, i.e. more favorable to the accused and it is therefore the default treatment. But it is easy to compare both calculations by toggling the T=... button to be sure.

» ignored (**I**) if the toggle is T=I.

This option is sound if you are confident that no real signal is masked by the stutter. Also if exceptionally this is the more conservative choice you might choose it for that reason.

**Note** Automatic stutter marking: By default the program will automatically mark **T** if the parent allele is observed and the rfu ratio is no more than 2×stutter ratio for the locus. (Delete the interpretation letter to allow the program to provide the default choice.)

V.D.5.f. Role letters

Below each suspect or reference allele is a role letter, such as **C** below the suspect **18** allele. This documents where the allele comes from. Multiple letters are possible, either for homozygous contributions or because of overlapping contributions when there are two references (or, unusually and probably inadvisedly, two suspects[4]).

V.D.5.f.i.) Considering the letters in combination with the rfu values can help the user to evaluate whether Hp (especially) is plausible.[5] (new – 2014)

V.D.5.f.ii.) Computation with roles: If and only if θ>0 the letters affect the LR computation.

V.D.5.f.iii.) You can type a role to add an allele: (new – 2014)

Click under the empty suspect cell in the allele **16** column, i.e. in the role row and in between the two **C**s. Type the letter **m** ***up-arrow***. Note that

» **M** appears in the cell (role letter entry is case-independent).

» **16** appears in the cell above.

V.D.5.g. Moving an allele

With the entry point in the **16** cell just created, type **17** ***enter***. Note that

» The allele along with its role tag **M** move to the **17** column.

---

[4] Even when the prosecution alleges (Hp) that the mixture is criminal defendants Jones and Smith, the fairest calculation is always to assume for purposes of evidence against Jones that Smith is a reference, not an additional suspect, and conversely for evaluating evidence against Jones.

[5] Future development: automatic plausibility evaluation

» The **17** column is automatically un-marked *stutter*. That seems the probable logical and useful behavior when a contributor is alleged for the column, but you can manually re-mark it as stutter if you insist.[6]

V.D.5.h. Buttons and parameters

There are four buttons and three parameters at the bottom of the screen. Activate/change any one by clicking on it, or typing the hotkey.

The parameters (# unknown persons Hp or Hd, and theta) and the T=... toggle button override just for this locus the default specified from the master calculation screen. (new – 2014)

V.D.5.h.i.) Hotkey tips. Beneath each in light grey is the name of the hotkey, i.e. type ***** to pop-up the dialogue to change the # of unknowns for Hp. (new – 2014)

V.D.5.h.ii.) RESET action button: Click on the RESET button and the screen reverts to its initial state.

V.D.5.h.iii.) CANCEL action exits the locus computation screen back to the master computation screen with no computation, result, or change.

V.D.5.h.iv.) OK button: Click this button – or ***end*** or ***ctrl-enter*** – to exit back to the master computation screen with the computation for this locus.

### V.D.6. Hints and techniques

If you understand how the mixture calculator works and use it expertly, then it has considerable flexibility to cater to dropout, ambiguous stutter, and peak intensity information.

V.D.6.a. How to handle stutter

See §V.D.5.e.ii.

V.D.6.b. Taking account of peak area/height information

The current version of *Mixture ("LR") calculator* assigns interpretation letter codes (V, U etc.) without regard to peak height/area. Under some circumstances it can be right to override the calculator's assumptions. For example suppose the mixture screen for some locus is

$$LR = 11 = 1/(2pr + 2qr + r^2)$$

| | | | |
|---|---|---|---|
| **victim** | 12 | 14 | |
| **stain** | 12 | **14** | **15** |
| **interpretation** | V | V | S |
| **suspect** | | 14 | 15 |

Suppose (as suggested by the boldface) that the mixture 12 is much weaker than the 14 and 15. Then the analyst may decide that the weak contribution from the victim does *not* explain the 14 allele in the stain. The way to take account of the extra information is manually type S as the interpretation of the 14 allele to *override* the automatic interpretation, then click `calculate`:

[6] Actually, marking it N makes a lot of sense in this situation.

| | | | LR = 20 = 1/2*qr* |
|---|---|---|---|
| **victim** | 12 | 14 | |
| **stain** | 12 | **14** | **15** |
| **interpretation** | V | **S** | S |
| **suspect** | | 14 | 15 |

V.D.6.c. Choosing the # of unknowns

The numerator and denominator of the likelihood ratio are probabilities that "belong" to the prosecution and defense respectively. Since the prosecution wants the likelihood ratio to be large and the defense wants it to be small, each side wants to make its own probability as large as possible.

For the prosecution this means choosing the most likely number — normally the obvious number — for **# of unknown persons Hp**, subject to the limitation that it must be a scenario that the prosecutor is willing to allege. This normally requires at least that the number of additional contributors be the same for each locus, although exceptionally it could be that different numbers are an appropriate way to cater to allelic dropout.

For the defense scenario, **# of unknown persons, Hd** similar considerations apply.

V.D.6.c.i.) Half an unknown person

The **# of unknown persons** is not required to be an integer; half-integers such as 0.5 or 1.5 are also permitted. The idea behind this experimental feature is a possible way to be helpful or at least fair to the defense by assuming (probably only for **Hd**) that an allele dropped out. Suppose for example that there are three unexplained alleles under **Hd**. The usual practice would be to assume 2 unknown contributors, or perhaps also to test the effect of 3 unknown contributors and give the defense the benefit of the smaller number. Taking this generosity one step further, test the effect of 1.5 unknown contributors. If that gives the lowest LR it may be the right number to report.

**V.D.7. Comments**

V.D.7.a. Crime Roles

V.D.7.a.i.) Use the role letters as you wish

The role names table (**Figure 154**, page 311) suggests for example using the role letter S for a suspect. This is not obligatory. The actual meaning of a role letter for calculation depends on where the user enters the letter in the screen shown in **Figure 19**, so you may use any letter however you wish . However, if you choose to use the letter S for the mixture for example, then it might be confusing if the report includes the word "suspect" next to the S. Therefore toggle the *Case Options* "Omit role descriptions" option (§V.C.5.a) to "yes".

V.D.7.b. Limitations

V.D.7.b.i.) Kinship

There is no provision to take account of possible relationship between suspect and unknown. It is expected to be included in Mixture Solution in the future.

V.D.7.b.ii.) Different races

All alleles at a locus are calculated as coming from the same population.

### V.D.8. Forensic Calculation Logic

Here are some examples of the way various scenarios are calculated.

V.D.8.a. Victim logic

Victim information is indicated by bands coded **V** on the calculation worksheet. Suppose for example there are 4 bands in the stain of which two match the victim and the remaining ones (the "unexplainable bands") match the suspect and are coded **S**.

Normally the bands matching the victim would be coded **V**. In that case the matching calculation assumes that there is only one assailant:

matching LR = $1/(2pq)$.

Whereas if there were no victim, or if for whatever reason you feel that there should be an additional assailant providing the extra bands, then assuming two assailants we compute

matching LR = $1/(12pq)$.

V.D.8.b. Two or one matching bands

If the suspect has *two* bands matching the two unexplained bands, then the obvious calculation results in a matching LR of $1/2pq$.

If the suspect has *one* band that matches an unexplained band then the calculation (questionably) ignores any extra suspect band. E.g. when there is only the one unexplained band, the matching LR is given as $1/q$.

V.D.8.c. Multiple assailants

If there are *extra unexplained bands* not matched by the suspect, code them **U** and the *multiple assailant* calculation is performed. More generally, if (# unknowns under Hd)>1, the calculation will take multiple assailants into account.

For example, if the unexplained bands are PQRS and the suspect is PQ, the logic and computation is as follows:

Imagine that the stain is fixed evidence, type=PQRS, and that the suspect's type, PQ, is the evidence to be evaluated probabilistically. That means we define

X=Pr(guilty suspect is PQ).

There are 6 consistent assailant types — PQ, PR, PS, QR, QS, RS — and they are equally likely once guilt and this stain are assumed, so X=1/6. Similarly, we define

Y=Pr(innocent suspect is PQ),

so $Y=2pq$. Consequently the matching LR, representing the strength of evidence against this suspect, is

matching LR = $X/Y = 1/(12pq)$.

### V.D.9. Mixture test suite

The *mixture examples* option in the (*Crime*) *Case* menu calculates the LR formulas for a set of mixture examples. Both θ-sensitive (per Curran *et al*, 1999; see §V.D.4.b.iii) and θ-less formulas are given. This suite can be used as part of testing and validation of the mixture LR calculations.

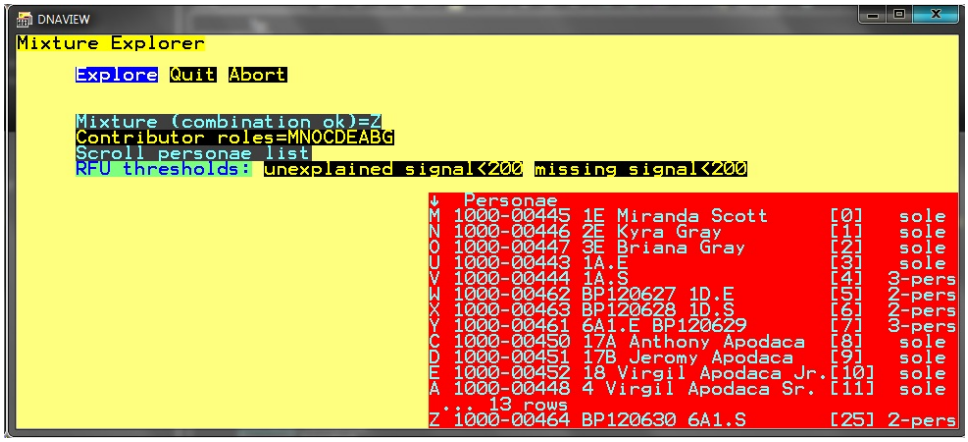
## V.E. *Mixture Explorer* (experimental)

(new with version 33.04 – 2014)

The *mixture explorer* is an option within *Automatic mixture*. It's useful to help sort out a situation a mixture and many profiles (e.g. reference) that may be contributors. It produces a report that can be used as suggestions for mixture scenarios to analyze with the *mixture ("LR") calculator*.

The *mixture explorer* considers combinations of possible contributors and evaluates the combinations by heuristics that take into account signal intensity. More below.

### V.E.1. Define search parameters




Each invocation of *Mixture Explorer* compares a single mixture against many combinations from a specified list of possible contributors.

V.E.1.a. The **buttons** in the screen above are used to define a search problem.

V.E.1.a.i.) Click MIXTURE or type **m** to enter a role letter (or letters, if you wish to artificially compose a mixture) representing the mixture to decompose.

V.E.1.a.ii.) Click CONTRIBUTOR ROLES or type **c** to enter a list of role letters to consider as possible components.

The *Personae* list is displayed to help you remember the role letters. If it doesn't entirely display

V.E.1.a.iii.) SCROLL PERSONAE LIST or **s** to see missing roles.

Then

V.E.1.a.iv.) EXPLORE to proceed with the analysis.

V.E.1.b. Suggested example: Case 128 (Npmix).

Mixture=**Z**. Contributors=**MNOCDEABG**.

EXPLORE!

### V.E.2. Exploring a mixture

The program tries to consider every subset of roles you specify as possible contributors. From 9 contributors as in this example, there are 511 combinations to consider. However for considerations of speed the program may limit itself to maximum 3- or 4- person combinations if the list is long.

The evaluation heuristic is as follows.

V.E.2.a. For a given combination, the program guesses what proportion of each contributor best explains the mixture. Thus, if nearly all of the alleles of some reference match high-rfu peaks in the mixture, a lot of the mixture is explained by assuming that a large proportion of the mixture DNA (molecules or rfu) is from that reference.

V.E.2.b. For the given combination and assumed proportions, another heuristic computes a degree-of-misfit score (lower is better, 0=perfect fit), the "CHB" (that's me) score.

**V.E.3. Understanding the results**

V.E.3.a. The combinations are sorted in order from lowest score to higher.

V.E.3.b. A report is produced showing the top several candidates. Following are a few examples with explanations:

V.E.3.b.i.) The top suggestion, CHB score=270, asks `Z ?=CG`, meaning is Z=**CG**?

```
Z ?=CG; 1.6=misfit/locus if Z= 0.44xC 0.55xG
   All mixture alleles are explained by the references.
   All reference alleles are observed in the mixture.
```

About 44% of the mixture DNA rfu would come from **C**, 55% from **D**.

All the reported mixture alleles are alleles that exist in the **C** or **G** reference profiles.

All the alleles in the **C** or **G** reference profiles are reported in the mixture.

In short, the mixture has exactly the alleles you would expect if it is a mixture of **C** and **G**.

`1.6=mixfit/locus` is a score reflecting allelic height discrepancies between observed and expected based on 44%**C**+55%**D**.

V.E.3.b.ii.) **CAG** explains the mixture nearly as well – not surprising if the **A** contribution is practically 0.

```
Z ?=CAG; 1.8=misfit/locus if Z= 0.44xC 0xA 0.55xG
   All mixture alleles are explained by the references.
   All reference alleles are observed in the mixture.
```

V.E.3.b.iii.) An example of a poor fit is **EG** with a bad (in context) score of 790.

```
Z ?=EG; 38=misfit/locus if Z= 0.084x? 0.3xE 0.61xG
Max unexplained signal=710rfu. * =unexplained allele
Max unobserved allele =660rfu. ()=unobserved but expected allele
 D3S1358   15-      16-         17-(E 503)    18-
 D16S539   11-      12-         13-*708
 D2S1338   22-      23-         24-           25-(E 367)
 CSF1PO    10-*625  11-         12-           14-
 TH01      6-       7-          9-            9.3-(E 663)
 VWA       14-      16-         17-(E 385)
 D21S11    29-      30-*223     32.2-(E 255)  33.2-
 TPOX      8-       10-(E 384)  11-
```

(I manually dug out the above analysis for demonstration. It would never appear on the report based on the rfu thresholds above because there are discrepant alleles with rfu intensities above the specified threshold of 200rfu.)

The heuristic best hypothesis includes a third unknown profile contributing 8.4% of the mixture.

Several **E** alleles are not observed in the mixture. These are noted in parentheses.

» `()=unobserved allele`. Based on a 30% contribution from **E** an allele #32.2 would be expected at about 255rfu (E 255), but no such allele was observed.

Those alleles in the mixture but not explained by either **E** or **G** are indicated with an asterisk.

» `*=unexplained allele`. In D21 we see that the observed mixture alleles are 29, 30, 33.2. The 30 allele was observed at 223rfu and – note the * – neither **E** nor **G** has that allele.

The loci listed are those at which either discrepancy occurs. The remaining loci that are not shown have exactly the alleles of **E**+**G**.

## V.F. *Mixture Solution – "continuous model"* **(experimental)**

(new with version 33.07 – 2014)

This program computes a mixture likelihood ratio using both allele size (i.e. repeat number) and intensity (observed RFU). It thus is able to

» take into account artifacts and complications such as dropout and stutter;

» realize that two peaks of different heights, although they may be possible from one contributor may be more likely from two contributors;

» estimate the proportional contributions from given references plus zero or more unknowns which best explain the data;

» take into account the possibility of dropin (provisionally – a model for dropin is implemented but I have not met anyone who seems confident that they know the right explanation for dropin);

The important consequence and benefit of all the above is that the program is able to

» make "sense" of complicated mixtures with 3 or 4, possibly even more, contributors;

» evaluate such mixtures objectively;

» produce a very high likelihood ratio against the suspect when justified;

» show why the suspect is not responsible when appropriate.

### V.F.1. Expert system

Mixture analysis requires formulating and comparing analytically (that is, by computation) hypotheses for prosecution and for defense. Mixture Solution, uniquely among mixture programs, automates that procedure. Every other mixture program[7] leaves to the expertise of the analyst to decide what hypotheses to evaluate, starting with specifying the number of contributors (per Clayton's widely cited but impossible advice), and also stating which reference profiles the prosecution and defense will assume are donors.

Mixture Solution offers a couple of alternatives:

V.F.1.a. Try all combinations

One alternative by Mixture Solution is that the program systematically tries all different combinations and prefers the hypotheses that explain the mixture best. Fortunately the good speed of the program makes this practical.

"All combinations" includes

V.F.1.a.i.) every subset combination of a list of reference profiles – i.e. from three people **A B C**, each of the eight subsets **ABC AB AC BC A B C** and **{}** (none);

V.F.1.a.ii.) for each such subset, any number from zero up of additional "unknown" contributors;

V.F.1.a.iii.) compare every such hypothesis against every other, importantly including comparisons between hypotheses with different numbers of contributors.

For example, see §xxx.

V.F.1.b. Wholesale evaluation of many donors

---

[7] The "exclusion" method doesn't require formulating hypotheses, but in part for that reason its results are meaningless hence we should not count implementations of the CPI, CPE, or RMNE method as "mixture programs."

A useful heuristic trick to screen quickly a large number of potential donors or elimination samples. This method is less rigorous than trying all combinations as §V.F.1.a but is even faster with a particularly large set of donors to consider.

See §xxx

### V.F.2. Caveat

A fair amount of validation work remains to be done. The program surely does reasonable things and provides useful insight. Preliminary checking tells me that the model is robust in the sense that the results are not very sensitive to parameter variations. However, it's too soon to rely heavily on the results. And a future version will surely include a provision to individualize the parameters for a particular lab's work product.

One possible approach for casework is to make a preliminary and tentative continuous version analysis and use it as an indication and a guide for a binary calculation.

The model permits user adjustment of the model parameters (§V.F.3.a.iii), but advice, especially automated advice, on the values of those parameters is not well developed yet.

### V.F.3. Operation of the continuous model

V.F.3.a. How to use

V.F.3.a.i.) Specify the role letters for mixture and possible reference profiles per §66.

V.F.3.a.ii.) Click DNA·VIEW MIXTURE SOLUTION.

V.F.3.a.iii.) Modify calculational parameters

Parameters affecting the calculation include maximum allowable number of unknown contributors, rate of stochastic variation, detection threshold, modification to stutter, tolerance for dropin. The initial, experimental release of the program does not allow the user to modify and experiment with these values.

They are documented on the output.

V.F.3.a.iv.) Wait for computation to finish.

V.F.3.a.v.) Save and or print the report.

V.F.3.b. What the program does

V.F.3.b.i.) Decide how many unknown contributors

The user has specified, under each of the competing hypotheses $H_p$ and $H_d$, which reference profiles are assumed to be contributors to the mixture. It is left to the program to decide how many additional unknown and untyped contributors best explain the mixture. It does this by starting at zero and incrementing (separately for each hypothesis). The searches continue until adding an additional contributor makes things worse. "Worse" ideally means reducing the likelihood of the observed mixture. But practically the search also has to stop when too many untyped contributors makes computation unwieldly, as tends to happen with 3 or at most 4 such contributors.

V.F.3.b.ii.) Consider contributor proportions

Corresponding to each hypothesis, for any particular assumption as to the proportional contributions a likelihood computation is rather quick.[8] The program evaluates each hypotheses (of assumed references

[8] as slow as a second or two if there are three unknown contributors, for each of whom all the trillions of possible genotype combinations are considered; orders of magnitude quicker for fewer unknowns.

+ unknowns) under a range of proportional contributions, an average of which goes into the final numerical likelihood ratio for or against the suspect as a contributor.[9]

Also of interest, though of unclear relevance, is the contribution proportions which best explain the mixture so these are also computed and reported, as well as likelihood ratios based on them.

As an example, suppose at some point the program, considering $H_p$, needs to evaluate the likelihood of the mixture Z assuming contributors C and G and no unknowns. It may be that the mixture is in fact a combination of those two contributors in proportion 4:1. If so, then hypothesizing C & G contributions in proportion close to 4:1 will probably come close to predicting both the observed alleles and the observed RFU values of Z (i.e. Z is "likely"), but under a moderately different assumed proportion such as 1:1 or even 3:1, Z is unlikely. Nonetheless, the entire range of contribution proportions must be considered since the prosecution and defense cannot reasonably hypothesize some particular ratio.

V.F.3.b.iii.) Produce a report

» Decide the appropriate likelihood ratio

The result report shows first the likelihood ratio for the hypotheses that seem most relevant:

```
Case 128 -- NP big mix                                  2014/2/1 22:47
Mixture=Z  Questioned=C  Known=G
appropriate conservative question:
Hp: AA male & MS S2.jeans (MrG) contributed -- VERSUS --
Hd:           MS S2.jeans (MrG) & 3 unknown people contributed.
LR (Hp vs Hd) = 19,32e15
```

» It includes tables with all likelihood ratios comparing various "number of unknowns" hypotheses for both prosecution and defense, and considering both likelihoods averages over all proportions,

```
      Component ratio: overall
 *LR Hp/Hd      | Hd=G&1unk   Hd=G&2unk   Hd=G&3unk
----------------+---------------------------------
   Hp=CG&0unk   | 30,43e15    96,08e15    19,32e15
   Hp=CG&1unk   | 1,473e15    4,652e15    935,4e12
   Hp=CG&2unk   | 179,1e12    565,5e12    113,7e12
   Hp=CG&3unk   | 23,13e15    73,02e15    14,68e15
```

(minimax LR indicated by boldface) and (for interest) likelihoods at the maximum likelihood proportion:

```
      Component ratio:  min pt
 *LR Hp/Hd      | Hd=G&1unk   Hd=G&2unk   Hd=G&3unk
----------------+---------------------------------
   Hp=CG&0unk   | 30,75e15    2,213e15    1,363e15
   Hp=CG&1unk   | 427,3e15    30,75e15    18,94e15
   Hp=CG&2unk   | 693,6e15    49,92e15    30,75e15
   Hp=CG&3unk   | 835,8e15    60,15e15    37,06e15
```

Note that both measure happen to show, in this case, that ever more unknowns benefits the defendant.

» Details of each likelihood computation are shown as well. For example:

[9] Up to 40 seconds are allocated for computing a scattering of contributor proportions.

```
 Hp       C       43%  <840rfu
          G       54%  <1055rfu
          unk#1   1%   <21rfu
          unk#2   1%   <21rfu
  4.78sec   117points   48  downhill steps
                        cost=116.46 @
                        C:0.434 G:0.545 0.01 0.011
  0.22sec     8points   nearby
 43.12sec  1140points   spaced overall
  -logL ave,minpt:123,3 116,5
d²-logL/d cof²=10785.18 at min (Δ=1/1000)
```

The algorithm doesn't seem to care much for the "unknown" contributions here, but doesn't want to dispense with them entirely. "<21rfu" means that, at the maximum likelihood contribution proportion, which includes a 1% contribution from each unknown, the maximum expected contribution from that contributor to any allele in the mixture is under 21rfu.

## V.G. *DNA Odds*

The *DNA Odds* command computes DNA profile matching LR's quickly on one screen for a variety simple (unmixed) stain scenarios in the forensic context that the suspect's DNA matches the crime scene DNA profile.

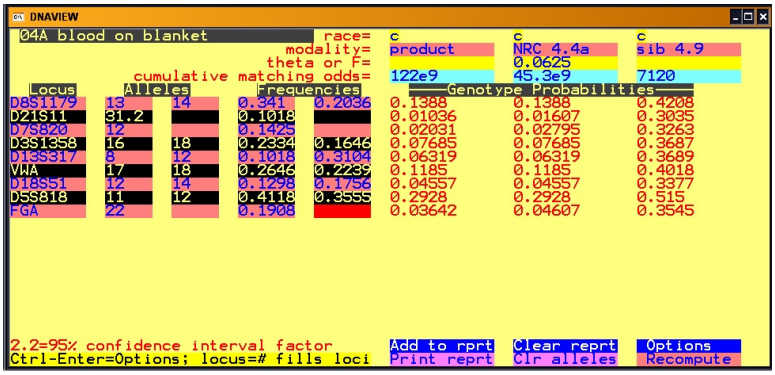



**Figure 24** *DNA Odds* screen

### V.G.1. Operation and capability

The program computes the strength of the evidence against the suspect as a likelihood ratio (§XII.A.2), the "matching LR".

Choose *DNA Odds* from the main menu or the **Casework** menu.

V.G.1.a. Outline of operation

V.G.1.a.i.) Enter autosomal genotypes either manually (§V.G.1.c) or automatically (§IV.E.1, §V.G.1.b)

V.G.1.a.ii.) Choose up to 3 computation "modalities" (i.e. product rule, sub-structured population, etc.) at one time (§V.G.3)

V.G.1.a.iii.) Choose a race for computation.

V.G.1.a.iv.) Print results or create a report

Move among the boxes with keystrokes (***space***, ***tab***, ***enter***, ***shift-tab***, arrow keys) or with the mouse.

V.G.1.b. Calculate an existing DNA profile

Bring up a profile according to its sample identifier (illustrated by the tutorial example §IV.E.1).

V.G.1.b.i.) ***ctrl-enter*** or ***left-single-click*** **Options**

V.G.1.b.ii.) choose *person*

V.G.1.b.iii.) `Data for whom?` Specify person either with accession number or role+case:

» accession number: Type the number, e.g. for `08-00123` type **`8`** ***space*** **`123`** ***enter***

» role+case: E.g. for case 1234, role M, type **`M`**

`Mother (M) of case #` **`1234`** ***enter***

V.G.1.b.iv.) If asked `race or ?` Just ***enter***

V.G.1.b.v.) `computation race?` **`c`** (enter desired race letter code)

The screen is now populated with the DNA profile and LR computation.

V.G.1.c. Calculate via manual entry or modification of a profile

V.G.1.c.i.) Optional: clear previous data

***click*** on **clear allele**, ***click*** **clear form** as desired

V.G.1.c.ii.) Populate first column with locus names

» type any abbreviation and the program will guess the locus. If it cannot guess uniquely, it will pop up a choice box., or

» type **#** for "next locus" according to the established locus order. Locus names will be filled in for consecutive blank cells. Therefore:

» to completely populate the loci, clear the loci (§V.G.2.a.iv) then type **#** in the first locus position. The loci and their order are defined per §IX.B.1.d.

V.G.1.c.iii.) Type in alleles as usual. (Only possible on a line that has a locus name entered.)

NOTE: If the allele resolution filter (ARF) is defined, then there will be a warning BEEP for any allele not mentioned in the ARF.

V.G.1.c.iv.) Make a typical or demonstration profile

For amusement or demonstrations: Enter **#** for an allele and the program will substitute an allele randomly selected based on the frequencies in` the relevant database.

Probabilities are calculated automatically, using the usual rules that are controlled by *Case Options* — including *Count this allele how many times?* You may override the probability specification.

FEATURE: Small numbers — e.g. 0.0001 — are shown as 1/ — e.g. 1/1000. 1/23e6 means "1 in 23 million".

You may enter a number by entering its reciprocal — e.g. type in 100 to get 1/100. (But you cannot type in the "e" notation.)

V.G.1.d. Choose alternate computation modalities(s)

V.G.1.d.i.) Move the type-in point (cursor) to any the three columns of the "modality" row, for example with ***single-click***.

V.G.1.d.ii.) Type all or part of the modality name if you know it. Or **x** ***space*** brings a pop-up menu of modality choices (§V.G.3). Choose the desired one.

V.G.1.d.iii.) In the `theta` space below the modality, enter a theta value if relevant for that modality.

The "1/" convention also applies here. So for example if *modality*=**relative** and you want to consider a 1/16 relative, just type **16** ***space***.

V.G.1.e. Choose an alternate race

» ***click*** on any of the instances of the race letter at the top of the screen.

» Type in a new race letter.

You can choose any race; databases are chosen according to the established "defaults," or you are prompted to select a database if there is no such. Present version uses the same race for all three computations.

Your work & your locus list is saved across sessions.

**V.G.2. Action keys and menus**

V.G.2.a. Action keys. ***SINGLE(!)-left-click*** for the indicated action.

V.G.2.a.i.) *Print reprt* — immediately print a screen.

V.G.2.a.ii.) *Add to rprt* – add this screen to the report. (Print or save the report later with *Print* or with **File** ▸ *save as*.)

V.G.2.a.iii.) *Clear reprt* – Start the report afresh.

V.G.2.a.iv.) *Clear alleles/form* – Clear all the alleles, leaving the locus list, or clear everything.

After clicking *clear alleles* clears the alleles, the button name changes to *clear form*. Therefore, click twice (slowly – these are single clicks) to clear everything.

V.G.2.a.v.) *Options* (also accessible via ***Ctrl-Enter***) gives you a menu including the above choices and also a few others:

» *Person* — prompts for an accession number, and collects the alleles for that person which is then calculated as a profile.

This method can accept a profile with many loci. The extra loci, beyond a screenful, couldn't be typed directly into the *DNA odds* screen, but the larger profile can be created in DNA·VIEW either through a method such as *Genotyper Import*, or *Type in a Read* (§IV.D.4).

» *Append allele counts* — include the allele count – database plus included observations – in the printable report

» *Uniqueness estimate* — an experimental feature that illustrates how commonly the profile would be seen in a random person who is not necessarily unrelated to the culprit.

### V.G.3. "Modalities" of computation – various NRC II options

The matching LR is the likelihood ratio comparing the hypothesis

$H_1$ The suspect is the donor of the stain

against some other hypothesis.

V.G.3.a. product rule and variants

These options compute the LR comparing $H_1$ versus the alternative hypothesis

$H_0$ The suspect is unrelated to the donor.

Three calculation modes are available:

V.G.3.a.i.) product rule (i.e. theta=0)

V.G.3.a.ii.) NRC 4.4a (correction for homozygotes, your choice of theta)

V.G.3.a.iii.) both NRC 4.4a+b (correction for heterozygotes as well, your choice of theta)

V.G.3.b. relationship situations

When one of the relationship "modes" is specified, the calculation under "genotype frequency" (perhaps misnamed) is $f$ = Prob( DNA profile | donor = relative of suspect). The reported matching LR is $1/f$ (cumulated over all loci) and represents the likelihood ratio favoring $H_1$ versus some "relative" alibi

$H_2$ The donor is a related to the suspect in a specified way.

V.G.3.b.i.) relative 4.8 (NRC II formula 4.8 with parameter F). By specifying an appropriate value for θ (=F), any unilineal relationship can be specified. For example, $\theta={}^1/_4$ means the relative is a parent or child and $\theta={}^1/_8$ means half-sibling, grandparent, uncle, niece, etc.

V.G.3.b.ii.) sibling 4.9 (NRC II formula 4.9 for chance of similar sibling)

V.G.3.b.iii.) relative 4.10 (Balding & Nichols formula for when suspect & culprit are in the same sub-population)

## V.H. *Y-haplotype matching LR*

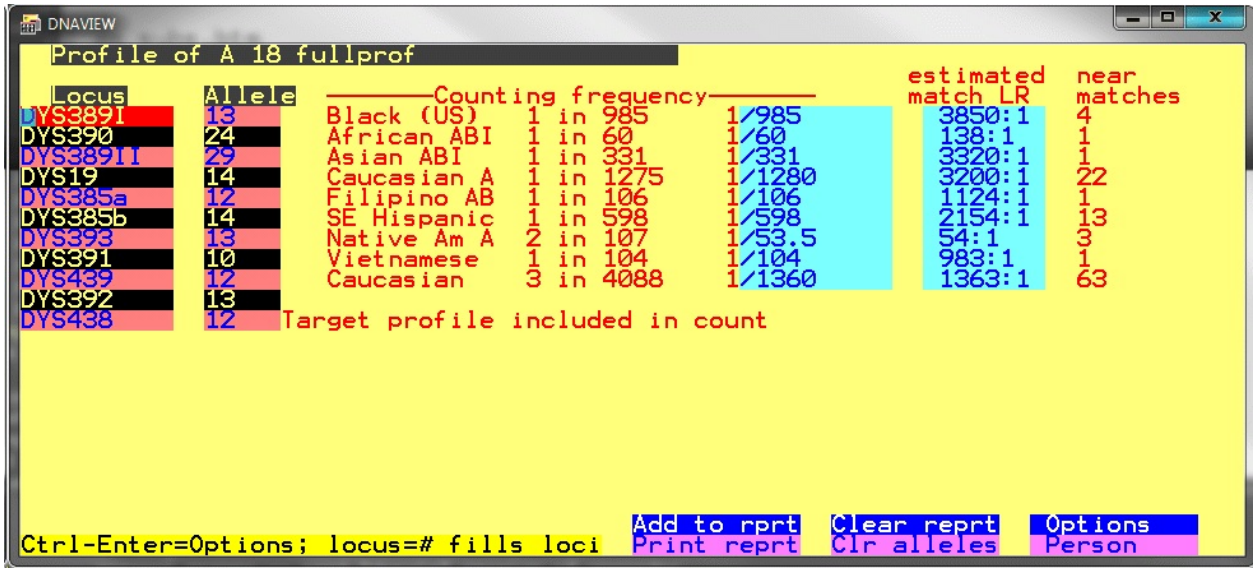



**Figure 25** *Y-haplotype matching LR* screen

*Y-haplotype matching LR* makes an assessment of the evidential strength of a Y-haplotype match.

The user may type in, or retrieve ("person") a Y-haplotype.

### V.H.1. Y-haplotype calculations

V.H.1.a. **Counting frequency** is the proportion of occurrences of the specified haplotype in the standard Y-haplotype databases. For purposes of counting, the specified profile may be temporarily included in the database, according to the setting of the option *Count observed allele in database how many times?* (§V.C.4.a) option. For example, in **Figure 24** the Caucasian comparison is with a database that has 2 matches to the target profile at all 11 reported loci out of 4087 comparable samples. (41 or the 4114 database samples are untyped at one of the target loci so are not comparable.) Temporarily including the target profile gives 3 matches out of 4088, hence 1/1362.7≈1/1360 as the "counting frequency."

The reciprocal of the counting frequency is a conservative estimate of the matching LR representing the evidential strength of a match to the population represented in the sample database.

V.H.1.b. **Estimated match LR** is the evidential value according to the model chosen per §V.C.4.d.

V.H.1.c. **near matches** – An experimental statistic, `near matches`, is also shown on the screen. This represents the number of haplotypes in the database that differ from the specified haplotype by at most one repeat unit at one locus (i.e. by 0 or 1 single-step mutation events). The only point of this statistic is to warn of a possible input error, in the suspicious circumstance that there are no near matches other than exact matches. The `near matches` statistic is not included in the printed report.

### V.H.2. Operation

Choose *Y-haplotype matching LR* from the main menu or the **Casework** menu. Navigation, printing reports, etc. are similar to the procedures for *DNA Odds* (§V.G.1) including the methods for populating loci (§V.G.1.c.ii) either by partial spelling or en masse with the **#** symbol.

V.H.2.a. Experimental demonstration feature: Select allele from randomly selected matching profile.

V.H.2.a.i.) The **#** symbol entered as an allele size searches among all the installed Y-databases.

V.H.2.a.ii.) A race letter (if accepted) as an allele selects a random matching profile from that race.

_____

See §VIII.F.2 for other Y-haplotype capabilities in DNA·VIEW including creating of Y-haplotype databases.

# V.I. *DNA Exclusion*

The *DNA Exclusion* command computes the so-called "exclusion probability" for a DNA mixture quickly and on one screen.

### V.I.1. Discussion of the "exclusion" statistic

V.I.1.a. Definition

The "exclusion probability", denoted $A$, is defined as the probability that a randomly selected person would have an allele not detected in the specified crime stain profile (which is assumed to possibly be a mixture), at any locus.

Examples:

V.I.1.a.i.) A suspect who is included in (or exactly matches) the crime stain profile for 10 loci, but has an allele not in the profile at an 11th locus, is counted as *excluded*.

NOTE: This may be an unrealistic assessment, for in practice perhaps it would be natural to re-examine the crime stain, and to reinterpret the odd-ball locus as "inconclusive" if there is even a hint of the "excluding" allele. If so, then actual "exclusion" decisions occur less often than the "exclusion" computation says they do.

V.I.1.a.ii.) If the crime stain appears to be the result of a single contributor — having only one or two alleles at each of many loci — and the crime stain is say 10,11 at some locus, the calculation counts a potential homozygous 10 suspect as being *not excluded*. The exclusion formula takes no account of the number of possible contributors.

V.I.1.a.iii.) If there is a non-zero null allele frequency (§VIII.C.12.e) specified for a particular locus, and the crime stain is for example 10,11, then the calculation counts a potential 10,null or 11,null (or even null,null) suspect as being *not excluded*.

The "probability of inclusion" or RMNE (proportion of "random men not excluded") is $1-A$.

V.I.1.b. Formula for "exclusion probability"

The exclusion probability, $A$, is computed as $A=1-B$, where $B$ is the probability of inclusion (also referred to as the RMNE). Note that $B$ can be calculated locus-by-locus, vis: $B = \prod_{i \in locus} B_i$. The screen display (§V.I.2, **Figure 26**) shows the numbers $B_i$ for each locus on the line for that locus, and the overall numbers $A$ and $B$ at the top of the screen. For each locus $i$, the fraction of people eligible at that locus is $B_i = \sum_{\text{genotype } G \in stain} Pr(G)$. For this purpose the probability Pr($G$) of a genotype $G$ is calculated as usin product rule, conservatively modified by using the NRC II formula 4.4a for homozygous types: $\Pr(QQ)=q^2+q(1-q)\theta$.

Genotypic combinations including the null allele are also counted. The null allele frequency is obtained from the database. See §VIII.C.12.e.

Each allele probability $f_i(a)$ is computed according to the rules detailed in §XIII.A.1.e covering minimum allele frequency and "counting the observed allele." Since each allele probability is computed separately and independently, the resultant total probability may be slightly generous. If the total probability adds to more than 1, a total of 1 is used instead. Note that the effect of the "generosity" is to understate slightly the exclusion probability — normally a favor to the suspect.

V.I.1.c. Limitations of the "exclusion" method

V.I.1.c.i.) Ignores part of the evidence

The computation of "exclusion probability" ignores the type of a particular suspect. If the stain has four alleles at some locus, two common and two rare, then the computation impugns a suspect with the two common alleles just as strongly as a suspect with the two rare ones. Occasionally the fact may be that the exclusion probability overstates the strength of the evidence against a suspect who has common alleles in common with the stain.

The exclusion probability might be unfair to the suspect.

However, while that can happen for one or two loci, it is unlikely that the net effect over all loci is unfair to a suspect.

V.I.1.c.ii.) Ignores the number of contributors

In this respect the bias favors the suspect.

V.I.1.c.iii.) Unrealistic assumption about exclusion

The point discussed above in §V.I.1.a.i suggests a bias against the suspect.

### V.I.2. Operation

Choose *DNA Exclusion* from the main menu.

Move among the boxes with keystrokes (***space***, ***tab***, ***enter***, ***shift-tab***, arrow keys) or with the mouse.

For loci, type any abbreviation and the program will guess the locus. If it cannot guess uniquely, it will pop up a choice box.

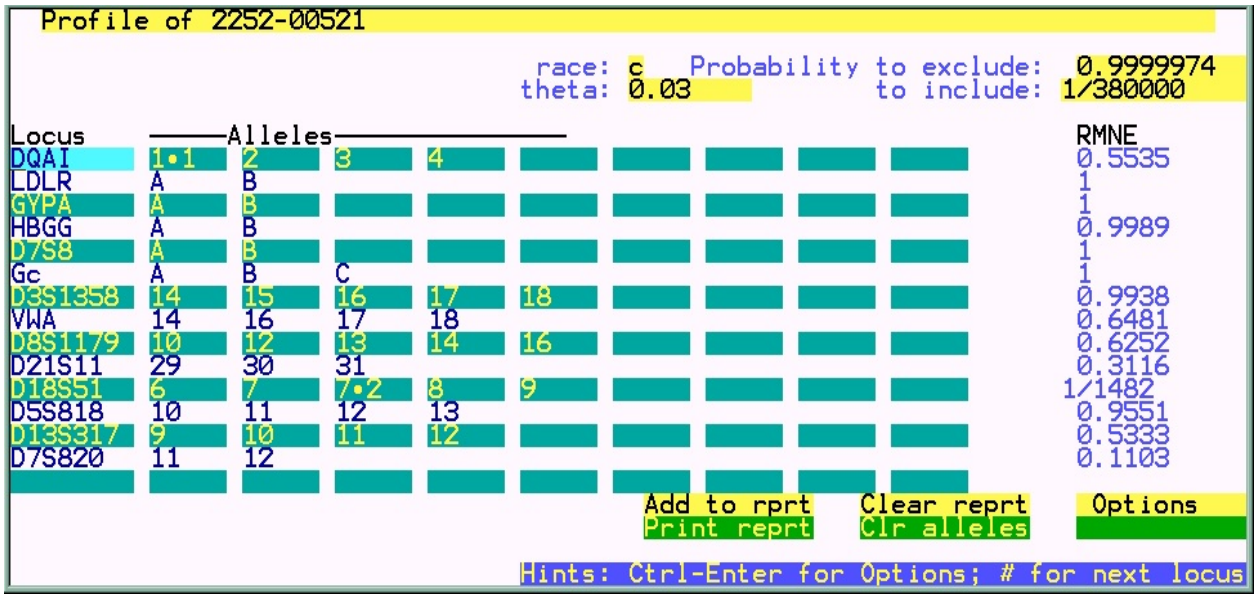



**Figure 26** DNA Exclusion screen

Type in alleles as usual. (Only possible on a line that has a locus name entered.)

NOTE: If the allele resolution filter (ARF) is defined, then there will be a warning BEEP for any allele not mentioned in the ARF.

Probabilities are calculated automatically, using the usual rules that are controlled by *Case Options* — including *Count this allele how many times?* During allele entry for a locus, probabilities are shown at the bottom of the screen, plus any null frequency, for the locus.

Your work and your locus list are saved across sessions.

V.I.2.a. Action keys

are the same as described in §V.G.2.a under *DNA Odds*.

V.I.2.b. Race

To compute with a different set of databases, click on the race letter and type a new race letter.

V.I.2.c. Theta

To compute with a different value of θ, click on the `theta` box and type in a new value. Note that a value larger than 1 will be inverted, so for example 0.01 may be entered by typing 100.

## V.J. Worklist

The *Worklist* program is used to create and modify worklist descriptions. A worklist description consists of

1) *Configuration* — obsolescent facility to configure calibration ("ladder") lanes for gel work.
2) *Roster* — the sample number or numbers associated with each lane,
3) *Identification* — a date and a comment.

For some laboratories, there will be a one-to-one correspondence between physical worklists/Genetic Analyzer (or similar) runs on the one hand, and worklist descriptions in the computer on the other. However, it does not have to be this way. For example, you may create your own large worklist of missing bodies or convicted offenders.


### V.J.1. Choose a worklist

Choosing a worklist is an operation that occurs from many programs. In *Worklist*, you select a worklist to examine or edit. *Import/Export* ▸ *DNA·VIEW Frequency Database Import* (§X.C.1) also shows the worklist list to begin. The names of worklists that have been created form a long list, one worklist per line, and each new worklist is added to the top of the list. Therefore worklists can be chosen by the usual menu selection procedures:

» The most recently referenced worklist is highlighted as a default choice and so can be immediately selected with ***Enter***.

» Context search: Typing **`car door`** will find those worklists with "car door" in the comment. It is a good idea to create comments for worklists with this in mind.

The worklist selection dialogue includes several extra useful buttons. (enhanced – 2014)

V.J.1.a. The NEW button is to create a new worklist, the normal procedure when importing profiles.

V.J.1.b. Two different **Show profiles per** options:

V.J.1.b.i.) WORKLIST – for a report of all the DNA profile data attached to (i.e. imported with) this worklist. Even data for unlabelled lanes will be shown. However, data from other worklists that pertain to samples on this worklist are *not* considered.

V.J.1.b.ii.) ROSTER – for a report of all the DNA profile associated with sample id's in the roster of this worklist. That includes data from *other* worklists for sample id's mentioned on this worklist.

V.J.1.c. **Find** options shrink the worklist list to just those which include the search target. Those can then be examined individually.

V.J.1.c.i.) Find worklist by CASE lets the user enter a case number, and finds all worklists including any sample ID from any sample in the case.

V.J.1.c.ii.) Find worklist by PERSON is similar.

V.J.1.c.iii.) (FULL LIST), because the previous operations shrink the list, is useful to restore it.

If you know a case number, you can reduce the list to all pertinent worklists.

V.J.1.d. **Change** buttons:

V.J.1.d.i.) DELETE OPTIONS to delete a worklist or some components of it per the pop-up choice menu shown at right.

```
Delete
   DNA -- all 12 loci ("reads")
   Roster -- all 4 lane labels
   both DNA and Roster
   altogether -- D+R+definition
   some of the DNA loci
   Rename the worklist
```

» *DNA -- all* k *loci ("reads")* removes the associated DNA data but leaves the worklist and roster intact.

» *Roster -- all* r *lane labels* is the quickest way to clear the worklist roster.

**Automatic deletion of cases:** (new – 2014) In addition, any *cases* which depend completely on the deleted roster entries *are also deleted*. But if, for example, even one person/sample ID of a case is in the roster of some other worklist, the case is *not* deleted. Automatic case deletion only occurs if deleting this roster means the case no longer has any connection to any worklist.

» *both DNA and roster* performs both of the above tasks (including automatic case deletion).

» *altogether -- D+R+definition* expunges the worklist entirely, along, of necessity, with associated DNA data, roster (and possibly cases as above).

» *some of the DNA loci* opens a menu allowing selection of individual reads to eliminate.

» *Rename the worklist* allows changing the date or comment in the name of the worklist.

V.J.1.d.ii.) RENAME goes immediately to the *Rename the worklist* option above.

V.J.1.d.iii.) ASSIGN RACE begins the operation in §VII.G, *Assign race to worklist*, operating on the currently highlit worklist.

### V.J.2. Create a worklist

This is a somewhat uncommon operation. Most worklists are created as a byproduct of *Import*.

Please see §IV.D.3 of the Tutorial for an example session.

*Worklist* begins by presenting a list of already created worklists. Assuming your worklist isn't already on the list, click the button NEW.

In response to prompts, you will supply identification — a date and a comment.

V.J.2.a. Select the SNP/STR configuration. (***Enter*** for OK).

At this point, the worklist is created and added to the list of worklists with an empty roster. You are presented with a button choice with NEXT>> highlit in yellow. Hence ***enter*** to continue.

### V.J.3. Enter or edit the roster


The roster information show for each lane is an accession number and possible an associated case number and role letter.[10] The case information is just that — information. You can't change it from the *Worklist* command.

You may put an (historically one or two, to allow for co-electrophoresis lanes) accession numbers into each lane.

Select a worklist for which you want to add or change the accession numbers.

The roster list, one line per lane, will be presented as a menu or choice screen (**Figure 29**).

Highlight (red bar) the lane/position you wish to change, then select a button:

V.J.3.a. OK to enter or change the accession number for the lane.

V.J.3.b. CLEAR LANE or ***delete*** to remove any accession number.

V.J.3.c. COPY FROM: WORKLIST to begin a dialogue to copy accession numbers from another worklist.

V.J.3.c.i.) Select the source worklist to copy from.

The screen splits with the source worklist on the top half, and destination worklist on the bottom.

V.J.3.c.ii.) Select a lane from the source worklist to copy. COPY LANE.

» OR CANCEL (STOP COPYING) to copy no more.

V.J.3.c.iii.) Select the next destination lane in the destination worklist and COPY FROM: WORKLIST. Continue from §V.J.3.c.ii.

V.J.3.d. COPY FROM: CASE opens the dialogue to find a sample/person within a case.

[10] If anybody is part of more than one case, the last case number is listed.

V.J.3.e. COPY FROM: QC opens a dialogue to copy a QC sample number.

V.J.3.f. NEXT>> to save the changes to the worklist. (FILE & QUIT)

**V.J.4. Done with roster definition**

When you click NEXT>> after editing the roster information for a worklist, a pop-up window with the menu of **Figure 30** appears. Select

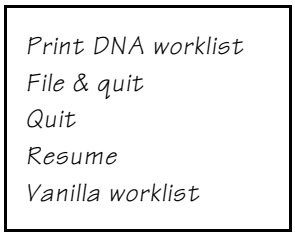



**Figure 30** *Worklist* exit menu

V.J.4.a. *Print DNA worklist?* to create and print the single column roster. Obsolescent; no obvious use.

V.J.4.b. *File & quit* to save the roster information & quit from *Worklist*. This is the normal way to exit from *Worklist*. If you forget to save, all is not lost though. If you invoke *Worklist* again and select the same worklist, you will be asked

```
File the interrupted work?
```

and you can answer **y**.

V.J.4.c. *Quit* to exit from *Worklist* aborting the editing operation. That is, discard all lane label additions or changes. Any changes in people definitions — race or accession number changes — however, have already become effective. ***Ctrl-Break*** (ABORT) would have the same effect.

V.J.4.d. *Resume* to make more roster entries or changes. Any "copy from" worklist you might have been using is forgotten.

V.J.4.e. *Vanilla Worklist* to create and print a simplified worklist.

## V.K. The File menu

Practically every command produces a report of some kind. This means that it leaves a printable result in the "report buffer."

### V.K.1. *Print*

*Print* makes a hard (printed) copy of the last report that was generated. The printed copy includes laboratory title (§IX.F.11), optionally workstation and/or software version (§IX.F.12) and the time and date.

The report buffer is erased by the action of any of the above programs beginning a new report.

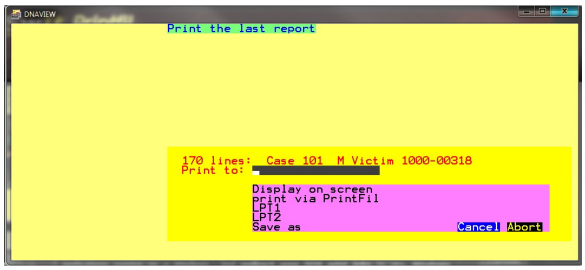


**Figure 31** printer "port" options

V.K.1.a. If the printer port is designated (§IX.F.10) as `Display on screen`, then the action of *Print* is to pop up a menu of options as shown in **Figure 31**.

V.K.1.a.i.) *Save as* (§V.K.2) to preview and/or save as a file.

V.K.1.a.ii.) The other options are per §IX.F.10.

### V.K.2. *Save As*

It is often useful to save a DNA·VIEW report – instead of, or as well as, printing (§V.K.1). The saved report can then be edited in any word processing program (Word, etc.)

V.K.2.a. Choose directory in which to save

`[drive:] path (folder)for text file:` **`reports`**

Careful – don't type the file name yet. Just the name of the folder.

» If the folder name does not begin with ****, then the address is relative to where DNA·VIEW is installed.

I.e. **`reports`** might mean **`C:\dnaview\reports`**.

» The special buttons DESKTOP, REPORTS, etc. are shortcuts for common locations.

The targets of those buttons can be customized by manually editing **dnaview.ini**. (new – 2014)

V.K.2.b. From the menu of files, either

V.K.2.b.i.) **replace/append to** and existing file by selecting it from the menu.

V.K.2.b.ii.) CREATE NEW FILE ***then*** type the name for a new file.

### V.K.3. *Show report*

*Show report* displays the current report buffer on the screen.

The display uses the bottom part of the screen, with the report arrayed using an attractive aqua color motif.

V.K.3.a. **Find:** Anything you type is interpreted as a case insensitive search key; **`. f`** finds all the lines like `#123. Family 2345.` NEXT to move to the next instance of the current "find" string. At the left middle of the screen, the first yellow-on-brown fraction – e.g. **23/114** – shows the ordinal count of the present instance of the find string, among all lines with the string.

V.K.3.b. PRINT or SAVE (like §V.K.2) if desired. CLOSE or ***esc*** to quit the report display.

Various situations in DNA·VEW use the report display in passing to display information.

## V.L. *DNA Profiles*

This command computes and reports the DNA profile probabilities for all the samples on a selected worklist.

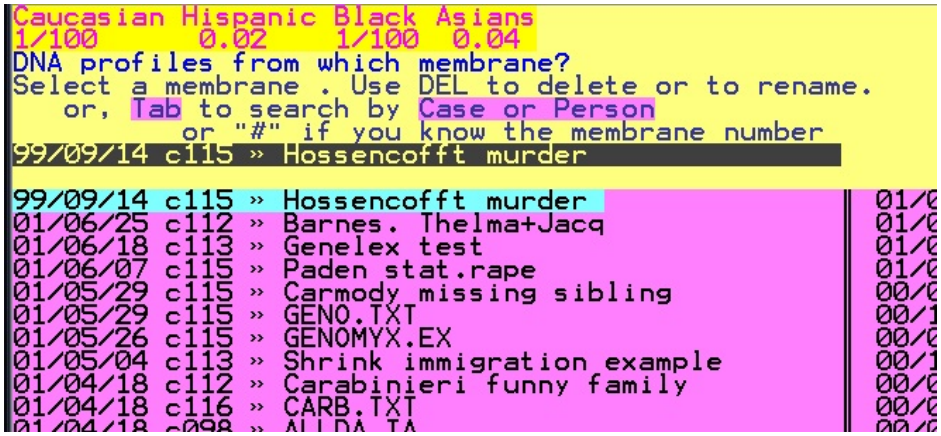

### V.L.1. Operation

Designate races for which to make computations. `Which races?` **chba**

V.L.1.a. Specify the parameter for population substructure for the calculations.

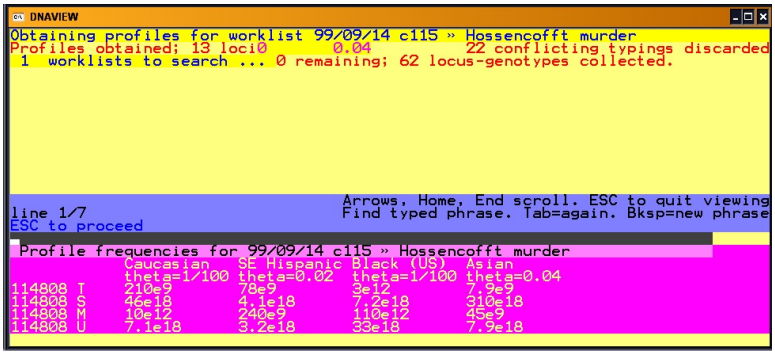
`What theta values(s)?` **0.01 0.2 0.01 0.04**

V.L.1.b. Select one or several worklists listing samples of interest.




V.L.1.c. The random match probabilities are now calculated under the various requested assumptions, and the report displayed. Hit ***esc*** when through viewing.

V.L.1.d. Print or file (=save) the report as desired.




### V.L.2. Comment

The selected worklists are treated as lists of sample identifiers, and based on those identifiers all DNA genotype data for those people anywhere in DNA·VIEW, even if associated with worklists other than those selected, are considered for the purpose of calculation. (This behavior is consistent with the way DNA·VIEW always calculates.)

# VI. KINSHIP



**Figure 35** Kinship chapter index

## VI.A. Conceptual Overview

The **Symbolic Kinship Program** solves general relationship problems of which paternity trio is the archetype. Other useful applications include:

i) inheritance disputes

ii) deficiency case (some relatives tested instead of alleged father)

iii) sibling questions (half siblings or full? identical twins?)

iv) incest situations

v) missing person problems

The program analyzes and compares arbitrary "pedigrees" or "scenarios," and makes computations, based on genetic data of the extent to which the evidence favors one scenario or another.

The algorithm is discussed in detail in Brenner (1997b), and some examples illustrating the method are presented throughout this chapter.

This chapter, VI, shows the steps using *Kinship* to analyze a typical kinship problem. §VI.L explains some special considerations: the tricky question of deciding twin-ship (§VI.L.1), and missing persons (§VI.L.2).

## VI.B. Kinship facilities in DNA·VIEW

### VI.B.1. Two versions of Kinship

There are two versions of the Symbolic Kinship Program in DNA·VIEW – "immigration" and "Kinship."

VI.B.1.a. *immigration/kinship*

The high-powered or "automatic" version of kinship (§VI.F, page 110) is a sub-command found in the *Automatic Kinship/Paternity/Automatic mixture* (in **Casework**) menus. Hence, it is available in the full DNA·VIEW system, but is not included in Abridged DNA·VIEW, in the DNA·VIEW Toolbox, or in PATER.

It is an automated and greatly refined version of the prototype *Kinship* (§VI.B.1.b), which requires less user attention and time, typically by one or two orders of magnitude, because it is integrated with the population databases and the imported and stored profiles of DNA·VIEW. Therefore

VI.B.1.a.i.) A user interaction is not necessary for each locus separately.

VI.B.1.a.ii.) The program automatically determines the genotype patterns; you don't type them in.

VI.B.1.a.iii.) The program automatically looks up the allele probabilities and calculates the likelihood ratios.

VI.B.1.b. *Kinship*

is a command in the **Research** sub-menu. It is the original version, a prototype for the later one, and is still essential in certain situations:

VI.B.1.b.i.) for the *Kinship* test suite (§VI.K.9);

VI.B.1.b.ii.) to obtain the formula for a hypothetical problem, without having any actual data;

VI.B.1.b.iii.) loci that are not defined as DNA loci;

VI.B.1.b.iv.) if you don't have the full DNA·VIEW system, but only Abridged, Toolbox, or PATER.

### VI.B.2. Limitations

The Kinship program (either version)

VI.B.2.a.i.) computes each locus independently and uses the product rule, so does not understand haplotype systems such as Rh or HLA, and is at least theoretically inaccurate (magnitude of error not clear) with X-linked markers, especially tightly linked ones, and with problems like sibship when any two markers share a chromosome,

VI.B.2.a.ii.) assumes the same population group for all individuals;

VI.B.2.a.iii.) takes a simplified approach to mutations (§XIII.C.4).

Future development work may cater better to mutations.

## VI.C. The Kinship notation

Kinship cases are thus defined to the program using a fairly simple but general notation. Using this language the user prepares a script and typically types it into the program using the *Type in scenario* option of the automatic kinship version, or, in the case of the stand-alone *Kinship* program, *Type in a new pedigree/locus*.

### VI.C.1. Kinship semantic concepts

The script defines normally two[11] *scenarios* that are to be compared:

VI.C.1.a. Principal scenario — The expected pedigree of a set of relationships among the people.

VI.C.1.b. Alternate scenario — An alternate pedigree or way they may be related.

It may include:

VI.C.1.c. Comments — these may describe the case, and perhaps (stand-alone *Kinship* only) make remarks about a particular locus.

The script refers to various

VI.C.1.d. People — some typed, some may be untyped.

and, only in the case of the stand-along *Kinship* version:

VI.C.1.e. Phenotypes — for some or all of the people.

### VI.C.2. Kinship syntax

Here are the basic syntax rules:

A *kinship script* consists of a collection of statements and/or comments.

Each statement either

(1) defines child-parent relationships,

(2) (stand-alone *Kinship* only) specifies genotypes for a person, or

(3) (stand-alone *Kinship* only) both.

VI.C.2.a. **Names**. The names of people must begin with a CAPITAL LETTER. Examples: **`Mom`**, **`C`**.

Note: The automatic version of kinship requires using *a single letter name* – namely the role letter – for anyone for whom there is a DNA profile.

VI.C.2.b. **Offspring-parent relationship**. The three-place predicate with the punctuation marks **`:`** and **`+`** denotes the biological relationship among child, mother, and father. Example: **`C:Mom+F`**.

VI.C.2.c. **Comment**. Anything after a semicolon (**`;`**) on a line is a comment and is ignored by the program. Example: **`C:Mom+F ; C is Charlie`**

VI.C.2.d. **Alternatives**. The symbol **`/`** designates the alternative scenario. It can be used in either of two ways. The two methods below **cannot** be used together – use one or the other!

VI.C.2.d.i.) Simple but verbose way to designate alternatives

Put the **`/`** symbol in front of each line of the alternative scenario.

---

[11] One hypothesis (§VI.C.4.b) or many hypothesis (§VI.D) are also possible.

VI.C.2.d.ii.) Concise but sometimes confusing way to designate alternatives

Let the / symbol separate two alternative names for the same role, one for each scenario.

Examples:

| | meaning: "Does the evidence support ..." | Simple way | Concise way |
|---|---|---|---|
| Paternity | ... paternity by **F** as opposed to by a guy named Joe? | **C : M + F**<br>**/ C : M + Joe** | **C : M + F/Joe** |
| missing body (equi-valent ways) | ... that the body **C** is the child of **M** and **F**, as opposed to of some other parents? | **C : M + F**<br>**/ C : Ma + Fa** | **C : M/Ma + F/Fa** |
| | ... that the body **C** is the child of **M** and **F**, as opposed to their child being someone else? | **C : M+F**<br>**/ Untyped:M+F** | **C/Untyped : M+F** |
| disaster identif-ication | ... that **C** is the child and **F** the father, as opposed to vice-versa? | **C : M + F**<br>**/ F : M + C** | **C/F : M + F/C** |

Advanced syntax rules (shortcuts):

VI.C.2.e. **Statement separator**

VI.C.2.e.i.) Multiple statements can be put on one line by separating them with the symbol **%**.

Example: **C:M+F % D:M+F/YY ; C & D share mother M. Do they share father F?**

The same effect can always be obtained by using multiple lines and putting one statement per line.

VI.C.2.e.ii.) When using the "verbose" syntax method (**/** at the beginning of alternative scenario lines), there is just one **/** or string of **/**'s at the beginning of a *line* of several statements, not one **/** per statement.

**C:M+F % D:M+F** ; primary hypothesis – full siblings

**/ C:M+F % D:M+YY** ; alternative hypothesis – half siblings

VI.C.2.e.iii.) **More attractive alternative separators**

| symbol | name | method to type |
|---|---|---|
| ♦ | diamond | ***Alt*** **~** (note: regardless of key caps, that's ***Alt*** with the upper-left-most key) |
| ☼ | starburst | ***Alt shift*** **8** |
| ∩ | cap | ***Alt*** **c** |

VI.C.2.f. **Full siblings**. As a shortcut, the comma (**,**) may be used to separate children who are full siblings.
Examples:

**C, D : M + F/YY** is equivalent to **C:M+F/YY ♦ D:M+F/YY**.

**C,E,D/Other : M+F** is equivalent to

**C:M+F ♦ E:M+F ♦ D/Other:M+F**.

Notes: (1) There is no shortcut for half-sibling situations; a comma won't help.

(2) It's better not to use the comma until you are fluent with the notation.

VI.C.2.g. **Computer-generated names**. For the lazy and unimaginative, the symbol **?** can be used instead of a name provided that

(i) The person is untyped.

(ii) The person is mentioned only once.

So for example **H:?+Pa ♦ J:?+Pa** is exactly equivalent to **H:Shirley+Pa ♦ J:Belle+Pa** (assuming the mothers are untyped); both mean that **H** and **J** are half-siblings.

Note: It is better not to use this shortcut until you are fluent with the notation. For pitfalls, see §VII.F.2. For a discussion, see §VI.C.4.a.

Examples using the shortcuts:

| | meaning: "Does the evidence support ..." | Simple way | Concise way |
|---|---|---|---|
| Paternity | ... paternity by **F** as opposed to by an unnamed person? | **C : M + F**<br>**/ C : M + ?** | **C : M + F/?** |
| missing body (equivalent ways) | ... that the body **C** is the child of **M** and **F**, as opposed to of some other parents? | **C : M + F**<br>**/ C : ? + ?** | **C : M/? + F/?** |
| | ... that the body **C** is the child of **M** and **F**, as opposed to their child being someone else? | **C : M+F**<br>**/ ? : M+F** | **C/? : M+F** |
| avuncular | ... that **F** is the uncle of **C**, as opposed to being unrelated? | **C : M + Bro**<br>**Bro, F : ?+?**<br>**/ C : M + ?** | **C : M + Bro**<br>**Bro, F/? : ?+?** |
| incest | ... that **C**'s father and uncle are the same person? ("Mom" assumed to be untyped.) | **C : Mom+U**<br>**Mom, U : ?+?**<br>**/ ; empty line!** | **C : Mom+U/?**<br>**Mom, U : ?+?** |
| sib-ship | ... that **A** and **B** are full siblings, as opposed to being unrelated? (Parents not typed.) | **A, B : ?+?**<br>**/ ; empty line!** | **A, B/? : ?+?** |

### VI.C.3. How to learn the Kinship Notation

The Kinship program computes a likelihood ratio comparing two different pedigrees as explanations for the same genetic data.

VI.C.3.a. **Two pictures**

The first step in transcribing a kinship problem into DNA·VIEW is to draw two pedigrees, on the left and right hand sides of the page, representing the two alternative explanations. Each pedigree is a diagram consisting of one or more family trees.

For example, a body is found – call it V – who ($H_1$) may be the missing child of F and brother of U, as in **Figure 36**. Alternatively ($H_0$, **Figure 37**), some "Other" body may be the missing child.

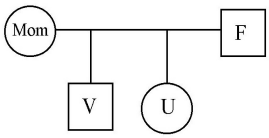



**Figure 36** full sibling

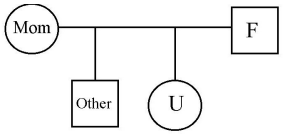



**Figure 37** unrelated

| $H_1$: The sample named V represents the victim who is the child of reference F and brother of reference U. | $H_0$: Some as yet untyped sample, called Other, is the victim who is the child of reference F and brother of reference U. |
|---|---|
| `V : Mom + F`<br>`U : Mom + F` | `Other : Mom + F`<br>`U : Mom + F` |

The trees should include all common ancestors. That is, don't stop with siblings connected by a horizontal bar; include their common parents even if those parents are not typed.

VI.C.3.a.i.) Names

Names begin with a CAPITAL letter. Subsequent characters may include digits.

Good names: `X F1 Bro DAD OtherWoman`

Bad names: `adolf saddam 1KNobe`

The people need to be named, and consistently in the two pedigrees.

VI.C.3.a.ii.) Single letter names

People who have been typed will be included in a "case," as such will have "roles" in that case, and the role letters (*capitalized!*) must be used as the names of such people. Other people – untyped people – may be given either long or short names at will.

VI.C.3.b. **Two pedigree transcriptions**

Under each picture – on the left and right sides of the page – transcribe the pictures into linear notation.

VI.C.3.b.i.) For each child, describe that child's relationship to its parents by writing a line using the three-place predicate with the punctuation marks **:** and **+** to separate **child**, **mother**, and **father**. See the captions of the figures above.

Consequently each pedigree diagram may result in several lines of transcription.

VI.C.3.b.ii.) Do not use the **?** or **,** shortcuts for either of the transcriptions.

VI.C.3.c. Two ways to enter the pair of pedigrees

VI.C.3.c.i.) Simple way

| | |
|---|---|
| Type in every line of the primary hypotheses | `V : Mom + F`<br>`U : Mom + F` |
| Type in each line of the alternative hypotheses preceded by a slash (/) | `/ Other : Mom + F`<br>`/ U : Mom + F` |

» If the alternative hypothesis is *empty* – no relationships – see §VI.C.4.b.

VI.C.3.c.ii.) Concise (classical) method – **Merge the two transcriptions.**

In the middle of the page, merge the two transcriptions into a single set of statements by using the symbol / wherever necessary to separate alternate versions of the same person. Thus:

```
V/Other : Mom + F
      U : Mom + F
```

To be rigorous about what is needed at this step: The program will take the merged description and scan it twice, once reading only name on the left side of each /, and once reading only the name on the right side of each /. These two scans must recover the two different transcriptions written in step §VI.C.3.b.

This method gives a more concise formulation but sometimes merging seems tricky.

The (either) result of §VI.C.3.c.ii may be typed into the program.

VI.C.3.d. The advanced **notational convenience options** – the **?** for a computer-generated name, the comma (**,**) for *full* siblings – can be introduced *after* the merging but I emphatically recommend against using them before gaining experience.

VI.C.3.e. **Genotype syntax** (stand-alone *Kinship* only)

The genotypes for one locus can be included in the kinship script in either of two ways:

VI.C.3.e.i.) as a separate statement (line) for any typed person; the name of the person, then the genotype:

```
V pq
Mom qr
```

VI.C.3.e.ii.) interlineated in a relationship statement:

```
V pq/Other : Mom qr + F
U : Mom + F
```

As permitted shortcut for the very economical-minded, the name can be omitted if the genotype is present:

```
pq/Other : Mom qr + F
U : Mom + F
```

Of course, if the person needs to be referenced twice, as `Mom` in the above example, you still need to include the name.

Genotypes are designated by one or two lower case letters, such as **pq**. A few special rules:

VI.C.3.e.iii.) The letter **o** can only be used for a null allele.

VI.C.3.e.iv.) Homozygotes are normally written with just one letter, like **q**. Writing **qq** instead is normally equivalent, but it does also mean that **qo** is not a possibility. If you write simply **q**, the *Kinship* analysis depends on the setting of the *silent allele* toggle — see §VI.K.2.a).

VI.C.3.e.v.) For an X-linked system, male types should be written with a dash, i.e. **r-**.

### VI.C.4. Kinship Semantics, clarification, and examples

VI.C.4.a. Understanding the **?**

The **?** does not mean "alternate father." It merely denotes any untyped person whom you do not care to take the trouble of naming. It may be and often is used to denote an alternative father, but the "alternative" meaning is not part of the semantics of the **?** but comes rather from the syntactic fact of following a **/**.

Thus, the definitions **Johnny : Mom + Dave/?** and **Johnny : Mom + Dave/Stranger** are completely equivalent (provided nothing else is said about *Stranger*). If there is only one occurrence of **Mom**, you can shorten further to **Johnny : ? + Dave/?**.

The **?** means a *different* person each time it occurs, and it is not permitted to attach to it genetic types. Therefore it may be used (instead of inventing a name) if and only if the following two conditions are met:

1. The person only needs to be mentioned one time.
2. The person is untyped.

VI.C.4.b. Design criteria for the kinship notation

The kinship syntax described above was designed with the simultaneous goals of being general (any problem can be defined), simple (easy to understand) and convenient (easy to type in — which mainly means concise). Whenever there are multiple goals, there is bound to be some conflict. To resolve the conflict between simple and concise, I allow two input forms – one simple (§VI.C.3.c.i), one concise (§VI.C.3.c.ii).

Almost any pair of pedigrees can be transcribed into the notation, but not quite all — identical twins pose a problem as discussed later in this chapter.

### VI.C.5. One hypothesis

It is also allowed to enter just one hypothesis. In that case the program automatically considers the alternate hypothesis to be that no one is related. So entering **C:M+F** is equivalent to

```
 C:M+F
/C:?+?
```

(

```
 C:M+F
/
```

).

VI.C.5.a. Why one hypothesis?

VI.C.5.a.i.) when the alternative is "no one is related."

VI.C.5.a.ii.) When using *Simulation* just to generate an example (§VI.G.4.d), the alternative is irrelevant.

### VI.C.6. Prior probability

A prior probability may optionally be specified for kinship calculations, from the kinship menu. If a prior is given then a posterior probability W is computed and presented according to the formula

$W=LR/[LR+(1/prior)-1]$.[12]

Specifying the value 0 means no prior probability; no posterior probability computation.

The value specified is preserved across sessions. To avoid accidents, the value, if non-zero, is advertised on the top part of the screen above the kinship menu in addition to being documented in the menu.

[12] This way of writing Bayes' formula has a nice interpretation. Suppose there are 20 equally probably identities for a victim. Then prior=1/20, and the formula becomes W=LR/(LR+19). In other words, add to the denominator the number of alternate identities.

## VI.D. Multiple hypotheses in *Kinship* and *Immigration/Kinship*

A *Kinship* scenario may compare more than two hypotheses.

### VI.D.1. Multiple hypothesis syntax

VI.D.1.a. example

The input syntax is based on the "simple method." Example:

```
   C:M+F    ; Lines without leading / are the primary hypothesis.
/  C:M+Unc  ; one leading / for the 1st alternate hypothesis
/  Unc,F:?+?
//    C:M+?       ; two leading /'s for the 2nd alternate, etc.
```

The three hypotheses are: The man F is the father; the man F is the uncle; the man F is unrelated.

VI.D.1.b. rules

VI.D.1.b.i.) Each line belongs to only one hypothesis. Thus any stipulated relationship – such as that **M** is the mother of **C** in the example above – has to be stated repeatedly under each separate hypothesis.

VI.D.1.b.ii.) Each line begins (except for permitted leading spaces) with a string of 0 or more **/**'s, indicating to which hypothesis the statement(s) of that line belong. If there are several statements on a line (separated by statement separators §VI.C.2.e), all belong to the same hypothesis and the string of **/**'s must be at the beginning of the line. If the statements for a hypothesis occupy more than one line, each line must begin with the requisite number of **/**'s. Thus, the lines may be in any order.

VI.D.1.b.iii.) Any number of hypotheses are allowed. The primary hypothesis consists of the lines with no **/**'s. A line with a string of *n* **/**'s is called the *$n^{th}$ alternative hypothesis*.

### VI.D.2. Multiple hypothesis result

The result is a list of the relative likelihoods corresponding to the several hypotheses. For example:

```
400 : 100 : 1.
```

These correspond to the hypotheses *primary*, *1st alternative, 2nd alternative*.

VI.D.2.a. Interpretation of multiple likelihoods

VI.D.2.a.i.) The ratio of any two likelihoods is the likelihood ratio supporting the first corresponding hypothesis over the other one.

Thus, from the example above,

» 400 = 400:1 is the LR by which the evidence supports the primary hypothesis over the 2nd alternate;

» 100 = 100:1 is the LR by which the evidence supports the 1st alternate hypothesis over the 2nd alternate;

» 4 = 400:100 is the LR by which the evidence supports the primary hypothesis over the 1st alternate.

VI.D.2.a.ii.) "Overall" likelihood ratio

If the likelihood for the primary hypotheses is much greater than all the other likelihoods – if the likelihood *ratios* are all greater than <threshold>, then we can conclude that the LR supporting the primary hypothesis is at least <threshold>.

Example: $5 \cdot 10^9 : 10^3 : 1$ – primary hypothesis is supported by at least 5 million over either other hypothesis.

Example: $5 \cdot 10^9 : 10^6 : 1$ – primary hypothesis is not so strongly supported compared to an alternative.

VI.D.2.b. Conclusions even when some LR's are less than <threshold>

If the primary hypothesis is not <threshold> better than all the other hypotheses we need to take a closer look. Suppose the nearest rival is either

» not different in a significant way, or

» inherently not very plausible.

Then the data still may be good enough for a decision.

VI.D.2.c. Hypotheses give the same conclusion

The first case is the easier. Suppose that the hypotheses are

*primary*: remains **V** is Edward and remains **W** is Katie

*1st alternative*: **V** is Edward but **W** is unrelated to Katie

*2ne alternative*: **V** and **W** are unrelated to Edward and Katie,

that the likelihoods are $5 \cdot 10^9 : 10^6 : 1$, and that the threshold for identification is $10^6$. Then we can conclude that **V**=Edward because each of the first two explanations are at least <threshold> superior to *any explanation under which* ***V*** $\neq$ *Edward*.

VI.D.2.d. Dismissing inherently implausible explanations

"Inherently implausible" means having a particularly low prior probability. Therefore if we are willing to assign prior probabilities to the various hypothesis we may be able to draw conclusions that go beyond the simple-minded approach of §VI.D.2.a.ii.

VI.D.2.e. Posterior probabilities

The normal behavior of the *prior probability* parameter, that, if it is non-zero the program should compute a posterior probability, does not apply in the case of multiple scenarios.

However, the *Immigration/Kinship* menu includes the experimental interactive option *Bayes calculator* (§VI.F.6.e) to compute posterior probabilities as explained in §XII.E.1.f.

## VI.E. Automatic Version of Kinship – Overview

The automatic version of kinship is accessed as the *Immigration/Kinship* option of the *Automatic Kinship*, the *Paternity Case*, or even the *Automatic mixture* command – collectively called a *Case* command, which are found in the **Casework** menu.

Calculations for all loci are performed with no (or minimal, or optional) user intervention, based on genotype data that has been supplied to DNAVIEW in any of the same ways that it would be supplied for a paternity case calculation. Probabilities are normally obtained from DNAVIEW databases automatically also (rather than being typed in by the user as is the case with the prototype *Kinship* command).

### VI.E.1. Preliminary

The case to be analyzed needs to be defined as a numbered case. Normally the case includes several people (i.e. accession numbers), each occupying a unique *role* within the case. Each role is designated by a single upper-case letter. Some or all of the people usually have genetic data – DNA profiles – associated with them. The case should also have one to six *race codes* – a list of lower case letters – associated as it's *race list*.

VI.E.1.a. Defining a case

A case is defined either by

VI.E.1.a.i.) using the *Genotyper import* or similar option of the *Import/export* command, with suitably specified *Sample Info*,

or by

VI.E.1.a.ii.) invoking the *select case* option from the *Case* menu, and typing in a case number.

VI.E.1.b. Including people in a case

To include people in a case, either

VI.E.1.b.i.) use the *Genotyper import* or similar option with suitably specified *Sample Info*,

or

VI.E.1.b.ii.) after *select case*, the *Case* options *edit people* or *add role*.

Put all the people of interest into the case, without particular regard to role (child, mother, etc.)

VI.E.1.c. Creating DNA profiles for people in a case

The DNA profiles are associated with a worklist. The worklist has person (accession) numbers associated with some of its lanes as its *roster*, and it has *reads* – each containing the DNA types for one locus – also associated with the worklist.

A worklist, its roster, and associated reads can be created either by

VI.E.1.c.i.) using the *Genotyper import* or similar option with suitably specified *Sample Info*,

VI.E.1.c.ii.) using *Worklist* to create the worklist, and *Type in a read* (§IV.D.4) to create each read manually,

or by a combination of the two methods.

VI.E.1.d. Creating a race code list

Both methods of creating a case generally create a race list. However, it may be desirable to edit it.

Select the option *edit race list* from the *Case* menu to change the race list. Include all races of interest in the list, with the most likely race first.

## VI.F. *Immigration/kinship* – operations

Select the *Immigration/kinship* option of the *Paternity/Automatic Kinship/Automatic mixture* command (in the **Casework** menu) to access the automatic kinship menu options.

MENU: BASIC options



MENU: PROBABILITY includes *Prior probability* above and also:



MENU: SCENARIO/CALCULATE includes some of the above and also:



MENU: OPTIONS includes some of the above and also:



**Figure 38** *Immigration/Kinship* options

MENU: Y-CHROMOSOME includes *Show pariwise relationships* above and:
§VI.F.6.b *Include Y-LR in result.* . . . . . 119
§VI.F.5.a *Y-haplotype summary.* . . . . . 118
MENU: X-CHROMOSOME includes various of the above.
MENU: ESTIMATE includes various of the above and:
§VI.F.4.d *Racial estimate.* . . . . . 117
MENU: REPORT/SUMMARY includes various of the above and also:
§VI.F.3.a *Add LR calculation to report.* . . . . . 115
§VI.F.3.c *Print summary of calculation.* . . . . . 115
§VI.F.3.d *Create summary file of calculation.* . . . . . 115
§VI.F.7.c *Eyeball check the raw sizes.* . . . . . 120
§VI.F.7.d *Eyeball check the genotypes.* . . . . . 121
§VI.F.7.f *Symbolic genotype chart.* . . . . . 122
MENU: POPULATION includes some of the above and also:
§VIII.C.2.b *Change database defaults.* . . . . . 160
§VI.F.7.h *Next race from: Caucasian/Hispanic/….* . . . . . 122
MENU: DATA includes several of the above.

MENU: ALL includes everything above.

**Figure 39** *Immigration/Kinship* options

**VI.F.1.** *Type in (or edit) scenario*

Typical steps for a kinship case are:

Type the problem definition into the large aqua box at the bottom of the screen using the syntax rules of §VI.C.

```
N:M+? ; N & V are half-sibs
V:M+F
/N:M+? ; or V is unrelated
```

OK (hotkey ***ctrl-enter***) when done entering the scenarios.

VI.F.1.a. Automatic and saved kinship scenarios

From *Type in or edit scenario*, click the button SPECIAL KINSHIP SCENARIO and the program offers several new button choices:

VI.F.1.a.i.) SAVE AND/OR FETCH FROM PICKLIST opens the pick list. You may pick a previously stored scenario from it, and/or re-save the pick list with the new scenario added.

Note that including the case number in a scenario comment in the pick list enables the pedigree searching program to find it for searching that case.

VI.F.1.a.ii.) GET SCENARIO FROM CASE COMMENT will extract any scenario found in the case comment.

VI.F.1.a.iii.) SAVE SCENARIO TO CASE COMMENT stores the typed-in scenario in the case comment.

VI.F.1.a.iv.) CREATE BODY ID SCENARIO creates a scenario based on interpreting the role letters.

VI.F.1.b. Example: creating and saving a body ID scenario in the case comment.

The case must use the letter V to designate the body to identify. Suppose in addition the case has references with roles M, U, A, H, P, D and the words corresponding to these role letters are mother, two siblings, a half-sibling, the spouse, and daughter of the missing person.

VI.F.1.b.i.) Invoke *Type in or edit scenario*, click SPECIAL KINSHIP SCENARIO and click CREATE BODY ID SCENARIO. Then the scenario

```
V/Missing, U, A:M+Fa
D:P+V/Missing
```

appears in the *Type in* box. Note that the scenario-generator didn't include H. The half-sibling relationship is ambiguous so you have to take care of it manually. So, if the half-sibling is paternal, add another line to create

```
V/Missing, U, A:M+Fa
H:?+Fa
D:P+V/Missing
```

VI.F.1.b.ii.) Proofread. The automatically-generated scenario may not be what you mean. Half-siblings aren't the only problematic relationship. Check it carefully.

VI.F.1.b.iii.) Then click SPECIAL KINSHIP SCENARIO and click SAVE SCENARIO TO CASE COMMENT.

VI.F.1.c. Typing rules

The standard rules for text or numeric entry (§XIV.A.4) apply including UNDO, INSERT LINE, DELETE LINE, and HISTORY of recently typed scenarios. Copy and PASTE are possible but not quite the same way as normal for Windows.

VI.F.1.c.i.) Long scenarios

» If you use up all the lines in the box and still need more room, OK back to the menu and re-invoke *Type in scenario*. This will give you at least three more blank lines.

» Several statements can go on the same line using % or other characters to separate them (§VI.C.2.e).

VI.F.1.d. More input hints

» Button (No) Names toggles a reminder display of the names and roles in the case.

» **Note** the on-screen note hints: ***f10*** (to type a **/**), ***f11*** (types the separator ♦), and ***f12*** (types all of **: + ♦** and you can then intersperse the child, mother, and father names).

VI.F.1.e. Table of genotype patterns

| D3S1358 | | VWA | | FGA | | Amelo | | D8S1179 | | D21S11 | | D18S51 | | D5S818 | | D13S317 | | D7S820 | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| pr | V | qr | V | pr | V | x | V | p t | V | p r | V | rt | V | p | V | p | V | pq | V |
| p t | M | p r | M | r | M | xy | M | st | M | r | M | r | M | p | M | p r | M | q | M |
| p u | N | p r | N | r | N | x | N | s | N | qr | N | pr | N | p | N | qr | N | qt | N |
| r | F | q | F | pr | F | xy | F | p t | F | p a | F | rt | F | pq | F | p | F | pq | F |

Legend: D3S1358 p=13 r=15 t=17 u=18 VWA p=16 q=17 r=18 FGA p19 r21
D8S1179 p10 s13 t14 D21S11 p28 q29 r30 a33.2 D18S51 p13 r15 t17
D5S818 p11 q12 D13S317 p9 q10 r11 D7S820 p9 q10 t13

**Figure 40** Table of symbolic genotypes

The table of genotype patterns can be helpful to understand the relationships and the results.

It is displayed on the *Type in (or edit) scenario* screen, and also as part of the *Estimate likely relationships* (§VI.F.4.b) screen.

It is normally included as part of the printed (log) report (§VI.F.3.b, §I.E.7).

**VI.F.2.** *Calculate & report LRs, all races*

Compute the likelihood ratios symbolically – that is, as algebraic expressions – for each locus. Then race by race (for each race in the race list (§VI.E.1.d)), evaluate per locus, compute the product over loci, and add to the report summary box (**Figure 41**) with the bottom line for each race.

```
Caucasian cumulative LR   7.44e9
Hispanic  cumulative LR   5.33e9
Black     cumulative LR   292e9
```

**Figure 41** Kinship LR's, all races

Repeat with additional scenarios if desired.

VI.F.2.a. Reporting kinship results – report log

The *Calculate & report* option automatically appends the summary box (**Figure 41** ) to the report log (§I.E.7). The more detailed per-race, per-locus calculation summaries (**Figure 42**) are available if desired, or the report log can be kept very brief:

```
Include per/locus details for each race in report? n
```

VI.F.2.b. Reporting kinship results – formatted report

Depending on the reporting options (§V.C.5) a results file may be created automatically.

The typical appearance of the file is approximately as **Figure 41**. Note that it is quite similar to the text file optionally created by *Paternity Case*, *calculate paternity(maternity)* (**Figure 19**).

Use the DNA·VIEW Report Viewer (§XV.D.2.a.iv) to view the report in Excel.

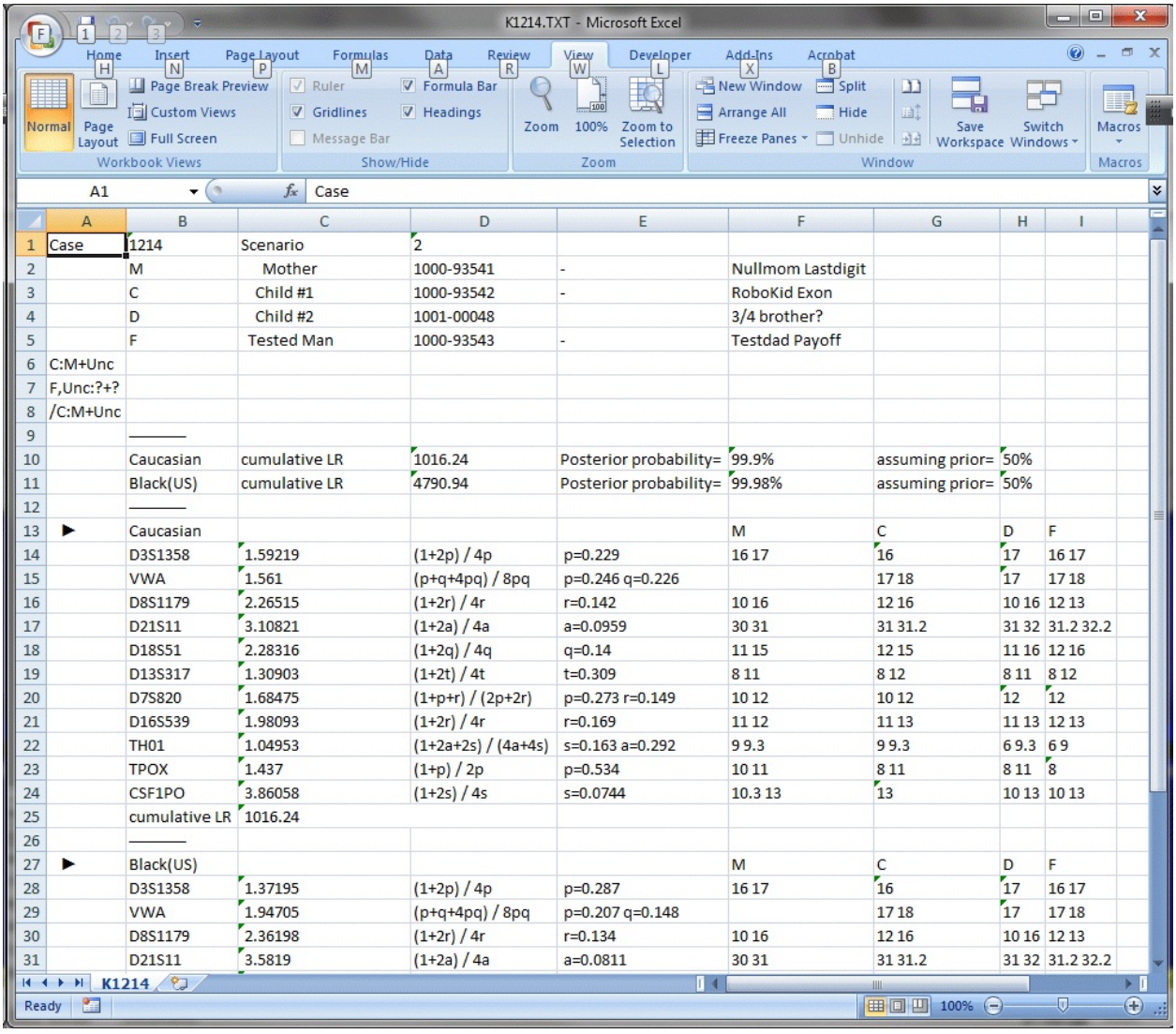



**Figure 42** Kinship "formatted report"

VI.F.2.c. Formatted report name

The name of the formatted report file is presented on the screen, and you may change it. Several points:

» Blank the name (delete it or just hit ***space***) and the report is skipped.

» Change the name, and the new name becomes the standard name for kinship reports.

VI.F.2.d. Genotypes and genotype probabilities

During the evaluation the program prepares tables of relevant genotypes and genotype probabilities. If it comes to a locus for which there is no appropriately specified (see *Set default database, per race*, §VIII.C.2.b) database, it will prompt you to select one. (And, optionally, you may specify the selected database to be selected automatically in the future.)

If you refuse to select a database (hit ***enter*** with no selection), then you will subsequently be prompted to supply probabilities. So this is a viable option.

**VI.F.3.** *Calculate LRs (current race)*

computes the symbolic likelihood ratios and evaluates them only for a single race, displays on the screen (**Figure 43**) and does not append the result to the report. As opposed to *Calculate & report LRs, [all] races* (§VI.F.2), use it for an experiment the result of which you might not want to keep.

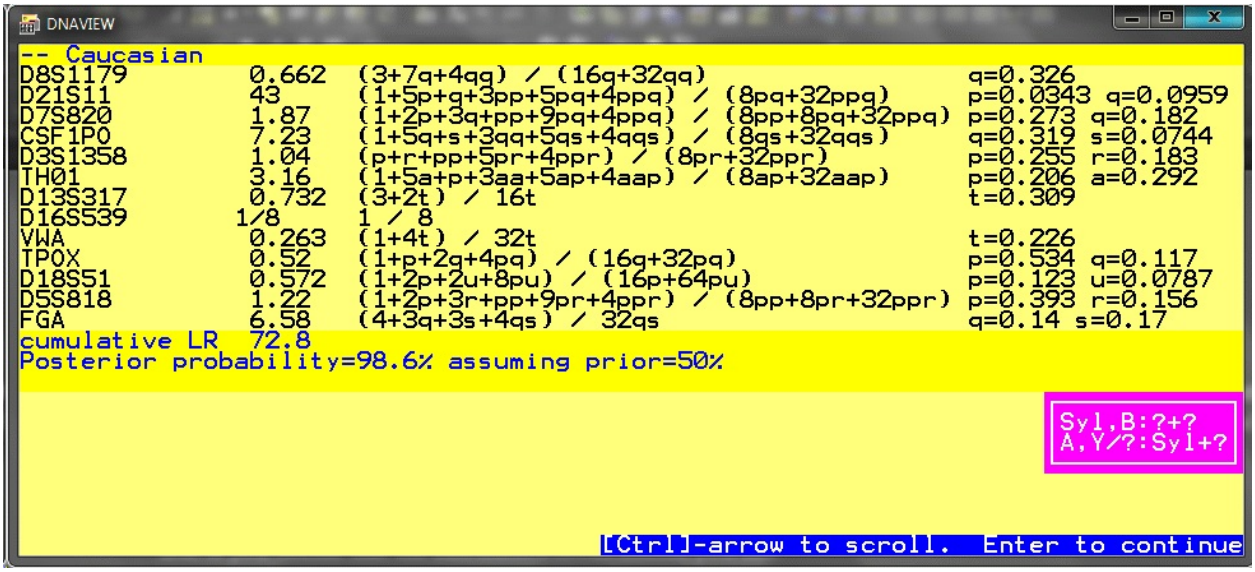



**Figure 43** *Kinship* LR details

But if you decide that the calculation looks useful, you can

VI.F.3.a. *Add LR calculation to report*

adds the (tentatively) experimental calculation of *Calculate LR (current race)* – §VI.F.3 – to the report.

VI.F.3.b. *Show (log) report* is equivalent for the kinship log to the *Show Report* (§V.K.3) command.

VI.F.3.c. *Print summary of calculation*

VI.F.3.d. *Create summary file of calculation* – same as the file created automatically by *Calculate & report LRs, all races* (§VI.F.2), except of course including only the one race.

### VI.F.4. Estimating races and relationships

VI.F.4.a. Which relationships?

To get a preliminary overview of a complicated case it's often helpful to begin by comparing the DNA profiles by pairs in order to guess who is related and how closely. The *immigration/kinship* menu includes three different computation options which are similar in spirit but differ in data and details.

| | loci used in calculation | | Display format | | |
|---|---|---|---|---|---|
| | autosomal | Y | LR's | symbolic genotype chart | |
| | | | | autosomal loci | Y loci |
| *Show pairwise relationships* | ✓ | ✓ | scrollable/sortable menu shows up to all 5 LRs for every pair | none | |

| | | | | | | |
|---|---|---|---|---|---|---|
| *Estimate likely relationships* | ✓ | | triangular distance chart | maximum 2 LRs shown | ✓ (left-right scrollable) | ✓ |
| *Y-haplotype summary* | | ✓ | | matching LR only | | ✓ |

The "pair-wise relationship" facilities described following, as well as the kinship search of *Screen disaster matches*, compute the pairwise relationship likelihood ratios (LR) for each of the following hypothetical relationships (using, in the autosomal case, the fast algorithm per Brenner Weir 2003, Brenner Bieber Lazer 2006SOM):

| abbrev-iation | name | description |
|---|---|---|
| `id` | identity | Matching LR is computed for the proposition that the two profiles from the same source *versus* unrelated. This computation is included not only to find identical twins, but also to permit including personal references as if a special kind of relative.<br>The identity index is tolerant of allelic dropout; the odds are computed as ½ for each locus that has a discrepancy that can be explained as allelic dropout. It is also slightly tolerant of a mistyping, using the STR mutation model. |
| `pi` | parentage | A 2-person parentage likelihood ratio is computed that includes tolerance for mutations. |
| `si` | sibling index | LR comparing whether the pair is full siblings *versus* unrelated |
| `hsb` | half-sib | sharing one parent. The LR for aunt, uncle, niece, nephew, grandparent, or grandchild are the same formula, except when there is a consideration of mutation. |
| `cuz` | cousin | LR comparing whether the pair is cousins, or unrelated. |

☞ Be aware that you will often get a slightly different number by doing an actual kinship calculation than the number given in a pairwise chart. The reason is the dropout and mutation approximation approach of the pairwise calculation. Therefore the pairwise charts are intended only to give preliminary clues as to likely relationships.

VI.F.4.b. *Estimate likely relationships*

Compare all the typed people in the case pair-wise. Display a chart showing all the plausible relationships (page 5). This is helpful for guessing at the likely results of explicit kinship tests and for deciding which experiments to try.

| | M | N | F |
|---|---|---|---|
| V | pi=20000<br>si=400 | pi=1/60<br>hsb=3 | pi=7000<br>si=100000 |
| M | | pi=10000<br>cuz=100 | cuz=1/8 |

**Figure 44** estimated relationships

The numbers in each cell evaluate the corresponding pair of people as potentially related per §VI.F.4.a. If the people may be the same (rule: `id>1` and `id` is the largest of the LR's), then the `id` is shown and nothing else. In any case no relationship index $<^{1}/_{100}$ is shown. Among the miscellaneous relationships beyond parentage, at most the single largest one is shown. In short, in order to conserve screen space at most two indices are shown.

☞ Note: Y haplotypes are *not* included in calculating the estimated relationship chart.

VI.F.4.b.i.) Genotype display diagram

Below the relationship chart (**Figure 44**) a colorful diagram of the symbolic genotypes is shown (see page 5). Among other details, note that if this chart is too wide


**Figure 45** Customizing genotype display for page 5.

for the scree n, the entire chart can still be viewed by horizontal scrolling using the cursor arrows or the ***home*** or ***end*** keys.

You can customize the selection genotype diagram vertically in response to the prom pt `Specify rows desired`. For example, if mother M has three children A,B,C, then it can be helpfult to create rows **`AMB CM`** – repeating the maternal genotype and introducing a blank row – to facilitate visualizing inherited genes.

» **Symbolic genotype letter assignment –** Note that for discrete allele systems (i.e. PCR), consecutive letters represent consecutive alleles (so a possible 1-step mutation), and similarly letters 2 or 3 apart represent a 2- or 3-step difference.

Larger alphabet distances do not correspond strictly to allele steps though.

VI.F.4.c. *Show pairwise relationships*

This option is similar to *Estimate likely relationships* but it presents a scrollable chart – a menu actually – which shows all relationship indices such that LR>$^{1}/_{100}$. It also differs in that the *Show pairwise relationship* ☞ *does* include Y haplotype evidence if there is any. *n*The chart can be edited and re-arranged for easier viewing.

VI.F.4.c.i.) sort

» by column using one of the letters **`a`**, **`b`**, etc above the columns to sort according to the LR values in that column. For example, **`b`** sorts according to `pi`. Sorting again on the same column reverses the sort order (like Windows does).

» by largest LR for the row using the key **`+`**.

VI.F.4.c.ii.) remove uninteresting comparisons

Move the red bounce-bar (single click or ***arrow***) to a row then ***del*** to remove it.

VI.F.4.d. *Racial estimate*

For each profile, compute the relative likelihood of seeing it in each of the races of the race list. In this example there is good evidence that **N** is not Black, but not much evidence of anything else.

```
Relative probabilities assuming
each racial origin:
    Caucasian  Hispanic  Black
 V    1         1/1.8     1/8.6
 M    1          1.8      1/3.6
 N    1          1.3      1/27
 F    1         1/1.2     1/6.6
```

**Figure 46** Racial origin likelihoods

The results table is included as part of the report log – i.e. the report that you can print or file after exiting from Immigration/kinship. A report with the same information can also be obtained by using the *Racial Estimate* command from the main menu (§VIII.N).

### VI.F.5. Y-haplotypes

Kinship does not know the genetics of the Y-chromosome. However, it does have useful facilities which allow inclusion of the Y-haplotype matching LR into the kinship likelihood ratio through user interaction.

These facilities depend on Y-haplotype databases being installed in your DNA·VIEW system (§VIII.F.3).

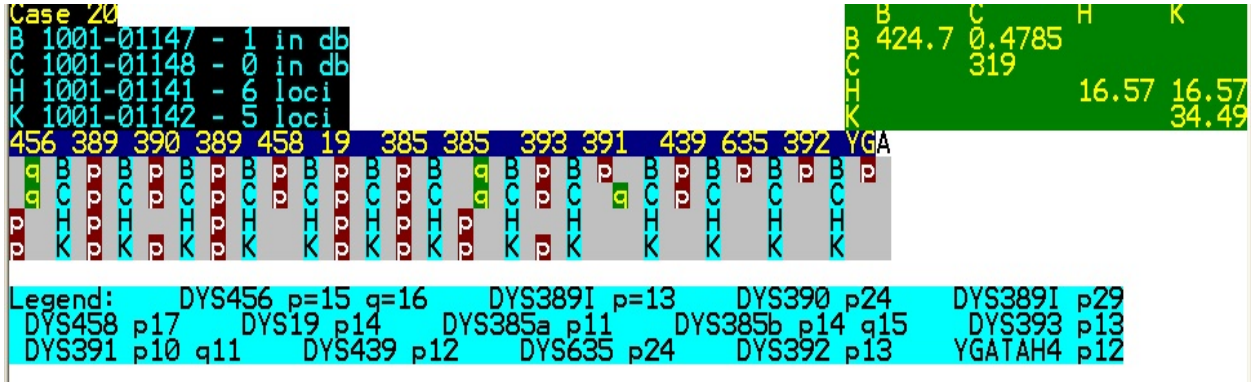



**Figure 47** Y-haplotype matching LR

VI.F.5.a. *Y-haplotype summary*

An inter-person comparison is made for any parties for whom there is Y-haplotype data, and the result is shown in a chart.

The example **Figure 47** illustrates various complexities:

VI.F.5.a.i.) Matching LR to a profile

The diagonal shows the matching LR for a profile with itself. For example, **B** vs. **B** has a matching LR of 424.7 (see §V.H).

VI.F.5.a.ii.) Paternity matching LR (=paternity "index") between two profiles

Suppose **H** and **K** are alleged father and son. These are compatible profiles. The LR supporting paternity is LR = 1/Pr(**K** match **H** | unrelated) = 1/Pr(common part of **H**, **K**), i.e. the LR is computed based on the six loci of **H** (since **H** is a subset of **K**). Of the 1275 comparable profiles in the extended database (adding **H** or **K**, not both), 77 match the common part of **H** and **K** giving a matching LR of 1276/77=16.57.

The matching LR 16.57 is shown in position **HK** of the table. Position **KH**, which could contain the same number, is left blank to avoid clutter.

VI.F.5.a.iii.) Paternity matching LR considering mutation

**C** and **B** match at all but one (DYS391) of the loci that they have in common. The matching LR of 0.4785 between them is an estimated paternity index trying to take mutation at one locus into account.

VI.F.5.b. *Add Y haplotype LR chart to report*

After creating the above table you may add it to the log report (§VI.F.2.a).

VI.F.5.c. Incorporate the Y haplotype likelihood ratio into the Kinship summary

See *Include Y-LR in result*, §VI.F.6.b, §VI.F.6.c.

### VI.F.6. Bayesian options – prior and posterior probabilities

The "natural" answer to a kinship problem is a likelihood ratio. However, if the user is willing to make some stipulations then probability statements can be made.

There are some options pertaining to posterior probabilities.

VI.F.6.a. *Posterior threshold*=99.9%

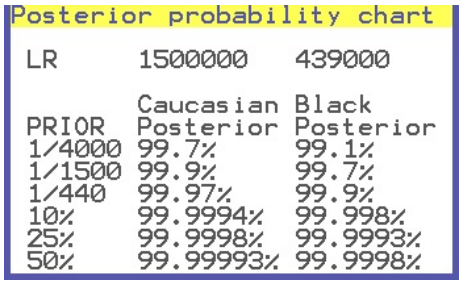
Select this option to change the value (see §XIV.A.5). The value is saved and remembered and utilized by the Chart of posterior probabilities (§VI.F.6.b).

VI.F.6.b. Include Y-LR in result

Same as §VI.F.6.c.

VI.F.6.c. Chart of posterior probabilities

This option becomes available after a kinship LR calculation has been made. It serves these purposes:



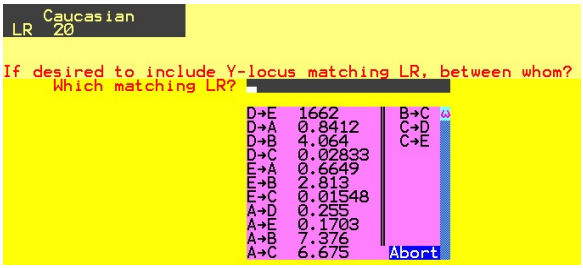

VI.F.6.c.i.) compute the posterior probabilities under various prior probability assumptions

The prior probabilities included in the table include:

» The designated prior (§VI.F.6.d)

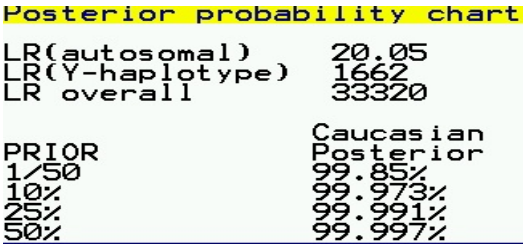
» A few standard values – e.g. 10%, 50%

» If the posterior threshold (§VI.F.6.a) has been specified, the "requisite prior(s)" – the prior probability values which, given the computed LR(s), are minimally sufficient to achieve the posterior threshold.




VI.F.6.c.ii.) allow the incorporation of a Y-haplotype matching LR (discussed in §VI.F.5.a), if any, into the bottom-line LR.

» If there is Y-haplotype data for some of the people in the case, a menu is displayed with the matching LR comparing each pair of men. Choose whichever one (if any) of those LR's is pertinent to the calculated scenario.




VI.F.6.c.iii.) Posterior probability chart including Y evidence

» Append the chart to the printable log report? YES

To view, print, and/or save the report *Show (log) report* (§VI.F.3.b) from the *Immigration/kinship* menu or **File** from the main menu.

VI.F.6.c.iv.) (coming soon) allow the incorporation of a sex-matching factor to be incorporated manually into the bottom-line LR.

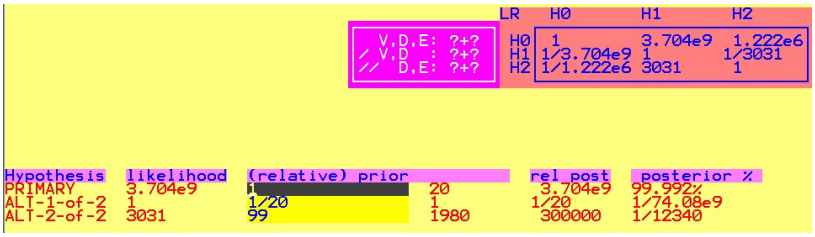
VI.F.6.d. *Prior probability=*

Specify a prior probability $p$ (other than *none* – see §XIV.A.5). Then in the ordinary situation of 2-hypothesis scenarios, posterior probabilities are calculated according to the formula $W=pL/(pL+1-p)$.

» The prior probability value that you specify is preserved across sessions.

VI.F.6.e. Bayes calculator

Bayes calculator is an interactive experimental tool to evaluate posterior probabilities especially when there are more than two hypotheses. See §VI.D.2.e for discussion and §XII.E.1.f for the theory.




Invoke it after doing the calculation (§VI.F.2 or §VI.F.3). A spreadsheet-like screen then appears with the calculated likelihoods copied in, and a column which you can change representing "relative prior." Modify the priors as you wish and the columns to the right adjust automatically. Use the buttons ADD TO REPORT, and CANCEL to quit.

### VI.F.7. Detailed control and analysis of the calculation

VI.F.7.a. *Silent alleles [NOT] allowed*

This toggle specifies whether silent (null) alleles should be considered as a possibility as discussed in §VI.K.2.a. Silent allele frequencies are defined as discussed in §VIII.C.12.e; however at loci for which the database has no null alleles, the user is prompted to supply a null allele frequency.

VI.F.7.b. *Calculate one locus, showing parsing*

This debugging tool gives the analysis shown in **Figure 51**. It replaces the switch *Parsing info: [NOT] shown*, which is deprecated since its output goes by too fast to see.

VI.F.7.b.i.) Operation

The screen displays detailed information about the kinship calculation for that locus, including

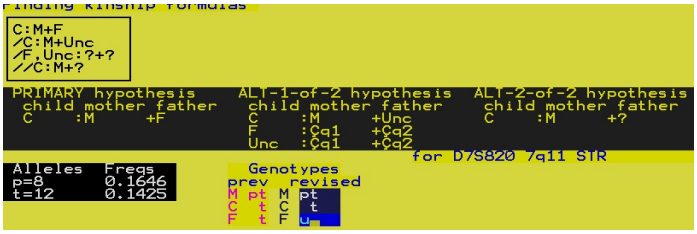



**Figure 52** parsing and kinship for one locus

» Parsing. The "parsing" (blue on white background) shows the child-mother-father trios that the program perceives the scenario to mean, for each of the hypotheses. This is debugging/training information.

» Alleles and probabilities

» Genotypes. The actual genotypes are shown, but you may enter different ones if you wish. Hit ***Esc*** to proceed to the computation.

VI.F.7.b.ii.) Uses

» Check that the program interpreted your scenario as you intended.

» Make trial computations to help make a mutation analysis. For example, when there is an inconsistency somewhere in a presumptive family, sometimes it is possible to reason that the likelihood in view of the inconsistency can be written as an expression involving one or more likelihoods of related problems in which one or another allele is changed (mutated).

VI.F.7.c. *Eyeball check the raw sizes* – checking size data

The *Immigration/Kinship* menu option *Eyeball check the raw sizes* is useful to examine the exact data available relevant to the currently selected case including presence of multiple assays, sizing conflicts,

and even multiple inputs. Upon selection it displays in a "print-preview"-like light-green window a raw size report. Suggestion for navigation:

» Type **work** to move to the first instance of the word "worklist". Notice at the top of the olive strip a notation like 1/3 -- meaning 1st of 3 instances of "worklist".

» NEXT successively to see the others.

» Take a note of the samples assayed, shown on the immediately following lines.

The list is a detail list, showing, by worklist every relevant allele including duplicates and conflicts – subject, however, to the possible effect of the *restrict data* (§VI.F.7.s) option.

VI.F.7.d. *Eyeball check the genotypes*

displays the genotypes for each person in a concise readable format. The display permits SAVE or PRINT as usual.

```
M      Mother  1000-10109   daughter of Fa
U  Sibling#1  1000-10110   brother of M
B  Sibling#3  1000-10111   child of Fa? U?

           M 1221          U 1221        B 1221
           daughter of Fa  brother of M  child of Fa? U?
 D8S1179   9 14            9 14          14
 D21S11    29 30           28 31         28 29
 D7S820    10 14           10 14         10 14
 CSF1PO    12              11 12         11 12
 D3S1358   15 16           14 17         15 16
 TH01      8 9.3           8             8 9.3
 D13S317   11 12           11 12         11 12
 D16S539   11 12           12 14         11 12
 VWA       17 18           14 15         17
 TPOX      8 10            8 11          8 11
 D18S51    15 17           17 20         15 17
 D5S818    11 12           11            11
 FGA       22              22            22

Genotype patterns are:
D8S D21S1  D7S  CSF  D3S13  TH0  D13S  D16S  VWA    TPOX  D18S  D5S  FGA
pu M  qr  M pt M  q M  qr  M pa M pq M pq  M   st M pr  M pr  M pq M p
pu U p  s U pt U pq U p  s U p  U pq U  qs U pq   U p s U  ru U p  U p
 u B pq   B pt B pq B  qr  B pa B pq B pq  B   s  B p s B pr  B p  B p

Legend:    D8S1179 p=9 u=14    D21S11 p=28 q=29 r=30 s=31    D7S820 p10 t14
 CSF1PO p11 q12    D3S1358 p14 q15 r16 s17    TH01 p8 a9.3    D13S317 p11 q12
 D16S539 p11 q12 s14    VWA p14 q15 s17 t18    TPOX p8 r10 s11
 D18S51 p15 r17 u20    D5S818 p11 q12    FGA p22
```

**Figure 53** genotype eyeball report

Be aware, though, that this report is the net genotype picture after resolving any conflicts, and after performing *restrict data* (§VI.F.7.s) if that option is active. To be sure of seeing even conflicting input data pertaining to the case, *Eyeball check the raw sizes* (§VI.F.7.c) with *Do NOT restrict data* (§VI.F.7.s).

VI.F.7.e. *Show summary of calculation*

Show the summary — on the screen, a 1-line-per locus, plus overall likelihood ratio, for the last scenario computed (**Figure 42**).

VI.F.7.f. *Symbolic genotype chart* (revised – 2010)

Creates a useful report documenting all genotypes, probabilities, and letter-allele correspondences for the individuals in the case. The report is displayed on screen; to save or print click the appropriate button.

| locus | M | C | F | p | r | s | t | p | r | s | t |
|---|---|---|---|---|---|---|---|---|---|---|---|
| D3S1358 | pr | pr | s | 15 | 17 | 18 | | 0.248157 | 0.213759 | 0.164619 | |
| VWA | s | pt | pt | 15 | | 18 | 19 | 0.114504 | | 0.223919 | 0.08651 |
| D16S539 | pr | pr | pr | 9 | 11 | | | 0.106173 | 0.274074 | | |
| D8S1179 | pr | pr | rs | 12 | 14 | 15 | | 0.147583 | 0.203562 | 0.111959 | |

VI.F.7.g. *Race is: [Caucasian]*

Select a different race for subsequent computations.

VI.F.7.h. *Next race from:* (e.g. Black/Caucasian/Hispanic)

The races in the "race list" for the case are enumerated. Selecting this option is equivalent to selecting *Race is* and selecting the next race on the list. It is mostly supplanted by §VI.F.2; it was designed for use in conjunction with *Calculate LRs ([specified race])* (§VI.F.3 and §VI.F.3.a).

VI.F.7.i. *Needless loci [NOT] INCLUDED*

A locus is *needless* if the program can recognize that it won't affect the result – for example if only one person was typed at the locus. Space is then saved from the genotype table by omitting such loci.

VI.F.7.j. *Dropout [NOT] allowed [for roles: V ...]*

This experimental option is a method under development to cater to degraded bones. It is not ready to be used!

VI.F.7.k. *[DON'T] SHOW formulas in summary*

A more concise and somewhat less technically-mysterious appearing summary can be produced by omitting the formulas.

VI.F.7.l. *DO [NOT] consider mutation*

When the LR as computed algebraically comes to 0 or to ∞, substitute μ or 1/μ respectively.

The Kinship program is not very clever about mutation. What it does is kind of a poor-man's version of the AABB recommendation for RFLP systems in paternity (§XIII.C.4). In the case of paternity or similar the approach is likely not to be very far wrong, but for cases with two meiosis – missing body of two parents – it figures to be a gross underestimate.

VI.F.7.m. *Let THETA=* $(0 \le \theta < 1)$ *Experimental feature* (new – 2011)

This feature – choosing $\theta>0$ – only makes sense for a body identification scenario. It will then calculated a reduced LR using a modified alternative hypothesis that the body, rather than totally unrelated to the missing person, is related to degree $\theta$.

VI.F.7.n. *Quit from Immigration*

Return to the *... Case* menu. **Esc** does the same job.

VI.F.7.o. *Simulate*

The *Simulate* option gives you a way to predict the likely benefit of typing additional people and/or additional systems. Since it is a major feature, it is described in a major section of its own (§VI.G).

VI.F.7.p. *Scenario from/to pick list* (modified – 2011)

Add the current scenario to the *immigration/kinship* menu. In the future it can then be selected (pick list SELECT button) to be used as a scenario – more convenient, for a complicated scenario, than retyping it.

This option also allows maintenance of the pick list. All pick list items including the newly-added one appears. Move the bounce bar to any desired item and you can EDIT it, DELETE it, MOVE it up or down. Then be sure to save any changes – SAVE & CONTINUE or SAVE & QUIT.

**Note:** For validation test cases, create and save a scenario in the pick list with the correct result for comparison included as a comment.

VI.F.7.q. *Read scenario from file* (new – 2014)

This option makes it possible to create a scenario in a text editor outside of DNA·VIEW then read it in. That would be useful on the rare occasion that the desired scenario is too big to fit in the DNA·VIEW type-in window of §VI.F.1 (for example if you have more than 22 hypotheses). **Note –** don't try to edit such a large scenario in DNA·VIEW or it will be truncated to the size of the type-in window. *Save scenario to file* does the converse but almost always *Save scenario to pick-list* (§VI.F.7.p) is preferable.

VI.F.7.r. *AUTOMATIC/ MANUALLY EDIT allele probabilities* (toggle)

Select this option to toggle between the "automatic" and "manual" states. "Manual" means that the program will pause during evaluation of the likelihood ratio for each locus and give the user a chance to specify the allele probabilities.

VI.F.7.s. *Do/Do NOT/ restrict the data* (toggle)

"Restrict" means to remove some of the DNA data from consideration for calculation. A special interactive screen allows you to remove or select data by person, by locus, or in other ways, as explained in §V.B.26.a.

**VI.F.8.** *Immigration/kinship* **– Kinship validation/consultation**

For some reason you may want to evaluate a kinship problem using specific allele probabilities that are on a piece of paper rather than in DNA·VIEW databases. Two points:

VI.F.8.a. In the kinship menu, turn on the toggle *MANUALLY EDIT allele probabilities* (instead of *AUTOMATIC allele probabilities*) per §VI.F.7.r.

VI.F.8.b. When entering the probabilities you may, within limits, type an expression. For example you may type **34/100**, perhaps useful when solving a paper challenge.

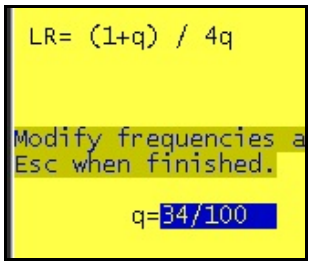



**Figure 54** probability expression

## VI.G. Kinship Simulations

### VI.G.1. Purposes

VI.G.1.a. The *Simulate* option of *Automatic Kinship* gives you a way to predict the likely benefit of typing additional people and/or additional systems. It can be used

VI.G.1.a.i.) for a hypothetical pedigree before any people are typed, or

VI.G.1.a.ii.) in a situation where some typing has already been done.

VI.G.1.b. Construct examples, experiment, test the kinship program (§VI.G.4.c includes an example).

*Simulate* includes a Monte Carlo genotype generator that assigns genotypes in a random, consistent, and unbiased fashion, on the basis of which likelihood ratios can then be computed. Executing multiple simulations then gives you a range of values of the likelihood ratio to expect if the speculated typings are performed.

MENU: BASIC options

| | | |
|---|---|---|
| VI.G.4.a | *Do multiple simulations* | 125 |
| VI.F.1 | *Type in (or edit) scenario* | 111 |
| VI.G.4.b | *Multiplex is (select to change): Prof+* | 125 |
| VI.G.2.e | *Assumed hypothesis is: PRIMARY* | 124 |
| VI.G.4.c | *Freeze the current simulated types & exit simulation* | 126 |
| VI.G.4.e | *Abort simulation (without freezing types)* | 127 |

MENU: SIMULATE includes some of the above and also:

| | | |
|---|---|---|
| VI.G.4.d | *Export the simulated types as an allele table* | 127 |
| VI.G.5.b | *Display/print simulation summary* | 128 |
| VI.G.5.f | *Browse (fetch/clear) (**n** simulations)* | 129 |
| VI.G.5.g | *Retrieve **nn** simulations from 2011/8/1 20:20?* | 129 |
| VI.G.5.h | *(Re)-assign genotypes then pause* | 129 |
| VI.G.5.i | *Likelihood ratio calculate* | 129 |
| VI.G.5.j | *Display last simulated types* | 129 |
| VI.F.3 | *Display details cumulative LR **xyz*** | 115 |

MENU: FILE/FETCH/SHOW includes some of the above and also:

| | | |
|---|---|---|
| VI.G.5.b | *Graph of this simulation (**n** LR data points)* | 128 |
| | *Graph of all simulation LR distributions* | |
| | *Add simulation summary to report* | |
| | *Show all LRs of this simulation (n LR data points)* | |

MENU: OPTIONS includes some of the above and also:

| | | |
|---|---|---|
| VI.F.7.g | *Race is (select to chage):* Caucasian | 122 |
| | *Assumed hypothesis is (select to toggle):* PRIMARY | |
| VI.G.5.n | *Log [NoIBrief\|Complete] simulation details to report* | 130 |
| VI.G.5.o | *Prior probability = [% or (none)]* | 131 |
| VI.G.5.e | *Simulation summary EXcludes / INcludes 0, ∞* | 129 |
| VI.F.7.l | *DO [NOT] consider mutation* | 122 |
| VI.F.7.a | *Silent alleles: [NOT] allowed* | 120 |
| | *Simulation does [NOT] complete partial profiles* | |

**Figure 55** *Kinship Simulation* options and menu categories

### VI.G.2. Outline of simulating

The parameters of a simulation include

VI.G.2.a. scenario – the hypotheses to be *compare*d

VI.G.2.b. the race of reference

VI.G.2.c. a multiplex of loci, or a locus

VI.G.2.d. the number of Monte Carlo simulations to do

VI.G.2.e. whether genotype assignment simulates the primary or the alternative hypothesis

### VI.G.3. Rules for simulation

VI.G.3.a. One-letter names

The people who are simulated are those who have 1-letter names and who are untyped in all loci of the multiplex.

VI.G.3.b. Partial profiles

Real profiles or partial profiles may exist for some roles (=people with 1-letter names) in the specified multiplex. They are used, not discarded. A partial profile may or be filled in by the simulation or not depending on the *Simulation does [NOT] complete a partial profile* switch setting.

VI.G.3.c. Reference databases

Simulation alleles are selected from the default databases for the currently selected race; or, if there is no default, the user is prompted to select a database.

VI.G.3.d. Comment on alternatives

The simulation is based either on the *principal* or the *alternate scenario* (at user option) of the currently specified pedigree.

Suppose the pedigree is a sibling test: `C : Mom+F ♦ D : Mom + F/?`. Simulation according to the principle scenario means that simulated genotypes are selected that are typical of full siblings `C` and `D`, and the likelihood ratio will therefore tend to favor full sibling-ship (LR>1). If you instead elect to simulate the alternative scenario, then genotypes typical of half siblings `C` and `D` are chosen and the likelihood ratios will generally be <1.

### VI.G.4. Simulation Operation

The simulation options mentioned below are selected from the simulation menu, and the setting for any particular options is generally documented in the menu. In addition, any non-standard settings, such as *prior*>0 (§VI.G.5.o) or *assume ALTERNATE hypothesis* (§VI.G.5.l) are shown prominently outside of the menu.

VI.G.4.a. *Do multiple simulations*

This option asks you to specify a number of simulations to perform, then proceeds with them.

VI.G.4.a.i.) Number of simulations

How many simulations are necessary? To get an accurate idea of the distribution of likelihood ratios, often the answer is 100 or more. The individual LRs tend to vary over a large range, which means that even the mean of the distribution cannot be accurately estimated from just a few observations (simulations). On the other hand, even a few simulations are much more information than none at all.

The prompt that asks for the number of simulations desired is phrased in terms of the total number (not the additional number).

```
Simulations already done of the primary hypothesis: 20
How many total simulations would you like? 30
```

» As the simulations are done, progress and cumulative results are kept visible on the screen. If you become impatient and wish to stop simulating before the requested number have completed you can freely interrupt the program; even if you close down DNA·VIEW the results so far computed will be retained and can be displayed, printed, or added to.

Simulation is slow – often seconds per simulation. Try a few simulations first to get a timing, then specify the number of additional simulations that you both want and have the patience for. The statistical summary of simulations is cumulative – if you do 2 simulations then 98 more, the summary includes all 100. In fact, the summary record is only reset when you explicitly clear it (§VI.G.5.f). If you simulate several different situations, separate summaries are maintained and they are saved on disk across DNA·VIEW sessions.

VI.G.4.b. *Multiplex is:* (multiplex or locus)

Specify the collection of loci for which the simulation is performed.

A set of loci can be selected from the *multiplex list*. That list isn't user-extensible, but still you may select any desired combination of loci on the fly, per §IX.B.7.

VI.G.4.b.i.) Arbitrary multiplex

```
Multiplex is (select to change): D3S1358 FGA VWA
```

The ability to select any arbitrary set of loci for simulation (§IX.B.7) is convenient for a couple of reasons:

» evaluate the power of any single locus

» simulate the effect of for example adding Cofiler (not available by itself in the multiplex list), without necessarily completing incomplete Profiler+ profiles (consider §VI.G.5.q).

VI.G.4.c. *Freeze the current simulated types & exit simulation*

Exit upward one menu level back to *Immigration/kinship*, and retain the last set of simulated genotypes for further analysis. This option gives a way to experiment with complicated what-if questions. The frozen set of simulated types can be used as the data for several different *Immigration/kinship* calculations. If *Simulation* is then re-invoked, the frozen types are regarded as known types for the purpose of further simulation.

VI.G.4.c.i.) Example – Incest experiment.

Consider the incest problem illustrated in **Figure 56** – siblings **M** and **B**, and the child possibly of incest **C** are typed – and suppose you are not sure whether it is necessary to include the sentence **M,B:Gma+Gpa** in formulating the hypotheses.

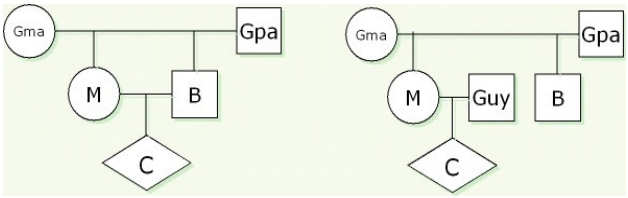


**Figure 56** Incest pedigree

Assume you have created case 14 as an empty case per §IV.D.2. Then starting from the main command menu:

| | |
|---|---|
| **C** *enter* **au** *enter enter* **14** *enter* | select case 14 |
| **im** *enter* **simu** *enter* | *Simulate* menu |
| *enter* | acknowledge that there is no data |
| *enter* | *Type in .. scenario* |
| **C:M+B** | For the moment, ignore the incest component. |
| **/ C:M+Guy** | Alternative hypothesis. |
| *Esc* | back to *Simulate* menu. |
| **geno** *enter* | *Assign genotypes then pause* |
| **fre** *enter* | *Freeze the current simulated types & exit simulation* |
| *Calculate LRs (Caucasian)* | Note the LR result. |
| **T** *enter* | *Type in .. scenario* |
| *Left-click* after end of 1st line | position the type-in cursor mark (§XIV.A.4.a.vii) |
| **% M,B:Gma+Gpa** | Alter primary hypothesis to include sibling relationship. |
| *Right-click* after end of 1st line | Capture (copy) the new text. |
| *Left-click* end of 2nd line, *ctrl-v* | Paste new text into alternative hypothesis also. |

```
      ESC after entering pedigrees.
    C:M+B  % M,B:Gma+Gpa
/   C:M+Guy % M,B:Gma+Gpa
```
**Figure 57** Paste saves typing

| | |
|---|---|
| *Esc* | back to *Immigration/kinship* menu. |
| *Calculate LRs (Caucasian)* | Compare the LR result with the previous one. Should be identical. |

VI.G.4.d. *Export the simulated types as an allele table*

This simulation option provides a convenient way to construct sample cases with data that is realistic for a specified situation. The export is the same as *Export case*, §V.B.14.

VI.G.4.e. *Abort simulation (without freezing types)*

Exit from *Kinship simulation* back to *Immigration/kinship*. Any simulation summaries you have developed can be extended by re-invoking *Simulation* and doing more of the same simulation (§VI.G.2).

### VI.G.5. Simulation Summary

The result of simulating is a report such as that illustrated in **Figure 57**. This data can be exported and graphed (§VI.G.5.b, §VI.G.5.d).

The illustrated simulation assumes that A and B share a mother and evaluates the chance to show that a third

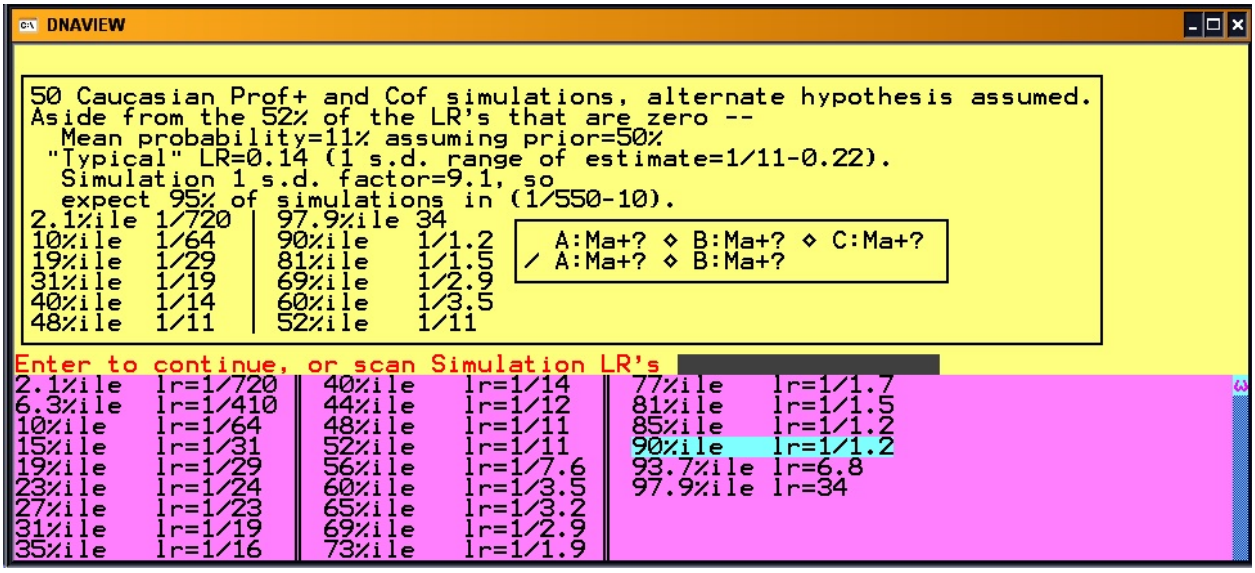



**Figure 58** Simulation summary

child C is not a half-sibling as well, when only the three children are typed. The simulation assumes that A and B share a mother and evaluates the chance to show that a third child C is not a half-sibling as well, when only the three children are typed.

From the example of **Figure 57**,

VI.G.5.a.i.) Note that "alternate hypothesis assumed" – genotype assignment modeled an *un*related third child C. In 52% of the 50 simulations, the third child was "excluded" (based on the no-mutation model) from half-sibship in some system.

VI.G.5.a.ii.) Of the remaining simulations, the "typical" LR – the geometric mean (i.e. the antilog of the mean log) LR, was 0.14. That is, the typical LR (correctly) supporting *non*-relationship of C was 7. This tells us that even among those 48% of cases with genotypes nominally consistent with three half-siblings, that consistent pattern is typically improbable – such for example as homozygosity for a rare allele.

VI.G.5.a.iii.) Due to the limited sample size (N=50), the estimate of typical LR has some uncertainty. Its value is probably in the range 1/11–0.22, and this range can be made as small as desired by continued simulations.

VI.G.5.a.iv.) The distribution of non-zero LRs is a rather broad distribution. If the number of markers is large, it should be approximately log-normal. N standard deviations of the distribution are approximately encompassed by the range[ $0.14/9.1^N$, $0.14 \cdot 9.1^N$].

VI.G.5.a.v.) The top part of the screen includes a summary of the distribution of the non-zero LR values at approximately each decile boundary. LR=1/11 is about the mode. The full list of non-zero LR's is shown at the bottom.

VI.G.5.b. *Display simulation summary*

The distribution described above is displayed after each simulation, and can also be recalled on demand by this option.

VI.G.5.c. *Graph of this simulation (**nn** LR data points)*

Create and save a file (dialogue per §V.K.2 *Save as*) with full details of the current simulation. The file includes

VI.G.5.c.i.) problem definition, parameters, and summary statistics

VI.G.5.c.ii.) **LR deciles** of the simulation distribution

VI.G.5.c.iii.) **all LR's** – every simulated LR

VI.G.5.c.iv.) **Graphable distribution** a chart with sample distribution points which, with the help of Excel, can instantly be turned into a charming and beautiful graph of the expected LR distribution.

» **Create the graph** Open the saved file in Excel. Select the **LR range** (X-axis) column and its neighbor(s) (Y-axis), including the titles. ***Click* chart**" (i.e. from the **Insert** menu), **XY(Scatter)**, choose **Scatter with data points connected by lines without markers**, **finish**.

**Logarithmic scale**. ***Right click*** on the X-axis, ***click* Format axis**, choose the **Scale** tab, checkmark **Logarithmic scale**. Now the graph looks ok.

» **Improve appearance** – ***right click*** and **clear** the Y-axis. ***Right click*** on the plot area, choose **Format plot area**, and under **Area** choose **None**.

**Fix labels**. ***Right click*** in the plot area and choose **Source data**. Select the **Series** tab. Type a descriptive name (e.g. "paternal incest") in the Name box. **Ok**.

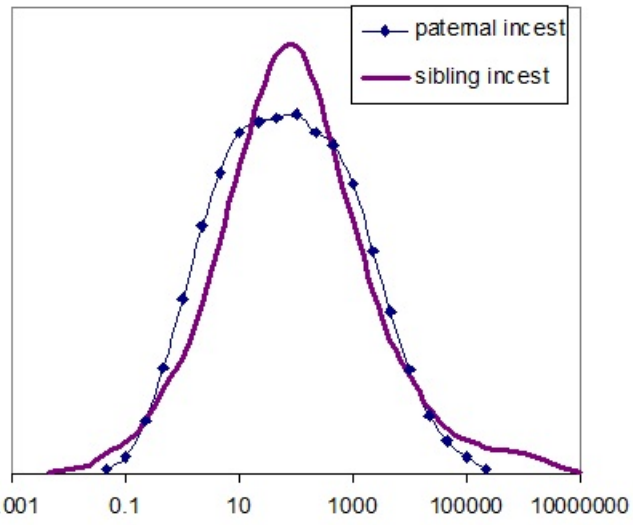



**Figure 59** Incest LR distribution; Identifiler test of mother and child

VI.G.5.d. *Graph of all simulation LR distributions*

Create and save a file (dialogue per §V.K.2 *Save as*) with data for making superimposed graphs of all simulations. Make the graph in Excel as described in §VI.G.5.c.iv.

» Labeling the graphs – The label for each simulation curve comes from the comment or the first comment in each scenario. Therefore it is a good idea to include a comment (§VI.C.2.c) as part of each scenario, viz: `V,C:M+? ; sib+1 parent`

VI.G.5.e. *Simulation summary EXCLUDES / INCLUDES O, ∞*. (toggle) Recommend: EXCLUDE

In most cases the EXcluding summary is probably more interesting. It means that the extreme values (which correspond to a nominal inconsistency for one hypothesis or the other) are counted and recorded, but not included in averages.

Suppose for example the paternity problem C:M+F/?, with mutation (§XIII.C) ignored (§VI.F.7.l) and assuming the `ALTERNATIVE` hypothesis (§VI.G.5.l). Then the vast majority of the likelihood ratios will be 0 so if they are INcluded in the summary then the typical PI (and the typical posterior probability if requested – §VI.G.5.o) would be 0 (and nearly 0), which is not helpful. Using the EXclude setting, averages of just "non-excluded" men are shown, which might be interesting.

VI.G.5.f. *Browse (fetch/clear) (**n** simulations)*

The *browse* list is summaries of the simulations you have done. You can selectively delete, or fetch one of them for further simulations, reporting, or graphing (§VI.G.5.b).

Note that if you close and re-open DNA·VIEW the *browse* list doesn't exist until you first *retrieve* (§VI.G.5.g).

VI.G.5.g. *Retrieve **nn** simulations from **date time***?

This option allows you to *retrieve* all the simulations – which can be for various scenarios – which were last saved on disk on the specified *date, time*. (The saving is automatic and continuous.) Note that if you re-start DNA·VIEW and run a new simulation *without retriev*ing, the old simulations are lost. (To avoid accidental loss, the simulation program always proposes *retrieve* when you first invoke the simulation program if there are saved simulations which would otherwise be lost.) If you do *retrieve*, subsequent new simulations are added to the old collection.

See also §VI.G.5.f.

VI.G.5.h. *(Re)-assign genotypes then pause*

Make a Monte-Carlo assignment of genotypes and display them (like **Figure 40**), without calculating the LR.

VI.G.5.i. *LR calculate*

Calculate the LR for the Monte-Carlo assignment. The combination of *(Re)-assign genotypes then pause* and this option is equivalent to doing one simulation (§VI.G.4.a).

VI.G.5.j. *Display last simulated types*

Show the genotype table (like **Figure 40**) corresponding to the most recent Monte-Carlo assignment.

VI.G.5.k. *Race is (select to change)*: `Caucasian`

Same functionality as changing the race in the next higher menu, the *Immigration/Kinship* menu.

VI.G.5.l. *Assumed hypothesis is (select to toggle)*: `PRIMARY`

Selecting this option toggles between assuming the `PRIMARY` or the `ALTERNATIVE` for generating the simulated genotypes.

Example: Suppose the simulation is a sibling question: `C, D/? : Mom+Fa`. Under the `PRIMARY` hypothesis, genotypes are generated for `C` and `D` as if they are full siblings, so the computed likelihood

ratios will mostly be greater than 1. Under the ALTERNATIVE hypothesis, genotypes are generated for C and D as if they are unrelated. But the likelihood ratio is defined in the same way as before, namely the likelihood ratio favoring siblingship. So the computed likelihood ratios will mostly be less than 1.

VI.G.5.m. *DO / Do NOT consider mutation* (toggle)

Same functionality as in the next higher menu, the *Immigration/Kinship* menu (§VI.F.7.l).

VI.G.5.n. *Log* NO/BRIEF/COMPLETE *simulation details to report* (selecting cycles among choices)

Choose how much detail is added to the "log" report (the report that goes into DNA·VIEW's report buffer, and is printed or viewed from the **File** menu).

```
   Beginning 5 simulations, PRIMARY hypotheses

 C : M+F/?
```

**Figure 60** Simulation record, NO details

```
   Beginning 2 simulations, PRIMARY hypotheses

 C : M+F/?

Caucasian cumulative LR   21.7
          Posterior probability=95.6% assuming prior=50%
Caucasian cumulative LR   39.6
          Posterior probability=97.5% assuming prior=50%

-------------------------------------------------------------

   Beginning 2 simulations, ALTERNATE hypotheses
 C : M+F/?
Caucasian cumulative LR   1/41700
Caucasian cumulative LR   1/20.2
```

**Figure 61** Simulation record, BRIEF details (2 examples)

VI.G.5.o. *Prior probability ...*

If the prior probability (§VI.F.6.d) is set (i.e. prior>0), then a posterior probability is calculated for each simulation and the average posterior is reported as a summary statistic.

For example, **Figure 57** shows a simulation of the "missing body" problem: the unrecognizable body X is either the child of J+K, or unrelated. The mean probability is 98%. The typical PI of 3100, with a prior of 1/20, corresponds to a posterior probability of 99.4%. Which is the more meaningful number?

At least in a case like this I would say the 98% has the more obvious interpretation. Suppose the context is a disaster with 20 children to identify, each with reference to the parents. Then prior to DNA analysis the probability is 1/20 for any particular body to belong to any particular set of parents. If an identity is then assigned according to a LR's whose expected corresponding posterior probability is 98%, then each such assignment means on expectation 0.02 misidentifications. That's a useful fact to consider when planning a policy for announcing identifications. If 20 identifications will be made under such a policy, then there will be about a 20·0.02=40% chance to get one wrong.[13]

VI.G.5.o.i.) Changing the prior

```
   Beginning 1 simulations, PRIMARY hypotheses

  C : M+F/?
2004/11/20 16:06

Simulated genotype patterns are:
D3S  VWA    FGA
p r M  r v M ps
pq  F p s  F p t
p   C  rs  C ps

 Legend: D3S1358 p=15 q=16 r=17  VWA p=14 r=16 s=17 v=20  FGA p22 s25 t26
Caucasian cumulative LR   5.13
Caucasian
D3S1358 3p STR  1.78     1 / 2p          p=0.281
VWA 12p13.3 STR 1.64     1 / 2s          s=0.306
FGA 4q STR      1.76     1 / (2p+2s)     p=0.203 s=0.0804
cumulative LR   5.13
Time for last simulation:  1.8sec
```

**Figure 62** Simulation record, COMPLETE details

Same effect as setting it in the next higher menu, the *Immigration/Kinship* menu (§VI.F.6.d).

Note that if you change the prior you may then immediately *Show simulation summary* under the new prior assumption; there is no need to re-simulate.

VI.G.5.p. *Silent alleles ALLOWED/NOT ALLOWED* (toggle)

Same functionality as in the next higher menu, the *Immigration/Kinship* menu.

VI.G.5.q. *Simulation DOES / does NOT complete partial profiles* (toggle)

Suppose `C` and `D` were typed already with some kit and a sibling calculation performed, but results were not obtained for all loci. If further testing will mean just testing the mother `M` of one of them, then the

[13] approximately. An exact computation is problematic.

appropriate simulation assumption would be to NOT complete the partial profiles. However, if it is proposed to re-test C and D (whether or not M will be simulated), then choose the simulation option DOES complete.

### VI.G.6. Saving simulation scenarios

The history of simulations performed are saved during a session (and NOT LOST when you exit from DNA·VIEW). Therefore it is possible to do 20 simulations for some scenario, then try a different scenario, then later return to the first scenario and do 10 further simulations for a total of 30. In order to make it convenient to return to an earlier scenario the scenarios are included in the "pick list", which is at the end of the *Immigration/kinship* menu.

## VI.H. The *Prototype Kinship* command (stand-alone Kinship program)

*Prototype Kinship* is the original (1993) kinship program. It is occasionally useful for experiments, such as to find the formula for a particular genotype pattern. It is stand-alone; it does not directly interact with DNA·VIEW databases or with people that have been defined as part of a *Case* or included in a worklist roster. Therefore for casework it is always easier, faster, and more convenient to use the *Automatic Kinship* version.

*Prototype Kinship* analyzes systems consisting of a collection of alleles *a*, *b*, ..., *n*, *p* , ..., *z* that exhibit co-dominance plus possibly one silent allele, which is always designated *o*. Or, it can analyze X-linked systems.

The result of *Prototype Kinship*'s calculation is an algebraic formula for the likelihood ratio of the relative probabilities of the two pedigrees. If you supply the probabilities, it will then evaluate the formula.

## VI.I. Kinship cases

*Prototype Kinship* thus computes one locus at a time. The necessary information for such an analysis is definitions of the pedigree alternatives, genotype/phenotype information for some or all of the people, and perhaps probability values for the various genes. For lack of a natural yet descriptive name for this collection of data, let's call it a *pedigree/locus*.

> *pedigree/locus* means the definition for a single locus of a kinship problem, and perhaps additional associated information including allele probabilities, and the likelihood ratio formula and value.

Part of this data is supplied by the user; part is determined by the program's computations. The user supplied part is the script, including symbolic genotype specifications, as describe in §VI.C.

*Prototype Kinship* also includes some filing and reporting features that enable you to combine computations across several loci into a unit which is unimaginatively called a *complete case*

> A *complete case* consists of a collection of pedigree/loci, one for each locus.

Complete cases can be filed and fetched from the a special kinship library in DNA·VIEW and/or PATER with the

> Kinship menu option: *File or Fetch a complete case*.

A brief locus-by-locus summary of the current complete case can be displayed with the

> Kinship menu option: *Display Case Summary*

Individual pedigree/loci (i.e. for one locus) can be retrieved and re-posted to the current complete case using the menu options *Rework, reorder, or delete a locus* and *Post this locus to the complete case*.

The currently active pedigree/locus can be displayed, edited, or calculated via *Display pedigree/locus*, *Edit pedigree/locus*, *Find formula*.

Once a formula exists, it is evaluated with *Evaluate formula.*

Several options are available to construct a report: *Clear the report, Insert case summary in the report, Insert pedigree/locus in the report, Print report (=Print)*.

## VI.J. Overview of using *Prototype Kinship*

### VI.J.1. a new problem

To work through a completely fresh problem, the typical order of operations is:

VI.J.1.a. preparation

Lay out the genotypes for each locus and assign letters in a consistent way to each allele.

VI.J.1.b. *Clear complete case*

VI.J.1.c. *Type in a new pedigree/locus*. Recommendation: First, make comment lines with case and locus. Second, define the genetic types, one line per person, as separate lines. Third, define the relationships. This format will be easy to modify from locus to locus. It is illustrated in §VI.K.11.

VI.J.1.d. *Find formula for locus*

VI.J.1.e. *Evaluate formula*. (Supply relevant allele probabilities as requested.)

VI.J.1.f. *Post this locus to the complete case*

VI.J.1.f.i.) *Post to what row?* Select *Locus* or *Serological test* or *arbitrary* as desired.

VI.J.1.f.ii.) Choose a name and the pedigree/locus is posted under that name within the complete case.

VI.J.1.g. *File or fetch a complete case*. For safety, file the case so far to disk.

VI.J.1.h. *Edit pedigree/locus*. Modify for the new locus — change the comment & genetic type lines.

Continue from *Find formula* until all loci are defined. Then prepare a report (§VI.K.13).

### VI.J.2. a modified problem

Quite often a kinship problem is a modification — different scenarios for example — of a problem already worked and filed. The steps in that case are:

VI.J.2.a. preparation (per §VI.J.1.a)

VI.J.2.b. *File or fetch a complete case*. Retrieve the model that will be modified.

VI.J.2.c. *Rework, reorder, or delete a locus*

VI.J.2.c.i.) Clear all the old likelihood ratios by moving the red bar to the first locus, then hitting **0** once per line.

VI.J.2.c.ii.) Select the [next] locus to rework.

VI.J.2.d. *Edit pedigree/locus*. Change the definition as necessary. *Find formula, Evaluate, Post, File* (using a new name) as per the steps in §VI.J.1 above.

Continue from *Rework* until all loci are defined. Note that the red bounce bars pretty well anticipate what you want to do — the *Rework...locus* menu automatically advances one locus at a time, the *Post ... locus* menu offers to return the work to the same locus slot from which it came, and the **Kinship** menu automatically cycles through the above steps.

When all loci are done, be sure to *File* (probably to a new complete case name).

Then prepare a report (§VI.K.13).

## VI.K. Using the *Prototype Kinship* Command

### VI.K.1. Define the relationships

To make a kinship analysis for a single locus, it is necessary to create block of text that defines the relationships and lists the genotype patterns. This most obvious method is to use the menu option

*Type in a new pedigree/locus*

although there are various ways to copy and modify previous work that are often convenient in practice.

The text so typed in we call a *Kinship file*. Kinship files can be saved and retrieved, through the **Kinship** menu options

*Read pedigree/locus from a *.KIN file*
*Save pedigree/locus as a *.KIN file*

although these old-fashioned facilities are not the most convenient method.

Create a kinship file following the rules explained in §VI.C. For example, the pedigree diagram **Figure 64** represents a deficiency paternity question. To do the first locus choose *Type in a new pedigree/locus*. I like to format the text in a way that will be easy to modify for the subsequent locus:

| | |
|---|---|
| VI.K.1 | *Type in a new pedigree/locus* |
| VIII.F, VIII.F.1, VIII.F.3, VIII.N, IX.B.3, XIV.A.4.a.viii | *Read pedigree/locus from *.KIN file* |
| VIII.F, VIII.F.1, VIII.F.3, VIII.N, IX.B.3, XIV.A.4.a.viii | *Save pedigree/locus as a *.KIN file* |
| VI.J.2.d | *Edit pedigree/locus* |
| VI.K.3 | *Find formula for locus* |
| VI.K.4 | *Algebraically simplify formula* |
| VI.K.5 | *Evaluate formula* |
| VI.K.6 | *Display pedigree/locus* |
| VI.K.13.b.iv | *Insert pedigree/locus in report* |
| VI.J.2.c | *Rework, reorder, or delete a locus* |
| VI.K.7 | *Post this locus to the complete case* |
| VI.K.8 | *Display case summary* |
| | *Clear complete case* |
| VI.K.13.b.i | *Clear the report* |
| VI.K.13.b.ii | *Insert case summary in the report* |
| VI.K.13.a | *Print the case summary* |
| VI.K.10 | *File or fetch complete case* |
| VI.K.13 | *Print report (=Print)* |
| | *quit* |
| VI.K.2.a | *Silent alleles: not allowed* |
| VI.K.2.b | *Locus is: autosomal* |

**Figure 63 Kinship** options

```
; Example reconstruction case
; locus D3S1358
Child pr
Mom pq
Auntie pr
Granny pr
Child : Mom + Allegedfather / ?
Allegedfather, Auntie : Granny + Gramps
```

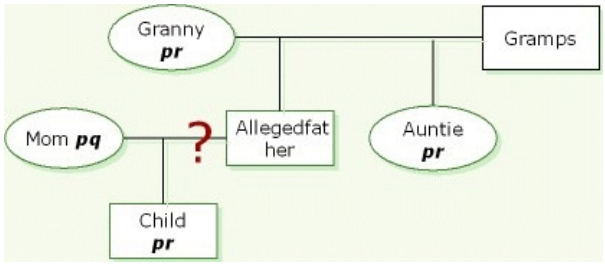



**Figure 64** Deficiency case pedigree

Type ***esc*** when finished with entry using *Type in a new pedigree/locus* or modifications (using *Edit pedigree/locus*).

The result thus created is not permanently saved to disk by the ***esc*** action, but only held temporarily for analysis.

### VI.K.2. Set option toggles

Several of the menu items are toggles that control the way the analysis is performed.

VI.K.2.a. *Silent alleles: allowed / not allowed*

Selecting this option toggles between the "allowed" and "not allowed" phrase. The distinction is in the way that one letter phenotypes, for example **Child p** are interpreted. If silent alleles are not allowed, then **p** simply means **pp**. But if they are allowed, then the analysis will take into the account the existence of a silent allele that is always called **o**, so that a person who types as **p** may be either **pp** or **po**.

For an example of the use of this option, see §VI.L.3.

If the ***o*** allele is specifically mentioned in the scenario definitions, then the analysis will automatically allow silent alleles.

VI.K.2.b. *Locus is: autosomal / x-linked*

Selecting this option toggles between the "autosomal" and "x-linked" phrase. Using this option male phenotypes should be written as for example **p–**, where the **–** is a pseudo-gene representing the missing position on the Y-chromosome.

*Example:* **Child pr : Mother pq + Man r–**. Likelihood ratio = 1/*r* for an x-linked trait. Whereas if the locus were autosomal and the man were **rs**, the likelihood ration would be 1/2*r*.

VI.K.2.c. *Parsing & hints are: shown /not shown*

If the *shown* option is elected, then extra, debugging, information about the analysis is shown when *find formula* is selected. The extra information is useful because, regardless of what you intended and of any errors in the documentation or of the program's ability to understand the problem correctly, it shows what problem the program actually solved.

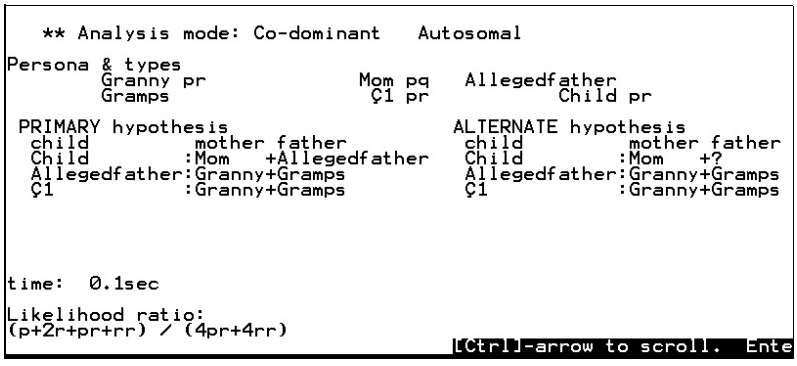



**Figure 65** Parsing output

**Figure 65** shows an example of the parsing result based on the problem definition

```
Child pr : Mom pq + Allegedfather / ?
Allegedfather, pr : Granny pr + Gramps      ;(the input)
```

Notice the symbol Ç1. It is a computer-generated name for the putative aunt who was denoted **?** in the input.

The parsing toggle also turns on these additional interesting effects noted below under *find formula* (§VI.K.3) and *evaluate formula* (§VI.K.5).

### VI.K.3. *Find formula for locus*

Choose this option to begin the analysis. The algorithm is explained in Brenner (1997b).

The result of the analysis is a likelihood ratio written in symbolic form:

`Likelihood ratio:` $(p+2r+pr+rr)/(4pr+4rr)$.

Symbols *p, r* etc. are used to denote gene probabilities corresponding to the symbols `p, r` etc. that designate genes on the input.

VI.K.3.a. Parsing toggle

An odometer-like counter at the bottom of the screen indicates progress as the program derives the formula for the scenarios that you have defined. If the *parsing* toggle (§VI.K.2.c) is turned on, the display is enhanced and also includes estimated time to finish.

VI.K.3.b. Interrupt

It is easily possible to define a kinship problem that will run for minutes (not to mention weeks). If you discover that you have done so accidentally, you may wish to cancel the computation, but without losing your temporary work which ***ctrl-break*** would seem to risk. To cater to this situation, ***ctrl-break*** has a special interpretation during *find formula* only. It produces the dialogue box:

```
Really interrupt (y)? Or continue (n)?
```

Typing **y** will abort the present computation, but does not quit from **Kinship** so the present pedigree/locus is not lost. Typing ***ctrl-break*** a second time will kill **Kinship** and return to the main menu. Typing **n** of course cancels the interruption and computation resumes.

### VI.K.4. *Algebraically simplify formula* (for amusement only)

The option *Algebraic simplify* in the Kinship sub-menu is a purely cosmetic operation that attempts to simplify the algebraic expression for the likelihood ratio. For the example, suppose the likelihood ratio is

$$(a+b+2ab) / (a+b)$$

*Algebraic simplify* will rewrite it as:

$$1 + 2ab/(a+b).$$

In some cases you can effect repeated simplifications — or at least rewritings of the formula.

This option is experimental and may not always work perfectly. However, the *evaluate formula* option always evaluates the *original* formula, so does not depend on whether *Algebraically simplify formula* is correct.

### VI.K.5. *Evaluate formula*

Select the *Evaluate formula* option in order to evaluate the algebraic expression with numeric gene probabilities (§VIII.C.13).

*Evaluate formula* creates a form on the screen in which you can type allele probabilities, and the likelihood ratio formula is evaluated as you do so. The value is dynamically displayed on the screen. After you have specified all allele frequencies to your satisfaction, ***esc*** or click DONE.

For the example above and locus D3S1358, the result is

```
Using p=0.113 r=0.223     value = 2.12
```

If *Parsing & hints* (§VI.K.2.c) is turned on, then the partial derivatives ∂L/∂v will be displayed showing how the likelihood ratio L varies with variation in each variable v. The reason for this hint is to give warning (by a positive partial derivative) of those special occasions where a generously large probability value is *anti*-conservative in the sense of exaggerating the strength of the evidence favoring the principal scenario.

**VI.K.6.** *Display pedigree/locus*

— can be used at any time to display all information and calculations for the current locus, such as illustrated in **Figure 65**.

**VI.K.7. Save the pedigree/locus in a complete case**

The pedigree/locus can be stashed to the currently active complete case through the command *Post this locus to the complete case*. The posting menu includes the options

VI.K.7.a. *Locus (I want to select a new one)*

Select this option in order to choose a name, from the DNA·VIEW probe/locus list, under which the pedigree/locus information is to be saved within the complete case.

VI.K.7.b. *Serological test (I will select from PATER list)*

Select this option in order to choose a name, from PATER's list of systems, under which the pedigree/locus information is to be saved within the complete case.

VI.K.7.c. *Arbitrary name (I will type it in)*

This option allows typing in a name of your choosing under which to file the present pedigree/locus.

VI.K.7.d. Extant name

*Post this locus* also presents a menu choice for each of the loci that already exist in the current complete case. Hence you can use this choice as part of the operation of retrieving a pedigree/locus (see *rework*, §VI.J.2.c), modifying it, then putting it back.

VI.K.7.e. no option (***backspace enter***)

has the effect of escaping from the *Post this locus to the complete case* submenu without doing anything.

**VI.K.8.** *Display case summary*

— can be used at any time to show a line per locus and overall likelihood ratio for the all loci so far posted to the complete case, as illustrated in **Figure 65**.

**VI.K.9.** *Run test cases; check program*

This option computes the formulas for a collection of test cases, and compares the answers with the correct formulas. It is a test that the algebraic formula derivation part of the program has not changed since earlier versions.

**VI.K.10.** *File or fetch complete case*

It is not necessary to file the complete case after each locus, or even to file it at all, but it is certainly safest. The *file or fetch* menu is the kinship case library directory. It also contains one option labeled

VI.K.10.a. *(a new case)*

to create a new complete case entry in the kinship case library.

VI.K.10.a.i.) The programs asks for the name. You may type any name including spaces etc up to about 50 characters. This will be the name that appears in the kinship case library directory.

After typing the name, either ***enter*** or ***tab*** and the complete case will be added to the directory and saved.

Use of the ***tab*** choice will insert the cumulative likelihood ratio at the end of the name. Thus it will be visible when you view the library directory.

VI.K.10.b. select a case name

After moving the red selection bar to the desired case, there are three keys normally used to confirm the selection:

VI.K.10.b.i.) ***PgUp*** to confirm the selection to *fetch* the selected case. It will thus become the new "current case" and supplant the one you have been working on.

VI.K.10.b.ii.) ***PgDown*** to confirm the selection to *file* the current case under the selected name. After selecting this choice, you then may rename the case (or re-insert the new likelihood ratio) per §VI.K.10.a.i above.

VI.K.10.b.iii.) ***Enter*** is ok either to file or to fetch. The program will then ask which you want, and proceed per one of the previous two choices.

Once the case is filed it resides on the hard drive and is safe even if you unplug the computer.

### VI.K.11. Calculate additional loci (if any)

Often it will be convenient to model the new locus on the previous one. For example, to continue with the example data of **Figure 64** to do the second locus, VWA, choose *Edit pedigree/locus* and modify the kinship file to read:

```
; Example reconstruction case
; locus VWA
Child pr
Mom pq
Auntie qr
Granny rs
Child : Mom + Allegedfather / ?
Allegedfather, Auntie : Granny + Gramps
```

Then proceed with step §VI.K.3 etc. The formula for this pedigree/locus will be given as:

```
Likelihood ratio:   (1+r) / (4r)
```

and the result of step §VI.K.5 is

```
Using  q=0.258     value = 1.22
```

### VI.K.12. Revising a complete case

Sometimes a new complete case is based on an old one. For example, it might be desired to pose a modified scenario problem — compare the same primary scenario with a different alternative scenario. In such an event the list of loci, the genotype specifications, and the allele probabilities from the former case may be all or mostly valid for the new problem, and it will be most convenient to model the new case on the old one, locus by locus.

The routine described in §VI.J.2 works well in such a case.

**VI.K.13.** *Print report*

The computations are displayed on the screen as you work. They are also appended to the report buffer, which you can print with this menu option or view/save/print from the **File** menu. However, this "log" report is probably not so useful because it will also include a record of mis-steps along the way. To put together a clean report, follow one of these procedures:

VI.K.13.a. *Print the case summary*

in many cases is quite sufficient as a record of the computations and the result. It will include a one-line-

```
Caucasian
--
D3S1358 3p PCR      2.12     (p+2r+pr+rr) / (4pr+4rr)     p=0.113 r=0.223
VWA 12p13.3 PCR     1.22     (1+r) / 4r                   r=0.258
FGA 4q PCR          0.25     1 / 4
TH01 11p15.5 PCR    1.81     (1+p) / 4p                   p=0.16
TPOX 2p25-p24 PCR   1.54     (3+p) / 4p                   p=0.583
CSF1PO 5q33-34 PCR  1.05     (2+p+r) / (4p+4r)            p=0.268 r=0.358
D5S818 PCR          1.34     (2p+q+pp+pq) / (4pp+4pq)     p=0.393 q=0.165
D13S317 PCR         1.56     (q+2r+qr+rr) / (4qr+4rr)     q=0.313 r=0.283
D7S820 7q11 PCR     1.69     (1+p) / 4p                   p=0.174
cumulative LR       6.64
```

**Figure 66** Kinship case summary

per-locus summary of the case including the overall likelihood ratio.

VI.K.13.b. Make a report with full detail

VI.K.13.b.i.) *Clear the report* — empties the report log

VI.K.13.b.ii.) *Insert case summary in the report* — puts the case summary, e.g. §VI.K.13.a as the top of the report-in-progress.

VI.K.13.b.iii.) *Rework, reorder, or delete a locus* — and from the *rework* menu, select the first or next locus. The next consecutive locus after the previous one that you selected will automatically be highlighted as the default. Therefore, after the first locus only one key, ***enter***, need be pressed.

VI.K.13.b.iv.) *Insert pedigree/locus in report* puts the kinship file, the likelihood ratio formula and value, and documentation as the chosen allele probability values, into the report.

On return to the **Kinship** menu, the *rework* option helpfully is highlighted, so you only have to press ***enter*** to return to step §VI.K.13.b.iii.

VI.K.13.b.v.) *Print report (=Print)* after all pedigree/loci have been added to the report.

# VI.L. Special situations



## VI.L.1. Twins

Sometimes the problem arises of deciding whether twins are monozygotic or dizygotic. This seems like a natural target for the *Kinship* program, but there is a snag: *Kinship* syntax requires that the scenarios being compared have the same list of persona. Since the monozygotic scenario implies a single (genetically speaking) individual where the dizygotic scenario has two, the persona lists are not the same and therefore the input language seems to be inadequate.

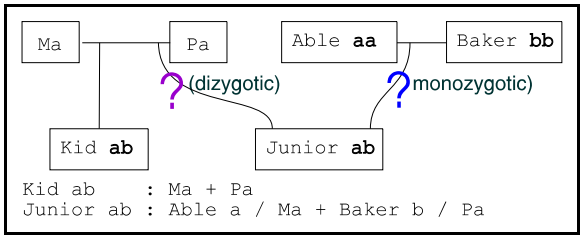



**Figure 68** Monozygotic vs. dizygotic twins

**Figure 67** shows an artifice that can be used to get around this kind of difficulty. We consider the situation where only the children, called *Kid* and *Junior*, are typed, and both heterozygous **ab**. We need somehow to represent the idea

```
TwinKids ab          : Ma + Pa    /*/ monozygotic hypothesis        (1m)
Kid ab, Junior ab    : Ma + Pa    /*/ dizygotic hypothesis          (1d)
```

in legal *Kinship* notation. The problem is to find a way to merge the above two hypotheses into a single set of pedigree definitions including the / symbol for alternate role-players. A correct solution is one that is equivalent to (1m) if you read the left side of each /, and equivalent to (1d) on reading the right side of each /. A first attempt might be:

```
TwinKids ab / (Kid ab, Junior ab)    : Ma + Pa
```

but this is no good because parentheses are not allowed. There is no way to have two people (the dizygotic twins) occupy a role equivalent to that occupied by a single genetic individual (the monozygotic twins) in **Kinship** notation.

The solution is to invent a new problem that will have the same likelihood ratio as the real problem. For this purpose two mythical parents

```
Able aa % Baker bb                                                   (2)
```

are invented because such parents always produce *ab* children. Line (2) appears the same way under both scenarios, so it just introduces the same factor (namely the factor $a^2b^2$) to the probability of each scenario and therefore doesn't affect the likelihood ratio.

Now we note that the monozygotic scenario (1m) is equivalent to the idea

```
Kid ab : Ma + Pa % Junior ab : Able aa + Baker bb                    (3)
```

whereas the dizygotic scenario is of course just:

```
Kid ab, Junior ab : Ma + Pa % Able aa % Baker bb                     (4)
```

which are the ideas of **Figure 67**. Finally, combining (3) and (4) in legal notation:

```
Kid ab : Ma + Pa
Junior ab : Able aa / Ma + Baker bb/ Pa
```

VI.L.1.a. Automatic Kinship treatment of twins

The same approach works with *Automatic Kinship*. Just set up the pseudo-parents Able and Baker with the appropriate homozygous type at every locus.

For a more detailed discussion, see http://dna-view.com/twin.htm.

Naturally, the method discussed above can be expanded to triplets, or to a case with additional reference relatives.

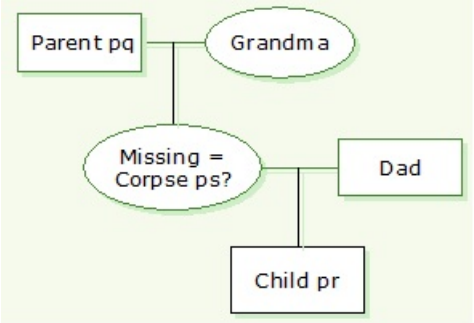



**Figure 69** Missing person and corpse

### VI.L.2. Missing persons

A person who is reported missing has both descendants and ancestors that can be typed, as in **Figure 69**. An unidentified corpse is found and typed. Is it the missing person? It seems that there should be a way to compare the two scenarios **same person** versus **different people**.

The necessary point of view to pose such a problem is to think of *Missing* and *Corpse* as two different people, and to consider them as alternative (using **/**) children in the two scenarios:

```
Child pr : Corpse ps / Missing + ?
Corpse ps / Missing : ? + Parent / ?
```

This way, under the **same person** scenario, *Child* will have a *ps* parent called *Corpse*. Under the **different people** scenario, *Corpse* will be an unrelated person and the untyped person *Missing* would be related.

*Kinship* computes the likelihood ratio 1/4*p* for this example. See **Figure 71** for another example.

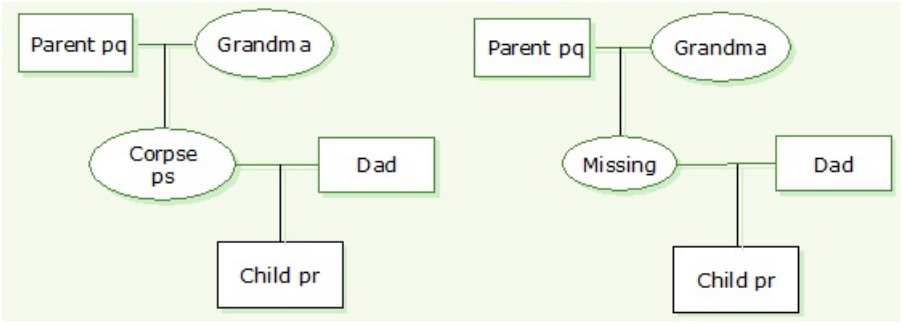



**Figure 70** Missing person solution

### VI.L.3. Indirect Exclusion

Select *Kinship* from the COMMAND menu, *Type in a new example*, and type the line

```
Child p : Mother pr + Dad q / ?
```

then ***Ctrl-e***. If silent alleles are not allowed, then Dad cannot be the father and the likelihood ratio is 0 (*Kinship* writes 0/*ppqrr*). However if *Silent allele: allowed*, Dad's **q** phenotype may be **qq** but it may also be **qo**, **o** being the silent allele. In the latter case, producing offspring like Child, with phenotype **p** is a possibility. *Kinship* gives as likelihood ratio:

```
o / (pq+2po+qo+2oo).
```

This can be factored as ($o$/(2$o$+$q$)) / ($p$+$o$). Compare the numerator and denominator with the formulas in §XIV.B.5.b.ii and §XIV.B.5.a.

# VII. DISASTER ANALYSIS

## VII.A. Overview

The DNA·VIEW disaster analysis module is designed to identify victims with references by DNA types. Victims are an arbitrary list of DNA profiles. References are another arbitrary list, but also, to the extent that the reference list includes several samples that belong to the same case, they are treated as a family. Multiple family members who seem to resemble the same victim are especially noted in the screening. The present version takes no particular systematic account of possible genetic relationships among victims, although the Kinship module of course permits assessment of all potential relationships.

Some of the programs are customized specifically for World Trade Center (WTC) identification data formats, and parts of this documentation reflect this. The modules are, however, ready to be used for any disaster.

The principal actions for victim identification are grouped in the **Disaster Screening** menu.

**VII.A.1. There are four major steps to the analysis:**



The input file can have all profiles – family reference, personal reference, and victim – mixed together.

Time: Importing 15,000 samples takes a few minutes.



Reduce the victim list by removing duplicates and probable duplicates



The screen computes likelihood ratios for identity, parent/child (accounting for mutation), and sibship between every family member and every victim profile. More importantly, it tests for two different family members resembling the same victim.

The resultant list is sorted by family in order of promise.

Time: 20 minutes to screen 2600 families (3+ members each) against 2000 (distinct) victims.



Compute a likelihood ratio for any combination of people.

Time: 1 minute - 1 hour per family, depending on success and complexity.

### VII.A.2. Additional – method+tool to establish reference allele frequencies



preparatory to compiling disaster-specific population frequencies.

## VII.B. Import DNA profiles

### VII.B.1. Overview

VII.B.1.a. Lists

The object of importing is to create lists – worklists of profiles to compare against one another. Typically create these lists:

VII.B.1.a.i.) Family+Personal references

The reference for each victim should be grouped together in a case, meaning that you assign a case number corresponding to each missing person.

Grouping them makes it possible for the screening heuristic to make better judgements than if it could only compare one-to-one.

VII.B.1.a.ii.) role letters for references

Putting the references into cases means assigning them role letters.

» For relatives of a missing person the role letter assignment under "Kinship roles" in **Figure 154** are suggestions.

» Eventually, when a likely candidate for the victim is recognized, that victim profile will also (tentatively) be added to the case, normally with a role letter of `V` or `W`.

» For personal references – DNA profiles or partial profiles that represent the missing person such as a profile obtained from a toothbrush or neonatal heel prick – the letters `J`, `K`, and `L` are preferred, especially if there are multiple personal reference profiles in the case. Those letters are treated in a slightly special way by the screening heuristic in that it recognizes they are probably versions of the same profile.

VII.B.1.a.iii.) Victim profile

The victim profile list is a set of profiles which have accession numbers but are normally not part of any case, at least not initially.

The screening program is arranged so that each list may be in several parts, i.e. may consists of several worklists.

VII.B.1.b. Repetitive screening

During the course of analysis of a mass disaster, new DNA profiles are likely to be developed continually over a considerable time. Screening and identification will go along in parallel. Therefore it will be desired to run the screening process repetitively. This can be done either

VII.B.1.b.i.) incrementally – by adding new data to the old in DNA·VIEW, possibly by adding additional worklists

VII.B.1.b.ii.) by starting over each time, re-installing DNA·VIEW with empty data files, then importing all accumulated data.

### VII.B.2. Import profiles

A large block of DNA profiles are imported once, or infrequently. Thereafter they are available for analysis within DNA·VIEW in various ways.

Import involves the following steps:

VII.B.2.a. (Optional) Clear away all data in DNA·VIEW per §XV.D.3.d.

Worklists (§V.J.1.d.i) can also be cleared with ***delete*** (then choose from the pop-up menu) when the bounce-bar is on the worklist name in the worklist menu. (But of course that wouldn't remove case definitions from DNA·VIEW.)

VII.B.2.b. Import using e.g. *Genotyper Import* §X.A.1

Normally you will import at least twice:

» once to create a worklist for the family and personal (direct) references

» a second import to create a worklist for the victim samples.

VII.B.2.b.i.) Automatic generation of cases from the input file

The family/personal reference data should be coded into *cases*. During the import, at step §X.A.2.h.i, designate that the cases should be **`kinship`**.

VII.B.2.b.ii.) Interpretation of off-ladder alleles

The *Option* (in the **Housekeeping** menu) *Off-ladder allele policy* determines how non-standard allele designations are interpreted. The best setting for disaster identification is `Treat as a rare allele` (rather than `Discard genotype`).

How rare is rare depends on the setting of the (*Case option*) *minimum frequency parameters* (§V.C.4). For *Disaster Screening*, see also *rare allele rate* (§IX.B.4.b).

## VII.C. Collapse Victim Profiles

The command *Worklist collapse* is found in the **Disaster Screening** menu.

It accepts a victim worklist as input, and creates a new worklist, a subset of the input list, as output.

### VII.C.1. Description

It produces a report with two sections

1. a "collapse" list of redundant profiles not used in the screening analysis (§VII.C.1.a),
2. a "deficient" list of those with deficient or nearly so (<1e11) matching LRs (§VII.C.1.b).

VII.C.1.a. Collapsed list of profiles

The "victim" collapse report fragment contains three samples (marked with ***=**) that are retained for the subsequent screening step, and below each of them are samples omitted. = means identical profiles. > means a common sub-profile according to rules that take into account common loci, possible phantom homozygous typings due to allelic dropout, and the matching LR threshold of at least $10^{10}$ comparing common alleles between two profiles.

The code "V 586" means a profile that has previously already been assigned as the victim of case 586.

```
*=V 586      e17
 >DM0221040 e12
*=DM0220262 e17
 =DM0226988
 >DM0220263 e16
 >DM0220401
 >DM0220264 e15
*=DM0220364 e17
 >DM0220363 e14
```

Collapsed victim profiles. The sample numbers ("DM"etc) reflect a World Trace Center custom modification.

VII.C.1.b. Deficient profiles

Non-collapsed profiles with matching LR $<10^{11}$ (when the profile is compared with itself) are listed. Possibly these need to be scheduled for further testing.

`e7.7`, which means $10^{7.7}$, applies to the samples until `e7.6`.

Note that sample id's such as `EDM FGJ4`, which have no systematic translation into DNA·VIEW numbers, are listed with both the Codis and the DNA·VIEW ID.

```
DM0226410     e 7.7
DM0225933
EDM FGJ4 1000-05135
DM0220404
DM0227111     e 7.6
DM0221045
DM0227666
DM0226513     e 7.5
```

Deficient profiles

**VII.C.2. Operation**

VII.C.2.a. `Choose race for computations`. It is not terribly important which databases are used for screening operations. **`C`** ***enter***, selecting "Caucasian," will do.

VII.C.2.b. Choose an input work-list.

`Choose the worklist to collapse`. Highlight the victim worklist, and ***enter***.

VII.C.2.c. Specify parameters for collapsing.

VII.C.2.c.i.) `Threshold odds for common part to collapse profiles`

The "common part" between two partial profiles means any loci in which typing are available for both loci, ignoring any discrepant loci for which the discrepancy can be explained by allele dropout. For example,

| **locus** | **a** | **b** | **c** | **d** | **e** | **f** | **g** |
|---|---|---|---|---|---|---|---|
| profile #1 | 11,12 | 10 | 21 | 17 | | 14,16 | 17 |
| profile #2 | 11,12 | 10 | | 17,18 | 9.3,10 | 14 | |

count as potentially matching profiles, with loci **a** and **b** considered as the common part.

If two profiles "agree" (defined per §VII.C.2.e.ii) at their common loci but each has types as some loci not found in the other, then inferior (lower matching LR) profile will be discarded if the matching LR for the common part is larger than the specified threshold.

For minimum collapsing, answer **`1E40`** and only sub-profiles will be discarded. This is the most conservative choice – and loses nothing but time – for a subsequent screening run.

For maximum collapsing, answer **`0`**. Intermediate values such as **`10000`** may give a fair estimate as to the number of distinct victim profiles available.

VII.C.2.c.ii.) `Threshold odds for smaller profile to delete a proper subset`

If one profile is a proper subset of another, it will discarded if it a matching LR (to itself) larger than the specified threshold. For purposes of subsequent screening there is almost nothing to lose by answering **`0`**, discarding *all* sub-profiles.

VII.C.2.d. `Threshold odds for "deficient profile" list` **`1E10`**

Specify parameter for appearance on the "deficient profile" list. The "deficient profiles" list is an advisory for possible additional DNA analysis.

VII.C.2.e. Choose whether to collapse, or only to count and report

VII.C.2.e.i.) `Collapse profiles (delete dups?)` **`y`**

VII.C.2.e.ii.) `Exact, Lenient, or Dropout, or STR dropout collapsing?` **D**

**Dropout** means 12 matches 12,13 or 13, but not 14,15. It is permissive of allelic dropout and seems realistic for degraded STR samples.

☞ **STR dropout** (recommended) uses the Dropout rule for STR's but requires an exact match for SNP's. This is a bit conservative for SNP's but there is no harm except computation time for retaining some unnecessary SNP profiles.

**Exact** matching means 12 doesn't match 12,13. Allelic dropout is considered impossible.

**Lenient** means 12 matches 12,13 or 13 or even 14,15. (This unrealistic choice was originally implemented as an approximation to the more complicated "dropout" option.)

VII.C.2.e.iii.) `Choose a worklist for saving the collapsed list`

Either create a new empty worklist, or select a scratch worklist that you are using for this purpose. The output of *Worklist Collapse* will be the input for a subsequent *Screen disaster matches* (§VII.D).

## VII.D. Screening

The command *Screen disaster matches* is found in the **Disaster Screening** menu.

### VII.D.1. Overview

You select two sets of worklists – family reference, and victim. All families are compared with all victims using several rough heuristics that tend to detect relationships. The several relationships listed in §VI.F.4.a are explicitly computed using fast algorithms.

If the lists are very large the screening will go much faster with no loss of effect if the victim list is a collapsed list (§VII.C).

The reference and victim list may be the same list. For example, comparing the victim list with the victim list may be useful in order to find relationships among the victims.

Also, the victim list may be selected as the "references" and vice-versa. This would be useful if it is desired to produce a list by victim profile of possible sources.

### VII.D.2. Result

The result is a Kinship Candidate report (§VII.D.2.a) with one paragraph per family, listing plausibly associated families and victims.

VII.D.2.a. Kinship Candidate report

The report is sorted with the families having the strongest, most plausible associations first.

```
#6. family 1199 MF
   DM0206582 vs FM LL>1E12
                vs F pi =60,000,000
             vs F si =600,000
             vs M pi =400,000
             vs M si =1,000
   DM0200368 vs M pi =100
```

```
#7. family 1699 UMF
   DM0206772 vs FM LL>1E12
               vs FU LL=300,000,000,000
                   vs F pi =200,000,000
             vs MU LL=90,000,000
             vs F si =3,000,000
             vs M pi =60,000
             vs M si =30,000
             vs U si =2,000
   DM0204434 vs FM LL=100,000,000
             vs F pi =200,000
             vs F si =60,000
             vs M si =600
   DM0227949 vs F si =2,000
   DM0227336 vs F si =2,000
   DM0227668 vs F pi =200
   DM0221943 vs U si =200
```

Above are two examples from early in the sorted list. The sample ID's reflect the input sample names except that prefixes are necessarily lost in the process of combining parts of the profile obtained from different analyses (i.e. with different prefixes).

For the first case above, #6 on the hit parade list, family 1199 (=case 1199) seems to be consistently associated with the victim sample DM0206582 (note the labeled and subsequent four lines) and to have a slight, probably coincidental similarity to the some other victim. Family 1199 is a DNA·VIEW case, which has roles F and M (corresponding to Codis codes BF and BM, father and mother of some victim). F and M each have a high PI statistic (single parent paternity index) associating them with the victim, and the record was sorted very high (position #6) based on the product of these two. (FM LL>1E12 means the product of two likelihood ratios, one for F and one for M, exceeds $10^{12}$. This number has no real meaning, but it is an indication to take a closer look at the suspected relationship. With so large an LL figure, the relationship will doubtless prove out.

The second case shows that family 1699 may well be full sibling (U) mother (M), and father (F) to DM0206772. The line `vs U si=2,000` means that U and the victim have a similarity that is characteristic of siblings (si=sibling index, that is, likelihood ratio favoring full-siblingship over unrelatedness).

The product of two likelihoods for DM0204434 is also so high (100,000,000), that probably these two samples represent the same person. The situation of two strong candidates, as here, is rare because the collapsing algorithm (§VII.C) will usually eliminate a redundant profile (the probable situation here).

VII.D.2.b. Screening

Then, the family-by-family analysis proceeds, comparing the families one at a time with all victims and preparing a paragraph per family as shown above. The screening step is very fast – typically several families per second although slower if there are thousands of victims or profiles have many loci.

You may interrupt the screening, for amusement, by hitting any key. Then you can ***space*** to step forward one family at a time, or **`r`** when bored to allow the program to resume at full speed. The program also responds to the **`b`** (back up) instruction, however beware: If you back up to a family for which the analysis just added a victim candidate, the second analysis of that family will regard the victim as previously, not newly, entered. This may affect the sorting of the final report.

The family paragraphs are then sorted according to a rough measure of likelihood of including a correct association, and compiled into a report. The report is again presented for cursory inspection (see §V.K.3). Hit ***esc*** when done cursorily inspecting. Then, from the main menu, you may print or file it as described above.

The families and associated victims may be regarded as kinship candidates.

**VII.D.3. Operation**

VII.D.3.a. Choose one or several races for searching computations. The best strategy is to nominate all the major races likely to be relevant. Enter the set of races with a string of one-letter abbreviations (per *Codings for races*, §IX.A.4)

See §IX.B.3.f for some details of *Screen* allele probabilities.

VII.D.3.b. Choose two sets of worklists to screen against one another (which need not be different)

`Choose the worklist with the families.` Highlight a family worklist, and ***enter***.

`Any more worklists besides those below?` is offered repeatedly to allow several worklists to be combined in case the references are not all on one worklist.

`Choose the worklist with the victims.` Highlight the victim worklist, and enter.

`Any more worklists besides those below?` is offered repeatedly to allow several worklists to be combined in case the victims are not all on one worklist.

VII.D.3.c. Select the screening heuristic rules and parameters

VII.D.3.c.i.) Family parameters/rules – if the reference list includes family groups (i.e. cases)

`Sort screening candidates (most promising first)?` **`yes`**
Normally **`y`** seems the more useful result. However, if screening victims against victims and the aim is to have a directory that can be manually searched by victim, the unsorted report would be easier to use.

`but sort families last if comment includes '>>'?` **`yes`**
To take advantage of this option, manually insert the character pair >> at any point in the case comment (§V.B.11; see also §V.B.28.a.ii) for any identification case that is complete or nearly

complete. Then, by answering **y** to this prompt, the hits to such cases, which are no longer exciting news, will be buried at the end of the list.

`"Name" to assign to automatically added victim` **Auto 2008/11/2 11:34**

Victim profiles sufficiently (§VII.D.3.c.ii) linked to a reference family will be added to the family as a convenience preparation for the detailed case analysis (§VII.F).

`What role for victim (usually V, maybe W)?` **V**

When the victim profile is added to a family, this is the role letter it will occupy.

`Remove previously assigned victims, personal refs, etc. from cases?`

`List roles to remove (V,W=victim, etc.)?` **V**

Discard the effect of a previous *Screen* operation if desired.

`Ok so far?` **y**

VII.D.3.c.ii.) Family parameters – numerical strength of link between family references and victim

`Automatically add victim to case if LL score >` **10E6**

If a victim profile possible association with a family is indicated by a sufficiently high score, then surely the victim will be at least a trial candidate for the missing relative of that family. Therefore the screening program can be helpful by adding sufficiently promising candidates automatically to a case.

The "LL score" means either a likelihood ratio, or product of two likelihood ratios comparing the victim profile with people in the reference family (case).[14]

`Report LL score (=product of 2 best LR's) if >` **100000**

This and the following several parameters specify which victim-reference pairs are interesting enough to report them as a line item in the screening kinship candidate report (§VII.E).

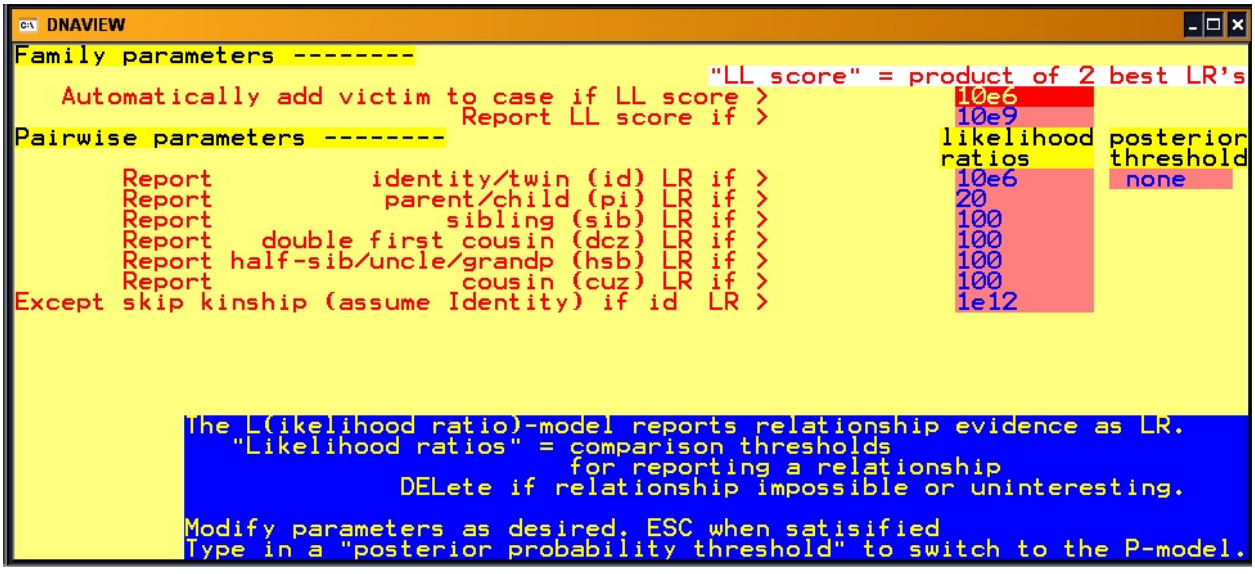



**Figure 71** *Screen* reporting parameters – Likelihood ratio model

[14] This statistic is intended as a rough approximation to the LR connection the victim and two people in the case. To be sure the right computation for that is quite different so this statistic is not necessarily meaningful. But it is quickly computable and after all is only used as an investigative lead.

VII.D.3.c.iii.) Pairwise parameters – the "Likelihood ratio" model

» The screening heuristic compares every reference profile with every victim profile in the six ways (identity, parentage, ..., cousin) shown in **Figure 71**. For each way you may set the threshold above which the likelihood ratio is considered interesting and should be included in the report.

» `Except skip kinship (assume Identity) if id LR >` **`1E12`**

If the LR of the evidence for identity is above the specified parameter then other relationships such as siblingship aren't interesting, and will not be reported notwithstanding the previous thresholds.

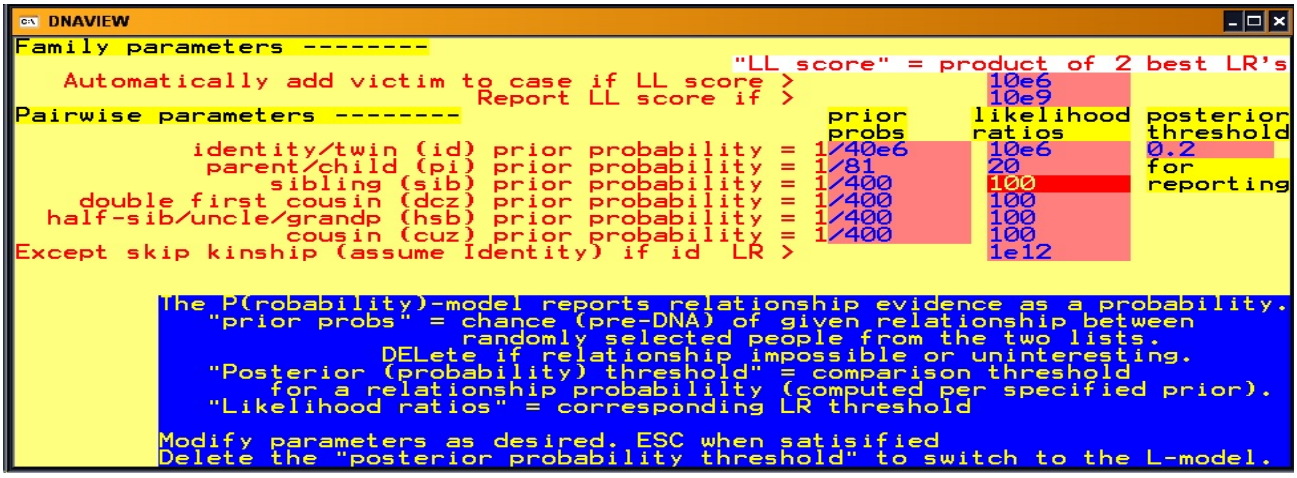
If you also want to do the opposite, to omit `id LR` lines for some reason, set the `Report id LR if >` parameter to an impossibly large value such as **`1E100`**.

» `posterior threshold` leave blank, set to **`0`** or ***delete***

Insert a value >0 to switch to the "probability model" per §VII.D.3.c.iii.

VII.D.3.c.iv.) Pairwise parameters – the "Probability" model

What likelihood ratio threshold is appropriate using the likelihood ratio model above (§VII.D.3.c.iii)? That depends on such things as the size of the identification effort. Instead of forcing the user to think through the arithmetic of prior probabilities mentally the program offers an alternative model which




embodies Bayes' Theorem and does the calculation for you.

» `posterior threshold for reporting` **`0.2`**

A threshold value >0 means that pairwise relationships that are at least [threshold] probable are interesting and should appear on the screening kinship candidate report.

To revert to the L-model (§VII.D.3.c.iii), set to **`0`** or ***delete.***

» `prior probabilities`

For each relationship, the *prior probability* means the average chance that a random reference and a random victim have that relationship. For example suppose there are 100 missing persons and 1/4 of the references are alleged to be siblings of a victim. Then the prior probability for a particular reference person to be the sibling of a particular randomly selected victim is 1/400. However, the number is closer to 1/100 for a reference that is alleged to be a sibling and to 0 for those that are not, so alternatively it may be more appropriate to write 1/100 for the prior. The

only risk is that somewhat more non-siblings who coincidentally look like siblings will clutter up the screening report.

## VII.E. *Screening summary*

Anytime after running a *Screen*, you may run this command to prepare a brief report based on the *Screen* run. The result is a 5-column, tab-delimited file that you can import to Excel to view, sort, and manipulate.

The file contains one row for each plausible connection between a sample from the victim worklists ("bone") and a reference family from the reference worklists, per §VII.D.3.b. "Plausible connection" is defined by the value chosen for the last of the screening analysis parameters of, `Report LL score` (§VII.D.3.c.ii).

The columns within each row are:

| | |
|---|---|
| RM# | a case number |
| bone | the accession number of a bone possibly related to the references of the case. |
| Role of bone | In case the bone already is part of a case |
| Case of bone | In case the bone already is part of a case |
| LL score | The qualifying score; product of the two largest relationship scores between the bone and two different references in the case. |

## VII.F. Test Candidate Relationship using Kinship

Start from the top of the family screening hit parade, and one at a time test the families with their likely associated victims using the *Automatic Kinship Case* program. Assuming the expected result with a satisfactory likelihood ratio is obtained, the victim can be regarded as identified.

### VII.F.1. Kinship steps

From the command menu, choose **Casework** ▸ *Automatic Kinship* ▸ *select case*.

VII.F.1.a. `Case #`

Type in a case number, e.g. **`1199`** ***enter*** to work the first case shown in §VII.D.2.a.

VII.F.1.b. Insert, if necessary, the suspected associated victim

If the case has no victim already, *Add role* should be highlighted. ***Enter*** to select it.

*Victim* should be highlighted. ***Enter*** to select it.

Type **`0200`** ***space*** **`06582`** ***end*** to add DM0206582 as role V of the case. (Actually leading 0's can be elided.)

VII.F.1.c. Go to kinship and evaluate

Strike **`/`** to highlight *Immigration/Kinship*. ***Enter*** to select it.

Test the relationships using the methods detailed in the DNA·VIEW manual. Chapter VI, Kinship, covers the principles of kinship scenario specification and analysis, and details of using the program. In outline:

VII.F.1.c.i.) *Type in scenario 1*

VII.F.1.c.ii.) Shortcut to analyze all races

*Calculate & report LRs, [all] races* determines the formulas, evaluates them for all races in the race list, and displays and adds to the report summary box with the bottom line for each race.

The more detailed per-race, per-locus calculation summaries are available if desired, or the report can be kept very brief:

`Include per/locus details for each race in report?` **`n`**

VII.F.1.c.iii.) Exit from kinship

Note that after some kinship operations a result will be displayed and a yellow wait banner will be at the bottom of the screen. It is not necessary to type ***enter***, then **`q`** to advance to the kinship menu and select *quit from immigration*; you can save a keystroke by typing **`q`** at the wait.

### VII.F.2. Technical comments about evaluating identifications

Alternates to the victim

VII.F.2.a.i.) Simple examples

Suppose the victim is denoted `V` and the suspected relationship is `V:M+F`. The alternative relationship can be thought of either as `V:?+?` (recall that each `?` asks the program to substitute a new unique name), or as `?:M+F`. The second version leads to the more attractive combined formula: `V/?:M+F` expressing the scenarios to be contrasted.

Similarly, if the suspected relationship is `S,D:?+V`, the combined formula can be written as `S,D:?+V/?`.

VII.F.2.a.ii.) Mistake to avoid

From the foregoing examples, it is tempting to conclude that the combined formula can *always* be obtained from the suspected relationship by noting each instance of `V` and substituting `V/?`. Not true.

It is safe, however, to substitute always `V/Other`.

The problem occurs when `V` needs to be mentioned twice. For example, suppose that the suspected relationships are

```
V : M + F
S : Spouse + V
```

Then the alternate relationships are *not* defined by the formulation

```
? : M + F
S : Spouse + ?
```

because each separate instance of `?` by definition refers to a different person, so it does not include the information that `S` is related to `M` and to `F`. Therefore the combined form

```
V/? : M + F
S : Spouse + V/?
```

is incorrect. It biases the computation towards the suspected relationships. Correct is

```
V/Other : M + F
S : Spouse + V/Other
```

VII.F.2.b. Likelihood ratio from Amelogenin

Almost all the victims (>80%) are male. Therefore a likelihood ratio of 2,000,000 from autosomal loci for identifying a male victim is not augmented very much by the evidential value of having the expected Amelogenin type.

### VII.F.3. More hints

VII.F.3.a. Pick list

The "pick list" can be useful. There are no doubt many cases with a reference mother, child, and sibling: Therefore it might be convenient to can the formula

```
V/Other:M + F
U : M + F
```

by adding it to the pick list.

Note that this same formulation is perfectly correct if the mother is not typed – there is no reason not to call her `M` anyway. Also, even if there is no sibling reference `U`, nothing is lost by including the second line except a barely perceptible amount of computation time.

VII.F.3.b. Mutations

The toggle *Do (don't) consider mutation* means that the likelihood ratio will never be reported as zero, but instead a small number will be reported. See §XIII.C.4, page284.

The small number is a reasonable estimate of the rate of paternal one-step mutations for the locus, but it is by no means therefore a reasonable estimate of the likelihood ratio for the present problem.

Example: Suppose a possible victim is being compared to two parent referenced: `V/Other:M+F` and in some locus the putative victim is PQ, the mother is PP, and the man is $Q^+$ R, where $Q^+$ is one step (repeat number) larger than Q.

Let *p, q,* μ be the probabilities of P and Q, and the mutation rate for the locus. Assuming, then, that half of all mutations are by minus one step,

Pr(genetic data | right victim) = Pr(PP woman)·Pr($Q^+$ R man)· ½ ·½ μ and

Pr(genetic data | wrong victim) = Pr(PP woman)·Pr($Q^+$ R man)·2*pq*,

so the likelihood ratio is μ/8*pq*, which may be much larger than μ although typically it is about 3μ.

In general, the mutation computation from the program should be regarded as no more than a signal. Mutations need to be worked out by hand.

VII.F.3.c. Allelic dropout

Assuming the same scenario as in the previous section, suppose the victim is P, the mother is P, and the father is QR. The program will of course report a mutation result, but in this case the real explanation may be not mutation but allelic dropout: the victim is really PQ.

A different, but still manual, analysis is necessary if this possibility is to be taken into account.

VII.F.3.c.i.) fudge the data

Examination of electropherograms may reveal a plausible Q in the victim. If so, modify (change and re-import the data file or use *Type in a Read*, §IV.D.4) the victim profile. Run the kinship analysis again. The formula will be 1/4*pq*.

VII.F.3.c.ii.) Possible dropout

Alternatively, suppose dropout is reckoned to be a possibility, but not a certainty. Then we need to use some estimate for the chance of dropout, the probability

*z* = Pr( not see Q allele | Q allele exists)

Assuming *z* is known, then

Pr(genetic data | right victim) = Pr(PP woman)·Pr(QR man)· ½ ·*z* and

Pr(genetic data | wrong victim) = Pr(PP woman)·Pr(QR man)·[2*pqz* + *pp*].

The right likelihood ratio is therefore *z*/[4*pqz*+2*pp*], which is roughly *z* times the result obtained above by assuming that dropout definitely occurred.

VII.F.3.c.iii.) Summary

A reasonable rough estimate of the likelihood ratio for a locus with possible dropout is obtained by modifying the data to the suspected (non-dropout) types, running the *Kinship* analysis, and multiplying the result by *z*.

## VII.G. *Assign Race to Worklist*

In assessing a possible relationship between a victim and a reference sample, the relevant probability is the relative likelihood that some unrelated victim sample would bear the observed resemblance with the reference. Therefore the relevant population sample to consider is the victims.

To create a set of victim allele frequency databases,

VII.G.1.a.i.) Import victim profiles to a worklist, per §VII.B.

VII.G.1.a.ii.) Use *Assign race to worklist* (in the **Disaster analysis** menu) to assign an otherwise unused race letter – e.g. `w` – to every sample in the victim worklist(s).

Equally, when viewing the list of worklists (e.g. using *Worklist*, §V.J), put the red bar on the desired worklist and click the ASSIGN RACE button.

You can optionally exclude assigning any race to children, which is usually a good option.

VII.G.1.a.iii.) Create databases, one for each locus, for the victim race. The protocol for doing this is detailed in §VIII.C.4 Building and maintaining allele population data ("database") in DNA·VIEW.

# VIII. POPULATIONS

## VIII.A. Population databases

A variety of population allele databases are included as part of a new install. Further or revised versions are supplied on an update CD ( in the **FREQ** folder) or may be had for the asking. Use the *Import/Export* method of §X.C.1.

DNA·VIEW has several facilities for creation, maintenance, examination, and comparison of allele population frequencies (“data bases”), which are discussed in this chapter.

## VIII.B. Population commands



**Figure 73** Population commands

## VIII.C. *Database*

The *Database* command comprises several options for creating, inspecting, and manipulating allele population databases for autosomal or X-chromosomal loci. **Figure 74** is the menu of options.



**Figure 74** *Database* command options

Note particularly §VIII.C.4 below describing procedures for maintaining databases of your DNA·VIEW data which can then be searched per §X.E.1.

### VIII.C.1. What a database is

"Database" means a database of allele frequencies for a particular locus or haplotype (e.g., Y-haplotypes), and a particular sample of people such as a specified race. However, the discussion in this section applies to autosomal databases. For Y-haplotype databases please see §VIII.F.2.

DNA·VIEW will maintain an arbitrary large list of databases and it is allowed to have many databases for the same locus and the same or similar populations.

Although the main information in a database is the list of allele fragment sizes and counts (number of times the allele was observed), further raw data is often stored such as the list of genotypes by person. This data can be obtained and analyzed in various ways.

The principal use of a database is probability computation. DNA·VIEW also has facilities to create a graph of a database (*Plot*, §VIII.E), or to search or list a database (*Import/Export* §X.E.1 *List and search database profiles* §X.E.1) or analysis (*Analyze database statistics*, §VIII.D, *List of homozygotes*, §VIII.C.11, *HW Check, RFLP*) of the information.

VIII.C.1.a. Database races

Every database has two sets of races:

VIII.C.1.a.i.) The "create races" — a list of 1 to 6 letters for the race codes of the people who go into the database. These letters are established when you create the database.

VIII.C.1.a.ii.) The "use-for races" (or "default races") — a list of zero to 6 letters that define when this database is used automatically for computations as discussed below (§VIII.C.2).

The default ("use-for") race(s) can be different from the creation race(s).

### VIII.C.2. Default databases

( – 2011)

DNA·VIEW is delivered with a large collection of databases, including typically many different choices for a particular locus and population (race). Therefore there needs to be a way to choose which one to use as reference data for calculation. For any particular locus and race, it is possible to mark one particular database as the *default database* – meaning the database designated to be used as reference data when any DNA·VIEW calculation program needs the probability of some allele.

When some DNA·VIEW program needs an allele probability to continue its computation (for case computation, simulation, racial distance or racial estimation calculation) it checks to see if there is a default already established and if so automatically uses the designated database. If not, then the user is prompted on the fly to supply the necessary information (§V.B.6.c), with the option to simultaneously set the databases choice as default for the future.

Alternatively, you may use *Set default database, per race* (§VIII.C.2.b) to establish in advance for each probe/locus and race, which database should be the default.

VIII.C.2.a. Specification concepts

VIII.C.2.a.i.) A particular database is indicated to be a default by virtue of a mark added to the database list in the "**Default for**" column as shown below.

| | DESCRIPTION | **Default for** | # of people | Date data compiled |
|---|---|---|---|---|
| CSF1PO | ABIdent Black (US) | Black( | 357 | 08/06/26 3pm |
| CSF1PO | ABIdent Caucasian | Caucas | 349 | 08/06/26 3pm |
| CSF1PO | ABIdent Native American | | 191 | 08/06/26 4pm |
| D2S1338 | ABIdent Black (US) | Black( | 357 | 08/06/26 3pm |
| D2S1338 | ABIdent Caucasian | Caucas | 349 | 08/06/26 3pm |

Note that there are defaults for `Black (US)` and for `Caucasian` for the two loci exhibited, but the third database isn't a default for anything.

VIII.C.2.a.ii.) Regardless of its creation race, a database can be assigned as default for several races (maximum of 6). This is useful in various ways. If for example you don't have D2 or D19 databases from population A but do have them for a similar population B, while for other loci you have ideal databases from both populations. Then as a reasonable expedient to enable Identifiler computations for both populations,

» set the available population A and B databases to be defaults to "use-for" their respective creation races (discussed in the above §VIII.C.1.a.i),

| | DESCRIPTION | **Default for** | # of people | Date data compiled |
|---|---|---|---|---|
| D8S1179 | Population A | A | 200 | 12/12/12 |
| D8S1179 | Population B | B | 327 | 02/03/04 |

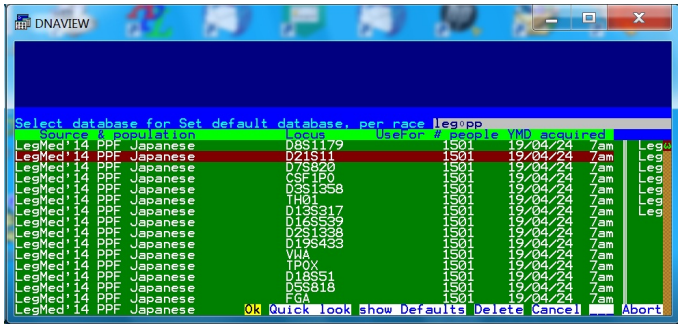
» and further set the population A D2 and D19 databases as defaults to "use-for" both A and B.

| | DESCRIPTION | **Default for** | # of people | Date data compiled |
|---|---|---|---|---|
| D2S1338 | Population A | A/B | 200 | 12/12/12 |
| D19S433 | Population A | A/B | 201 | 12/12/12 |

VIII.C.2.b. *Set default database, per race*


The submenu option **populations** ▸ *Database* ▸ *Routine* ▸ *Set default database, per race* lets you specify or re-specify which databases are to be used for computation when a database is required for a particular race and locus by *Paternity, Kinship, Mixture Solution, DNA odds, DNA exclusion, Racial estimate*, etc.

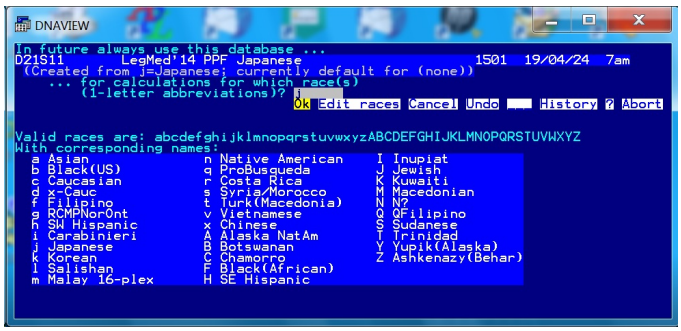
Typically related databases exist for a related set of loci such as the loci of a multiplex, and for the most part you will use the entire set together, as defaults. The method for establishing defaults is designed to make this easy while still allowing the user to mix and match if desired. The method is two steps: Choose the default for some representative locus, then choose the same default for similarly-named databases for other loci as follows.




VIII.C.2.b.i.) **Choose a default database for a locus**

Choose any database from the menu. The list of race names is displayed for reference, and you are prompted to select the race(s) for which the selected data base is to be used.

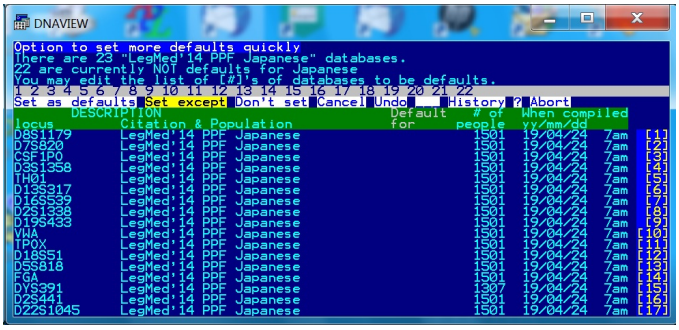
» Enter from zero to six race letters, then ***Enter***. The selected database will be used whenever any of these *default race* codes matches the *computation race* at the moment of computation.




- Common is to put the one race letter of the database's population.
- Entering *zero* races is a way to *remove* the default designation from some databases. (But a more common way to remove is to assign the default to another database of the same locus. Naturally, setting a database as default for a particular race removes the default status of any prior database for that race and the same locus. Only one database can be the default for a given race and locus.)
- Specifying multiple default races may be justified when two (or more) populations (races) are reasonably similar and one of them lacks population-specific database for some loci.

VIII.C.2.b.ii.) **set related databases**

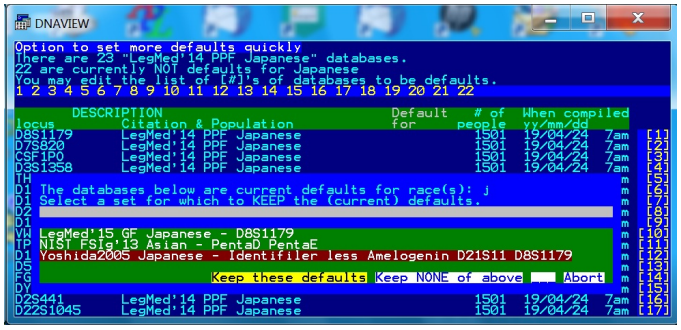
Now comes the good part. If there are similarly-named databases for other loci, you are given the option to set (some or all of) them as defaults wholesale.




Similarly named databases are listed on the screen. There is a reference number in brackets – e.g. [7] – at the right of each database. The suggested (default) response to the prompt includes all the numbers. There are some helpful buttons:

» SET AS DEFAULTS (optionally – after editing the reference numbers) to accept all the numbers, so that similarly-named databases will be defaults for the same race or set of races;

» DON'T SET to refuse changing any additional defaults;




» SET EXCEPT gives automated assistance for selecting just a subset of the [ ] numbers, in case you do not want to override some previously established defaults. It brings a popup selection window with descriptions of any conflicting (previously existing) groups of defaults. To *avoid* over-riding a group, select it then click KEEP THESE DEFAULTS.

VIII.C.2.c. default database on-the-fly

If there is no database already established as the default at the time the computational need for it arises, the program will pause and offer the user the opportunity to

» select (from among all databases for the locus) the database to use for the computation,

» establish default status for the selected database, and for similar ones for other loci, per §VIII.C.2.b.ii.

Therefore there is no need to explicitly invoke *Set default database, per race* except to *change* defaults. Initial default will quickly be established through the course of doing casework and answering the prompts.

VIII.C.2.d. List the default databases

VIII.C.2.d.i.) *Show default assignments per race for all races*

Prepare and display a report summarizing *all* the "default" database assignments, that is, the set of allele population data marked to be used as reference for calculations for a particular race. The report is in two sections:

» The *summary* has one line for each race for which there are default, summarizing the set of loci for which a default is specified such as

```
c=Caucasian    Identifiler less Amelogeni
x=Chinese      PowerPlex 18 less Amelogeni plus D1S80
```

» The *detail* is a sorted list of all the default databases.

VIII.C.2.d.ii.) Show the set of default databases for one race

From any menu display of databases, if the bouncebar is on a database the SHOW DEFAULTS button appears. Clicking on it results in up to three choice buttons as for example:

» C=CAUCASIAN DEFAULTS – to show all the defaults corresponding to the *creation race* of the highlit database (§VIII.C.1.a.i),

» G=GERMAN DEFAULTS – to show all the defaults corresponding to *default race* (§VIII.C.1.a.ii) of the highlit database,

» ALL DEFAULTS – equivalent to §VIII.C.2.d, *Show default assignments per race for all races*.

**VIII.C.3. Ways to make a database**

Following are ways to make an allele population database in DNA·VIEW:

VIII.C.3.a. Imported from any of various standard formats, discussed in §X.B;

VIII.C.3.b. Typed in. Select *Database* and then *Type in data to create a database*. See §VIII.C.12;

VIII.C.3.c. Compiled from casework or population study data that has been imported as a worklist (e.g. *Genotyper import*, §X.A.1), typed in, or digitized on DNA·VIEW (or that has been imported in a manner equivalent to digitization). Compiling a database is discussed below – §VIII.C.5 for a new one, §VIII.C.7 for rebuilding ("*update*");

VIII.C.3.d. Imported from another DNA·VIEW user per *Import/Export* ▸ *DNA·VIEW Frequency Database Import* (§X.C.1). See §X.C for the companion export and import methods;

VIII.C.3.e. Special method of import. This includes importing from a tab-delimited text file, or even from an idiosyncratic form by special arrangement (i.e., call and ask).

**VIII.C.4. Building and maintaining allele population data ("database") in DNA·VIEW**

VIII.C.4.a. Overview

At any time, DNA·VIEW can be asked to compile a population allele database based on accumulated profiles.

The genotypes (i.e., the DNA profile) of anyone with an accession number can be included in the database.

That certainly includes anyone in a *Case* (*Paternity*, *Automatic Kinship*, etc.), but more generally anyone who occurs on a worklist roster even if they are not used in any *Case*.

Any single database pertains to

» one specified locus

» one or more specified race letters.

That is, people are chosen for inclusion according to the race letter that is part of the definition of that person. In fact, selection for inclusion in a database is the whole reason for associating a race letter with a person. (The specification of the race to be used for calculation comes from a race letter associated with a Case (§V.B.13) or user-selected from a menu (§VI.F.7.g).)

» a range of accession (person) numbers.

Most often I select the full range of accession numbers from 80-00000 to 9999-99999, but if perchance you have in mind to create many categories for population studies, advance planning of your accession number scheme may be helpful.

» a set of "Reader numbers" (§IX.A.5, §X.A.2.d)

The default, usually most useful, is to use all of them (099=Genotyper might be the only one) except for 000=Student.

VIII.C.4.b. Preparation

VIII.C.4.b.i.) Routine

So keep the above in mind while designing data encoding and calculating cases. In particular, you probably want to reserve particular letters for the likely races of interest – e.g. **p** for Puerto Rican. Then, every man and every woman (but preferably not the children) should have the letter attached to their person-description.

There are a few ways of doing this. You may want to use a combination.

» Assign race to an entire worklist

With *Assign race to worklist* (§VII.G, **Disaster Analysis** menu)

- select a collection of worklists;
- say whether to exclude children (probably **yes**);
- choose some race (letter) such as **p**.

Then all the denominated people will be assigned to the desired race.

» Assign race to people in a case

For each case, select the case (in *Paternity case*), choose *edit people*, and the editing cursor will appear on the first person, the mother.

- ***Space*** twice to go past the accession number
- type e.g. **p** in the race cell
- ***tab*** to skip to the next person
- ***tab*** to skip past a child
- fix the (alleged) father(s) in the same way as the mother.

The most efficient practice is to make a habit of doing this while you are computing the cases. Then the data is ready to create databases whenever you like.

» Assign race to individual people in a worklist

- Open the worklist with *Worklist* from the **Casework** menu.
- Select the lane with the person of interest.
- ***Space*** through the fields to edit the person and introduce the desired race.

VIII.C.4.b.ii.) Preliminary check

The *List cases* (§V.B.17) option can be used to quickly review a sequence of cases to check if you have properly filled in the race letter for each person.

VIII.C.4.c. Compile database

Though 100 or so cases is typically regarded (for no particular reason) as a minimum size for a database, the most efficient way is to do the initial database creation (§VIII.C.4.c.i) when you have very little or even no profiles at all to fill them.

VIII.C.4.c.i.) Initial creation of databases

Even with no cases at all, you can build the (empty) databases for your lab. When you do it the first time, there is a separate operation for each locus, so you will need 2 or 3 minutes to do all 13 or so loci. The procedure is straightforward:

| keystroke(s) | reason |
|---|---|
| **Pop** *enter* | **Populations** from main menu |
| **D** *enter* | *Database* |
| **Com** *enter* | *Compile a new database from DNA·VIEW data* (§VIII.C.5) |
| **D8** *enter* | `Which locus?` Select the desired locus. |
| *Enter* | `Which readers?` *Enter* for default choice(s). |
| **p** | `Which race(s)?` Type 1 to 6 letters designating races to be included in the database. |
| *Enter* | Earliest acceptable accession ID = `80-00000`. |
| *Enter* | Last acceptable accession ID = `9999-00000`. |
| (time passes, probably a few seconds, and counters indicate progress as the program finds and compiles the relevant data) | |
| *End* (probably) | `Select the database to update, or End to make a new one` and the list of already existing databases is displayed. |
| *Esc* | to proceed to the next locus. |

VIII.C.4.c.ii.) Updating the databases

Once the databases have been built the first time, they can easily later be updated to reflect additional people who have been entered into DNA·VIEW. There are **two easy ways** to do this.

» The **first method** assumes that you have marked as defaults the databases that you want to update. If you've already been using them for casework, then this would be the case. Or, if you're planning to use them for casework after the re-compilation, then go ahead and mark them as defaults now, then proceed with re-compilation.

| keystroke(s) | reason |
|---|---|
| **Pop** *enter* | **Populations** from main menu |
| **D** *enter* | *Database* |
| **Up** *enter* | *Update the default databases* (but only those marked as from your own laboratory) |
| *Enter* | `Recompute these databases?` **y** |
| *Enter* | `Shall I skip printing the exception reports?` **y** |

» The **second method** is more general, but takes (just) a few more keystrokes:

| keystroke(s) | reason |
|---|---|
| **Pop** *enter* | **Populations** from main menu |
| **D** *enter* | *Database* |
| **U** *enter* | *Update arbitrary list of databases* |
| *Enter* | Accept lines containing `YourLabName` |

If desired to restrict the list further, type a context then ***enter*** (see §XIV.A.3). For example, if you are maintaining a universal set of databases for the purpose of offender searching with the race name `anonymous`,

| | |
|---|---|
| **anon** ***enter*** | limit the database list to the `anonymous` databases |
| ***Enter*** | to accept the list |
| ***Enter*** | `Recompute these databases?` **y** |
| ***Enter*** | `Shall I skip printing the exception reports?` **y** |

Obtain and drink coffee. Recompiling 100,000-person databases will take 1–10 minutes per locus, and will need 10mb of RAM available to DNA·VIEW (see §XV.B.9.1).

### **VIII.C.5.** *Compile a new database from DNA·VIEW data*

To compile a data base with you specify parameters to define the population of alleles to collect. See §VIII.C.4.c.i for more operation details.

VIII.C.5.a. a locus

`Which locus?` **D8S1179**

VIII.C.5.b. the list of readers whose data to use – usually OK

VIII.C.5.c. a race (or collection of races)

VIII.C.5.d. a range of accession numbers

`Earliest acceptable accession ID?` **80 -00000**

`Last acceptable accession ID?` **9999-00000**

The suggested default range of accession numbers, from **80-00000** to **9999-00000**, probably includes all actual samples (particularly if you use the leading part of the accession number as a year), while omitting "special"numbers such as **1-564** that might represent quality control samples.

Next, the program proceeds to gather all data consistent with the specified parameters, which may take some seconds. Progress is indicated on the screen.

The data is compiled from readings of lanes labeled with an accession number in the specified range. Control subjects and other miscellaneous lanes can be put in or left out in a variety of ways according to your choice, depending on how you have coded the roster information for the lanes. One convenient way to omit incidental samples from the database is to assign them a very small year number – e.g. some use `1-562` for K562 samples. Other possibilities include assigning a race code of –, or labeling the second roster column. All these ways still permit analysis with *Statistics* or *Quality Control*.

In the case of a PCR locus or single-locus probe database, each person contributes two alleles. A homozygous allele is counted twice. Multiple reads and/or lanes for a person are averaged (as described by algorithm §XIII.F).

VIII.C.5.e. Filing the new database

When the new database is ready for filing, if there are any databases that you previously created for the same locus and race(s), you will be given the option of updating an old slot with the new data, or creating a new slot.

A menu of already existing databases for the specified locus is presented. You have three choices:

VIII.C.5.e.i.) REPLACE *an existing database*

Choose one of the already existing databases and click REPLACE. In this case before any action is taken the two are presented side-by-side on the screen for you to compare, and you are given the option to confirm or change your mind, as shown in **Figure 79**.

(If you change your mind and choose *not* to overwrite the selected database, you will next be given the option to skip filing the database altogether.)

VIII.C.5.e.ii.) CREATE NEW to add a new database in addition to those that already exist.

VIII.C.5.e.iii.) CANCEL to *skip filing*

### VIII.C.6. Side effects

There are several side effects to compiling a data base:

- An Exception Report (see §VIII.C.8.a) is created covering any suspicious circumstances. The report can be printed using *Print*. It is also saved, and can later be retrieved using *Retrieve exception report*.

  For a single-locus probe, suspicious circumstances include inconsistent data for any individual, such as readings from the same lane, or from different lanes, that don't agree to reasonable precision; inconsistent decisions on homozygosity among different readings or lanes; and lanes that are labeled but never read, or read only once.

  In the multi-locus case, the report lists worklists with lanes read more than once, and people with data from more than one lane.

- [PCR- or single-locus] A special record is made and saved of homozygotes, which may be examined using the *List of homozygotes* (§VIII.C.11) sub-menu option of the *Database* command.

- [PCR- or single-locus] A special record is made of individuals run more than once — i.e. on several worklists or lanes. See *Printout of a database* (§VIII.C.13).

### VIII.C.7. Updating

Updating a database means to recompile a database as a collection of profiles available in among your own data – case data and/or orphan profiles. Updating is in effect a way to re-run *Compile a new database from DNA·VIEW data* (§VIII.C.5) automatically and en masse. Hence it is only possible for database that were compiled locally per §VIII.C.5. Updating imported databases, whether shared from another DNA·VIEW installation per §X.C.1 or imported per §X.B, is not possible.

Since you may often want to update all or many of your databases at the same time and there are likely to be quite a few of them (one for each probe-race combination), several options are available for convenience.

VIII.C.7.a. *Update the default databases* lets DNA·VIEW take over the keyboard and automatically replaces each database that is

(a) marked as a default, and

(b) is from your own laboratory.

Each newly compiled database then replaces the older version.

VIII.C.7.b. *Update arbitrary list of databases* first lets you select an arbitrary collection of databases. The method of selection is by contextual phrases, as described under §XIV.A.3, "Multiple selection". DNA·VIEW then takes over the keyboard, recomputes each specified database, and replaces the old version with the new one.

Exception reports are automatically printed out for each recompiled database under either of the last two options. But if you want to save paper you can temporarily change the printer to *(none)* (see §IX.F.9).

VIII.C.7.c. Detail remarks about updating

The updated version is compiled based on the same race(s), accession number range, and probe number as were specified for the previous version. DNA·VIEW suggests the accession number range of `80-00000` to `9999-00000` when you initially create a database so that it will be convenient to use the *Update* method to create ever more inclusive versions.

Also, if you have manually changed the name of the database, your new name will be retained after the update.

The set of readers, however, is not remembered from the previous database compilation. Rather, the updated database is always based on the default set of readers which is those numbered from 1 to 999. STUDENT (000) is therefore omitted, as would be any irregular readers (since normally there aren't any) with numbers 1000 and above.

**VIII.C.8.** *Review exception report*

This is an obsolescent option mainly intended for RFLP. For most of the description, see an old version of the manual.

VIII.C.8.a. 3-banded patterns

The exception report includes a list of people with 3 bands.

**VIII.C.9.** *Delete or rename*

to select a data base for deletion or to rename. As usual, you are asked to confirm your selection before the data base is deleted. If you do *not* confirm deletion, then you are given a prompt to rename it.

**VIII.C.10.** *Combine*

lets you create a single database as a combination of several. Two likely reasons to do this:

» combine databases from several different laboratories into one larger one,

» combine a database compiled with DNA·VIEW and one entered by typing.

An important point to appreciate: Suppose that you have created a database without DNA·VIEW, then you acquire DNA·VIEW and after a time have accumulated data in DNA·VIEW that you would like to add to your earlier data. *Do not* select the typed in database for "updating." That would simply *replace* the typed-in data with the accumulated data. The only method that works is to create three databases: The typed-in one, a second one from the DNA·VIEW data, and a third one of the combined data.

VIII.C.10.b. Example steps for combining databases:

**populations** ▸ *Database* ▸ *Construct* ▸ *Create a combination of databases*
`First Combination of databases component?` Choose a database, OK
`Additional ... component?` Choose a database, OK
`Additional ... component?` Choose a database, OK
`Additional ... component?` DONE
(# of people) OK; repeat for each component.
`Is all the information correct?` **y**
`Create a new database or replace an existing one.`
*edit the name* [retype if you like] OK

**VIII.C.11.** *List of homozygotes*

As a side effect of compiling allele frequencies (see *Database*, *Compile ...* or *Update ...*),a record is saved of all people for whom only one allele was digitized. The list of their accession numbers can be retrieve using this sub-menu option, then printed with *Print*.

The accession number lists for several data bases with the same probe may be combined into a single sorted list.

Accession numbers are not available for imported data bases compiled at a foreign site.

**VIII.C.12.** *Edit or type in a database*

Editing a database is done with a friendly full-screen editor. Features include:

» Edit an existing one, or create a new one and type in the alleles and count of occurrences.

» PCR allele notation or base pair notation both allowed

» Search for sizes, and automatic scrolling.

» Automatic scaling allows you to enter data from a table that gives decimal frequencies (enter on a per-thousand basis), then adjust the frequencies to an allele-count basis by changing the total to the correct total number.

VIII.C.12.b. Allele name and count

The input screen requires allele name (allele number for STR, letter codes for SNP's, Amelogenin, or poly-markers, base pairs for RFLP) and *count* – not frequency. However, if you have a source document that lists frequencies, it is easy to make the program do the conversion.

VIII.C.12.c. Protocol that converts frequency to count

Suppose for example that the input document lists the 3-allele population sample shown in **Figure 79**, not explicitly listing the actual "counts." However, that is no problem. for here's a simple trick. Enter the decimal number scaled by 1000 – i.e. enter the database on the basis of n=1000 alleles, as shown at the right:

Then, at the bottom of the screen where it says "Total alleles", change the scaled total of 999 to the correct number, 114. The numbers for allele count automatically change proportionally.

Now, click `Save and exit`, and the computer will round to the exactly correct values – 3, 12, 99 – and save them.

VIII.C.12.d. Automatic conversion of frequency to count

Alternatively, you may enter or scale the numbers to actual frequencies (total alleles =1). Then click `Save and exit` and a dialogue, beginning with the program's plausible guess as to the correct total, guides you through conversion of frequency to count.

VIII.C.12.e. Null alleles

Null alleles are handled in DNA·VIEW by adding a count to a database. To enter null alleles, type the letter **n** in the `allele` column, then an appropriate count in the `count` column.

**VIII.C.13.** *Document sample frequencies etc. tables*

(redesigned – 2011)

VIII.C.13.a. Choose information to report

» C=Count – # of occurrences

» F=Frequency, i.e. sample frequency

» P=Probability, i.e. conditional probability to see the allele at random given the user options currently selected (which are shown on the screen)

VIII.C.13.b. Choose the locus

From the menu of loci, choose the first (remaining) one you wish to report.

Example: `Which locus?` **`D16S539`**

VIII.C.13.c. Choose some databases

Use the "mass database" method (§XIV.A.3) to select one or several databases to print out.

Example:

`Accept lines containing?` ***JFS enter***
`Accept lines containing?` ***44(6) enter*** reduces the list further.
`Accept lines containing?` ***Enter*** proceeds to the reporting step.

VIII.C.13.d. Database list displayed

The selected databases are appended to the database report, and the report so far is displayed.

**D16S539 STR**

| | JFS 44(6) Black | | | JFS 44(6) Caucasian | | | JFS 44(6) SW Hispanic | | |
|---|---|---|---|---|---|---|---|---|---|
| | Count | Freq | Prob | Count | Freq | Prob | Count | Freq | Prob |
| 8 | 15 | 3.59% | 3.82% | 8 | 1.98% | 2.22% | 7 | 1.68% | 1.92% |
| 9 | 83 | 19.86% | 20.05% | 42 | 10.4 % | 10.62% | 33 | 7.93% | 8.15% |
| 10 | 46 | 11 % | 11.22% | 27 | 6.68% | 6.91% | 72 | 17.31% | 17.51% |
| 11 | 123 | 29.43% | 29.59% | 110 | 27.23% | 27.41% | 131 | 31.49% | 31.65% |
| 12 | 78 | 18.66% | 18.85% | 137 | 33.91% | 34.07% | 119 | 28.61% | 28.78% |
| 13 | 69 | 16.51% | 16.71% | 66 | 16.34% | 16.54% | 43 | 10.34% | 10.55% |
| 14 | 4 | 0.96% | 1.19% | 13 | 3.22% | 3.46% | 10 | 2.4 % | 2.64% |
| 15 | | | 1.19% | 1 | 0.25% | 1.23% | 1 | 0.24% | 1.2 % |
| | 418 | 100 % | 102.63% | 404 | 100 % | 102.47% | 416 | 100 % | 102.4 % |

**Figure 80** Database printout

CLOSE the report display

`Append more databases to the report?` **n**

Or answer **y** to resume at step §VIII.C.13.a.

**VIII.C.14.** *Print allele list, frequencies* **(RFLP)**

This obsolescent options produces a detailed report of only one database which was defined with RFLP in mind. For STR, prefer §VIII.C.13.

## VIII.D. Database statistics and information

The command *Analyze database statistics* (also included as a menu option under *Database*) gives, for a selected collection of databases (but see §VIII.F.5 for Y-haplotype databases), a number of useful statistics the measure the discriminatory power of the locus/race the database represents. Summary statistics over each natural group – databases representing the same laboratory+race description, presumably representing one population sample for various loci – are presented, and the per-locus statistics are optionally available as well.

### VIII.D.1. Operation

VIII.D.1.a. Select a set of databases to analyze using the "Multiple selection" method, §XIV.A.3. For example,

`Context to filter on` **`JFS`** `ACCEPT` selects the 331 or so whose names include "JFS".
`Context to filter on` **`44(6)`** `ACCEPT` reduces to 40 databases.
`Context to filter on` **`D1S80`** `REMOVE` reduces to 39 databases – 3 races times 13 loci.
***Enter*** (`ACCEPT` empty phrase) to continue to the next step

VIII.D.1.b. `Do you want only the group summaries as the report` `YES`/`NO`

### VIII.D.2. Results

VIII.D.2.a. per-locus details example (if you answer `group summaries:` `NO`)

```
D8S1179  STR JFS 44(6) Black              180  01/10/30 12pm
        A = 60% = mean paternity exclusion power, based on
        H = 22±2% = effective homozygosity rate (discrete alleles).
        PD = 92% = expected power of identity discrimination.
        PI = 2.89 = typical PI (trios).
        mPI = 2 = typical PI (motherless).
        S = 24% (=43/180) single-band rate
        Pr(overlooked band) plausibly from (S-H)/2 = 1% to S/2 = 12%.
```

The statistics are mainly various ways of measuring the discriminating power of the system. Some of them are requested by the AABB as part of the process of accrediting laboratories.

VIII.D.2.b. per-multiplex summary example – summary over a collection of loci:

```
Cumulative mean statistics for 13 JFS 44(6) Black databases
        D3S1358 VWA FGA D8S1179 D21S11 D18S51 D5S818 D13S317 D7S820 D16S539 TH01 TPOX
        CSF1PO
        Matching chance=1/900e12  A=99.9991%   PI=1.92e6(=3.04/locus)  mPI=13800(2.08)
```

VIII.D.2.b.i.) **Matching chance** is the typical probability for two randomly selected unrelated profiles to be identical. What is sometimes called PD (probability of discrimination) is 1−this number.

VIII.D.2.b.ii.) **A** is the typical probability of so-called "exclusion" for paternity casework – the chance that a random non-father would be excluded if we did not believe in mutation.

VIII.D.2.b.iii.) **PI** means the *typical* paternity index for the given battery of loci. The number in parentheses is the (geometric) average contribution per locus. (That is, $3.04^{\text{number of loci}}=1.92\cdot10^{6}$).

VIII.D.2.b.iv.) **mPI** means the typical **motherless** paternity index – i.e. when the mother is not available for testing. The number in parentheses is again the average contribution per locus.

## VIII.E. *Plot*

The *Plot* program creates graphs of allele frequency distributions. Several distributions can be superimposed using different colors, and the user can choose parameters determining just how the plot will be drawn. The plot can also be printed or (better) exported as a graphic file.

Consequently, *Plot* is an extremely powerful tool for exploring a variety of interesting and practical questions about DNA allele population frequencies.

For example, from the plot of a single database, you can see immediately the sizes and probabilities of the main alleles.

Comparing different races lets you see where, and how significant, the frequency differences are. Do Hispanics differ significantly from coast to coast? Are there perhaps certain alleles for which they differ? Artfully drawn graphs tell the story clearly.

A practical test as to whether population statistics are significantly different is whether they result in markedly different calculations of paternity index. Instead of comparing population frequencies themselves, you can ask for and visually compare plots of the paternity indices, calculated and graphed for every different fragment size.

### VIII.E.1. Operation

VIII.E.1.a. Open DNA·VIEW in "Plot" mode.

*Plot* won't work if you open DNA·VIEW with the regular icon. You have to use the "DNA·VIEW Plot" icon (§XV.D.2.a.iii), which requires running in Windows XP or *below*. Plotting from Windows 7 can be achieved indirectly by using DOSBOX or Virtual PC.

VIII.E.1.b. Invoke *Plot* (in the **Populations** menu)

VIII.E.1.b.i.) Choose the "primary data base" from a list of databases (allele frequency distributions) to plot. Selection can be made by the usual menu selection methods. Selection by multiple contexts (§XIV.A.2.a.iii) – e.g. **D3Caucas** – is particularly useful here.

VIII.E.1.b.ii.) *Start from?* and *End near?*

refer to the abscissa limits. The defaults will encompass every last allele; you may want to trim back from this, or to include more if you are planning to superimpose another plot. A chart (**Figure 82**) showing the numbers of alleles in broad brackets will help you decide on appropriate abscissa values.

The bracketed %'s (e.g. [0.1%]) in the chart indicate alleles not present in the selected database and show what is their overall frequency among all the installed population studies.

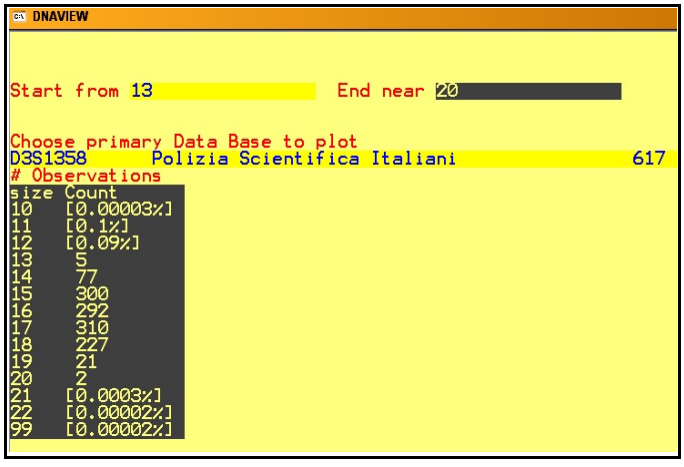



**Figure 83** selecting parameters for a Plot

That concludes the selection of parameters (in the STR case). You now have a choice menu to opt from, shown in the screen appearance illustrated in **Figure 83**. The choices are:

VIII.E.1.b.iii.) *DISPLAY* is the initial default. It results in creating the graph and displaying it on the video monitor. When you have looked at it for long enough, ***Enter*** or ***ctrl-c*** to return to the choice menu.

VIII.E.1.b.iv.) *PRINTER* – A (not very attractive) copy of the graph is printed on the printer. A better option:

VIII.E.1.b.v.) ☞ ☞ *METAFIL* – to create an editable graphic file for use in a publication or other document

A coded image of the graph is written to a file. The file format is called CGI or CGM. It can be imported into Word, Powerpoint etc., or edited with a graphics program such as Corel Draw.

VIII.E.1.b.vi.) *start over* – Go back to the beginning of *Plot*, except that the choices you made previously will be remembered and used as defaults.

VIII.E.1.b.vii.) *quit* – Exit from *Plot*.

VIII.E.1.b.viii.) *more plots* – Get another database to superimpose onto the graph.

If you select a database that is already included in the graph, the prompt `smooth by?` – intended for RFLP databases – offers to simulate measurement uncertainty.

VIII.E.1.b.ix.) *fewer plots* lets you back up by removing a database from the plot.

VIII.E.1.b.x.) *homozygotes* – similar to *more plots*, but the graph is of just the homozygous individuals found for the given probe. This information is usually available only for data compiled by DNA·VIEW, as opposed to data that was typed in.

VIII.E.1.b.xi.) *paternity index* creates a special superimposed plot consisting of the paternity index calculated point by point by considering in turn each size on the X-axis to be the obligatory paternal size for a typical paternity case.

As a useful side effect, the graph legend will report the "typical PI" (geometric mean, see Brenner & Morris (1989), page 36).

Notes on scales: If one or more graphs of paternity index are requested, then the Y-axis is labeled with the PI. The PI's will be packed close together toward the top of the screen, so that an infinite PI corresponds to the top margin.

VIII.E.1.b.xii.) *turn histogram on/off* – This option is only for demonstration when displaying RFLP data.

### VIII.E.2. Experimental *Plot* features

The features mentioned in this section are not meant to be counted as part of the DNA·VIEW product. They are not guaranteed to work, nor to persist in future versions. The descriptions are in most cases admittedly and intentionally sketchy.

VIII.E.2.a. Modify *Plot* Colors

While looking at a plot, you can modify the colors.

Sketchy background information: Each color is a mixture of Red, Green, and Blue, intensities ranging from 0–1000, and these intensities are typed in using the letters a–z. Each color in the plot has a number, 0, 1, 2, 3, 4, 5, 6, 7, 8, 9, A, B, C, D, E, F. 0 is the background, 2 & 3 are the grid, 4 the centered caption, 5,6,... the colors of the individual graphs.

As you are changing the colors, there is some coded feedback in the lower right hand corner of the screen. ***Space*** makes it disappear.

Examples: **`5zaa`** changes color 5 to solid red. Then **`ufo`** changes color 5 to fuchsia. Now hit **`-`** or **`+`** and watch the red intensity diminish or increase. Type **`.`** to advance to the next primary constituent. **`0;1`** exchanges colors 0 and 1. **`/`** and **`\`** restore the current color, and all colors, to their values at the beginning

of the plot. **#** switches to an alternate palette. This is necessary before printing to a color printer if you want a white (unpainted) background.

The key **!** creates a gradation of intensities merging to the background. **$** undulates the colors over time.

VIII.E.2.b. Show Marker Sizes (RFLP feature)

Marker sizes can be superimposed on the plot by choosing the option `Ladder` from the option menu.

VIII.E.2.c. Comparing Different Smoothings (RFLP feature)

If you choose the *Plot* option *more plots* and choose a plot that's already been chosen, DNA·VIEW suspects you want to use a different smoothing the second time, and gives you an opportunity to specify it.

# VIII.F. Y haplotypes in DNA·VIEW

improved

## VIII.F.1. Y haplotype overview

new

VIII.F.1.a. Y haplotypes

The Y haplotype for an individual is the alleles for a collection of loci on the Y chromosome. Importing Y haplotypes is therefore the same as importing autosomal data – specify the alleles for some loci.

For calculation and "databases" though Y haplotypes of course need to be treated as a haplotype unit, never as individual loci, and this is quite different from the treatment of autosomal (or X-linked) loci in DNA·VIEW.

VIII.F.1.b. Y loci

Some collections of Y loci, such as YFiler, are included in the multiplex list (§IX.B.7). However, any arbitrary set of Y loci is allowed to comprise a Y haplotype. Any locus is considered by the program to be a Y locus or not according to the genetic type (§IX.B.3.f) coding associated with the locus.

VIII.F.1.c. What are Y haplotype databases?

A DNA·VIEW Y-haplotype database consists of a population sample – a collection – of Y-haplotypes from test subjects, for a particular racial category.

Y-haplotype databases are different from single-locus (autosomal or X) databases. There is a special list of Y-haplotype database, and the ways to manipulate, maintain, and use the Y haplotype databases are quite different from the autosomal database facilities.

You may have Y-haplotype databases for as many populations as you wish. Each database

- has identifying parameters
  - race (a race letter)
  - name (arbitrary text description)
- haplotypes
  - for any set of Y-loci
  - any number of individuals
    - incomplete haplotypes (missing locus here and there) are allowed
    - multiple alleles per locus are not allowed as of now

It's permitted to have several Y-haplotype databases associated with the same race. The first database for a given race letter has priority. Therefore the order of the databases (§VIII.F.4.a.i) is significant.

VIII.F.1.d. Y haplotypes for identification

Several DNA·VIEW programs use Y haplotypes in computation.

VIII.F.1.d.i.) Crime stain matching

*Y-haplotype matching LR* (§V.H) looks up a given haplotype in the Y haplotype databases and reports the evidential value of a match.

VIII.F.1.d.ii.) Kinship and body identification

A Y haplotype LR can be incorporated into a kinship calculation. Some pairwise matching LR tables include Y haplotype evidence. See §VI.F.5 **Y in kinship**.

VIII.F.1.d.iii.) Disaster screening

The *Screen disaster matches* module takes Y-haplotype comparison into account.

VIII.F.1.e. Y haplotype database operations

» Import or enter Y profiles in the same ways as autosomal data. The Y-haplotypes are input and organized within DNA·VIEW as collections of individual loci. However, computations and databases always treat them as haplotypes.

» *Y Databases* (§VIII.F.2) (in the **Populations** menu) includes functions to create (§VIII.F.3), manipulate, and analyze (§VIII.F.5) Y-haplotype databases.

**VIII.F.2.** *Y Databases* **functions**

| | |
|---|---|
| | *Clear away all Y-haplotye databases* |
| | *Install Y-haplotype databases from catalog* |
| VIII.F.3 | *Install Y-haplotype databases from worklist*. . . . . 175 |
| VIII.F.4 | *Rename, reorder, delete Y databases*. . . . . 176 |
| VIII.F.5 | *Y-haplotype database statistics*. . . . . 177 |
| V.H | *Y-haplotype matching LR*. . . . . 87 |
| | `quit` |

**VIII.F.3.** *Install Y-haplotype databases from worklist*

improved

You may have as many Y haplotype databases as you wish. ☞ You can install them one at a time or several at a time.

VIII.F.3.a. Preliminary – prepare worklist

Each population/database corresponds to a worklist. Therefore before adding one or several Y-haplotype databases, first import the haplotypes for each population into separate worklists. (**Note:** Under **Import/Export**, use an option (such as *Genotyper import*) from the*DNA profile data import* menu (§X.A); do *not* use *Database import* (§X.B).) Importing is the same as for autosomal data – the haplotype for each person consists of multiple loci worth of data.

VIII.F.3.a.i.) **DYS385 note** – For yconvenience a special provision is made for DYS385a/b. If you enter genotypes for just one of these loci the program will automatically split it between the two loci for you. But the loci DYS389I and DYS389II are treated as two completely different and independent loci; you need to explicitly supply the allele for each one.

That done, invoke **Populations** ▸ *Y Databases* ▸ *Install new Y-haplotype databases from worklist*.

VIII.F.3.b. Select worklist for a Y-haplotype population

The profile data associated with the specified worklist is gathered.

VIII.F.3.b.i.) Any roster attached to the worklist is irrelevant and ignored. This is exceptional compared to the way DNA·VIEW normally gathers data from a worklist. In particular this means (exceptionally) that further data, on another worklist, will not be collected even if the other worklist mentions some of the same sample numbers.

VIII.F.3.b.ii.) Illegal or unacceptable genotypes (note §VIII.F.3.a.i) are ignored, hence leaving a partial profile for the person.

VIII.F.3.b.iii.) Choose the race to associate with that worklist. You are also given an opportunity to enter a name for the worklist. If you don't, then by default the name is the name of the race.

VIII.F.3.c. After each selection, a summary screen shows the populations/worklists so far selected. For any sample, any loci with more than one allele are discarded. This means that in terms of matching, they will be considered not to match with any profile that has an allele in that position – a reasonable rule.


**Figure 84** Installing Y-haplotype databases

Repeatedly select more worklists for further populations until there are no more.

VIII.F.3.d. `Is that ok?`

Answer **y** and the new databases are installed.

You are now presented with the `Y-haplotype maintenance screen`. See §VIII.F.4.

**VIII.F.4.** *Rename, reorder, delete Y databases*

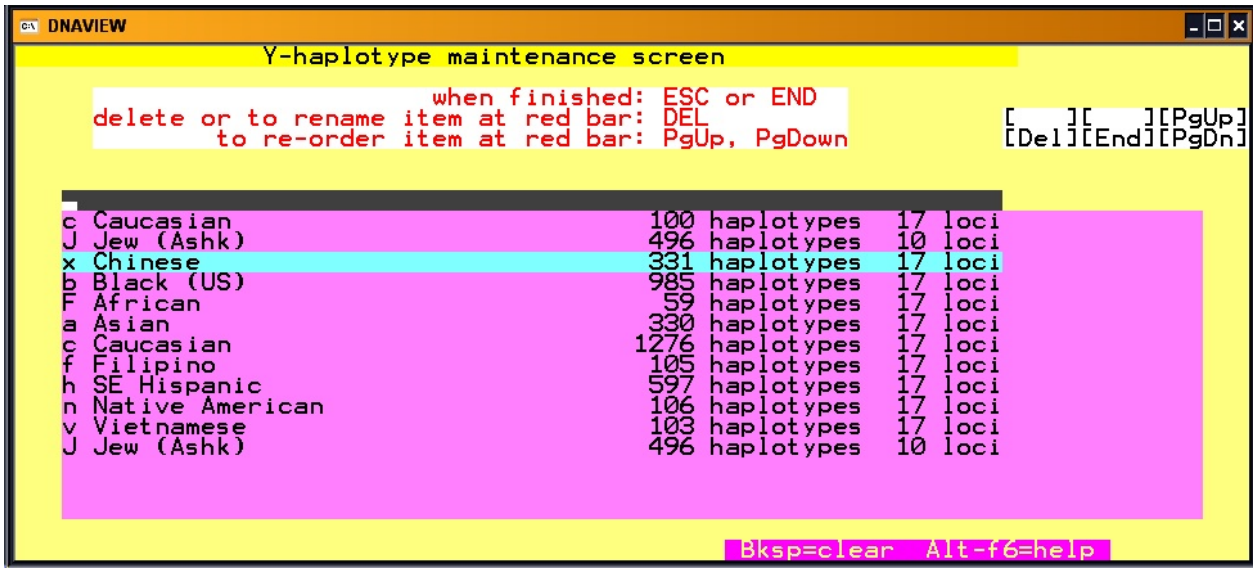



**Figure 85** Y-haplotype maintenance screen

The menu lists the installed Y-haplotype databases. Position the bouncebar to highlight an item and:

VIII.F.4.a.i.) *Reorder* the highlit item using MOVE↑, MOVE↓, ↑↑TOP, and/or ↓↓BOTTOM.

If there are two databases associated with the same race letter, then a calculation for that race in *immigration/kinship* will use the earlier one. Therefore the order can be important.

VIII.F.4.a.ii.) DELETE to *delete* the highlit item. Then confirm YES at the prompt.

VIII.F.4.a.iii.) RENAME to *change the race letter* and/or *description* of the highlit item.

## VIII.F.5. *Y-haplotype database statistics*

Creates and displays a statistical summary report of the installed Y-databases.

The "Spectrum" section, including κ, refers to ideas explained in Brenner (2010).

```
Statistical analysis of installed
Y-databases
Black (US) (b)
  17 loci: YFiler
  Number of profiles       985
  Number of haplotypes     954
  Random match probability 1/921.4
  Power of discrimination  99.89%
  Spectrum
   925 singletons  1 4-tons
    28 doubletons
   Kappa (singleton proportion) 0.9391
   Inflation factor=1/(1-K)     16.42
African (F)
  17 loci: YFiler
  ...
```

## VIII.G. Exact Tests for Independence

This section is a background discussion for the programs which are described in the following sections.

Some forensic calculations — including paternity calculations — implicitly assume that alleles are randomly assorted through the population. DNA·VIEW includes commands implementing the statistical tests of Guo (1992) and Zaykin (1995) to test for random assortment.

Multiplication of paternity indices or of matching LRs between loci is valid only to the extent that the loci are independent. Independence might be violated if loci are physically linked or by reason of non-random mating.

To say independence between loci holds for an infinite population means that for each allele b at locus B and each allele c at locus C, b occurs at the same rate among people who have c as it does among all people. In practice it is necessary to test this hypothesis with only a finite sample of people, for whom some variation from the ideal proportions inevitably occurs because of sampling variation — i.e. chance. The question for a statistical test, then, is this: How likely is the observed variation to occur just because of sampling variation, even when the independence hypothesis holds?

VIII.G.1.a. Logic

Independence is a property of a population, but in practice we only test a sample (subset) of a population. Therefore, in typical statistical language, the test result can never be definitive, but only says whether there is evidence of non-independence based on the sample.

For major populations it is by now well established that independent assortment of alleles is close to true but not exactly true. In the early 1990's there was a debate in the forensic community – or perhaps a debate between the forensic and parts of the legal community. One side alleged that the populations must be in equilibrium so the product rule for DNA probabilities is accurate, the other side that the product rule may be far from true and traditional DNA probability calculations completely unfair and unjustified. I think it is now clear that the truth is in between: completely random assortment is a fiction, but real populations come close.

However, unfortunately there is no particular relationship between “passing” the exact test and sufficient accuracy of the product rule. Passing could and does simply mean not enough data was examined. Conversely, if we examined sufficiently large population samples – of 10000+ individuals would be my guess – then we would see clearcut statistical evidence of *non*-independence, even though the magnitude of the departure from independence is probably forensically unimportant.

In summary, these tests are less valuable for database “validation” purposes than I thought when I implemented them. They do still serve some limited purposes:

VIII.G.1.a.i.) Errors in compiling the data sometimes show up as vast departures from statistical independence. Hence, failure of these tests may point to systemic errors.

VIII.G.1.a.ii.) When loci are quite close – e.g. when one is evaluating potential new forensic loci such as SNP sites – there can still be a real question of independence or not.

VIII.G.1.a.iii.) The forensic literature is still adding to the large volume of database papers using pointless “validation” methods, and you can check them or imitate them if you like.

VIII.G.1.b. Exact test for independence of loci

The so-called “exact test” for independence of loci is described in Zaykin et al (1995). As described in the paper, the following choices are allowed for consideration of each locus:

VIII.G.1.b.i.) assuming that the sample will necessarily have the same allele counts as this sample does, what is the probability $\boldsymbol{p}$ to observe exactly this set of (multi-locus) genotypes?

VIII.G.1.b.ii.) assuming that the sample will necessarily include the same numbers of each genotype, locus by locus, as this sample does, what is the probability ***p*** to observe exactly this set of multi-locus genotypes?

VIII.G.1.b.iii.) a hybrid — condition §VIII.G.1.b.i for some loci, and condition §VIII.G.1.b.ii for others.

The smaller the probability, the smaller will be the ***p***-value. The only trouble is, how to convert from a probability to a ***p***-value? It will depend on the specific collection of alleles and/or genotypes chosen. Therefore, the ***p***-value will be determined by Monte-Carlo experiments. Each Monte-Carlo trial consists of shuffling the sample data set randomly in one of two ways, depending on whether condition §VIII.G.1.b.i or §VIII.G.1.b.ii was chosen, for each locus. The probability of the shuffled data is calculated. After a thousand or so such Monte-Carlo experiments, the ***p***-value of the original data can be reasonable accurately estimated by noting where it's probability lies within the collection of 1000 random probabilities.

VIII.G.1.c. Exact test for Hardy-Weinberg equilibrium

The above method is a generalization of the method put forth in the seminal paper Guo & Thompson (1992) Performing the Exact Test of Hardy-Weinberg Proportion for Multiple Alleles, Biometrics 48:361-372. If you take the above method for the case of just one locus — in which case you necessarily use shuffling method §VIII.G.1.b.i — you have the Guo & Thompson method.

Hence the DNA·VIEW menu items *Independence of Loci* and *HW Exact Test* are the same program.

VIII.G.1.d. Independence on the Y-chromosome

The same method as in §VIII.G.1.b.ii can be further generalized to allow loci that occur on only one chromosome, or to treat a combination of loci as a haplotype. Of course, these situations occur mainly on the Y-chromosome.

## VIII.H. Hardy-Weinberg Exact Test; Independence of Loci

These command menu options are a stand-alone program that makes the special statistical computations discussed above. They are only useful for systems with discrete alleles and co-dominance, such as STR systems.

Also offered are some traditional statistical tables, which are useful for finding which particular allelic combinations are responsible for an appearance of non-independence.

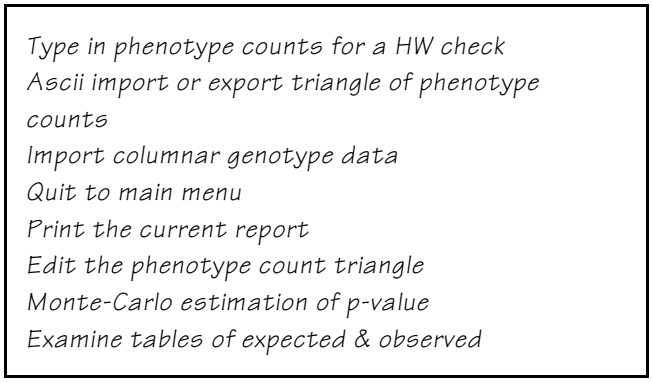



**Figure 86** Exact Test options

The program is operated through the menu shown in **Figure 86**. First you select one of the several menu options to enter data, then use some of the various options to analyze it. These steps add information to the report buffer — a sort of log of the analysis you have done. Whenever you like you can print the report buffer, or export it as a DOS file.

There are three ways to enter data. The first two are only for the HW test:

1. Type in a triangle of numbers representing genotype counts at some locus
2. Read such data from a text (“ASCII”) file.

Good for a HW test or a test of independence between loci:

3. Import data from a text file with multi-locus genotypes.

Having created a data set by one of the above methods, you can choose either or both of these methods of analysis:

1. Monte-Carlo

2. Tables showing various statistics between alleles.

The various menu options are described in more detail below.

```
     |  A     B     C     D
-----+-------------------------
  A  |  20
  B  |  7     4
  C  |  4     2     0
  D  |  12    4     1     7
-----+-------------------------
     |  63    21    7     31     allele count

        .516  .172  .057  .254   frequency
   122 alleles
Assuming co-dominant system in HW equilibrium:
 0.636  h=rate of heterozygosity
         =chance random alleles mismatch
 0.379  Expected exclusion rate for paternity trios (exact)
 0.811  Expected rate to exclude a random suspect from
         a stain sample
p-value & 1 std dev confidence interval:
p=  173.0 /  1712 = 0.10105  ( 0.09377 - 0.10834 )
```

**Figure 87** HW exact test

### VIII.H.1. Type in phenotype counts for a Hardy-Weinberg check

Select the first menu option from **Figure 86** to enter a triangular array of numbers such as **Figure 87**. These numbers are genotype counts of a population sample population data for a single polymorphic genetic locus with discrete co-dominant alleles, such as an STR locus. The triangular array is interpreted as if the rows and columns were each labeled with names of alleles (the same set of names for rows and for columns), and each entry is the number of people with a particular genotype. For example, among the 61 people represented in the figure are 7 BA people, 4 BB's, 2 CB's, and 4 DB's. These seventeen people account for 21 B alleles. The allele counts are updated as you enter the numbers, so they can be useful as check totals.

After you have entered observed phenotype data to your satisfaction, strike ***Esc***.

Total heterogyzosity and several other statistics will be calculated as shown in **Figure 87**. Then the Monte-Carlo run begins.

When you select this option the old report buffer is first discarded, so that the new triangle of data that you enter will appear at the top of the new report buffer. This is necessary in case you later want to save it and later rework it (VIII.H.3).

### VIII.H.2. Monte Carlo calculation

Monte Carlo trials are initiated by selecting the menu item *Monte-Carlo estimation of p-value*.

Each Monte Carlo trial consists of shuffling the set of genotypes that is the input data, computing the probability of both the original and the shuffled sets of genotypes and scoring 1 for a success every time the original data is more probable. The number of successes (ties count as ½) as a fraction of the total number of trials is displayed by running counters on the screen. In **Figure 87**, 173 successes have been scored out of 1712 trials, for a success rate of 0.10105. The numbers in parentheses represent the upper and lower 68% confidence interval around 0.10105.

The success rate is a direct computation of a ***p***-value. The interpretation of ***p*** is, "Assuming equilibrium, what fraction of random samples would be less likely than this one?" Hence if $p<0.05$ it is traditional to doubt the hypothesis. Conversely, if $p>0.95$ the data looks too good to be true. By definition though under equilibrium the values for ***p*** will be uniformly distributed, so every value is equally likely by chance.

VIII.H.2.a. Controlling the Monte Carlo run

Strike **n** to stop the Monte-Carlo run, or hit any key such as ***space*** to interrupt it whereupon you are asked *Resume Monte-Carlo run?* You may then:

VIII.H.2.a.i.) type **n** to return to the menu of **Figure 86**.

VIII.H.2.a.ii.) answer **y** to continue for a specific number of trials. When asked

`Do how many trials`, answer **1000** for example, and ***enter***. The trials continue.

VIII.H.2.a.iii.) If you wish, you may then type **q** before the trials are complete. Then the trials will automatically stop after 1000 without asking again the question at §VIII.H.2.a.ii. The advantage of this is that any further keystrokes you type are stacked and used to select from the exact test menu, etc, rather than being interpreted as a request to interrupt the trials.

### VIII.H.3. Ascii export, or import a triangle of phenotype counts

VIII.H.3.a. *Export* prompts you for a file name to which to save a text file of the report buffer, just as the *Save As* (§V.K.2) command from the **File** menu of the main menu.

VIII.H.3.b. *Import* allows you to retrieve to rework a triangle of numbers for a HW check that were previously saved in a named text file.

If you have previously created a triangle of phenotype counts for a HW check, then the triangle will be at the beginning of the report buffer and hence the file created will be of such a form.

### VIII.H.4. *Import columnar genotype data*

is the way to input data for a test of independence of loci.

VIII.H.4.a. Input file format

The menu option *Import columnar genotype data* expects an text file of the typical appearance as in **Figure 88**.

Columns are defined either by spaces or by tabs. The title row, if present, need not conform to the column separation though.

Since the title is optional, you will be prompted to answer whether it exists or not.

*D21S11 FIBRA is titles not data, right?*

Answer **y** if there is a title line.

```
                D21S11   FIBRA

D133/ 92   6    10  2    5
D134/ 92   4    15  1    2
D135/ 92   10   15  2    7
D136/ 92   8    15  1    7
E008/ 92   4    10  1    6
E009/ 92   4    10  4    6
E010/ 92   4    8   4    6
E011/ 92   10   13  5    8
```

title line (optional — may be omitted)

blank lines are ignored

each row=genotypes for a person

**Figure 88** Genotype file format

Alleles for a locus occupy a pair of consecutive columns. Additional columns don't hurt, but can be used if present to define a subset of the people in the file as a subpopulation, and to perform an analysis limited to that set. If the file doesn't fit on the screen, the program will attempt to squeeze it in by eliminating blank columns. Even if it still doesn't fit you are allowed to designate columns off the screen for analysis.

Since it takes some considerable number of seconds to read and parse a genotype file which you may wish to use for several analyses, for any import after the first you are asked

*Reuse same file?*

Answer **y** for instantaneous retrieval of the same file, or **n** to get another file.

The format rules are rather permissive:

VIII.H.4.a.i.) You can use any name (not containing spaces) you like for the alleles, and not just numbers.

VIII.H.4.a.ii.) The columns of data are separated by one or more columns of spaces.

VIII.H.4.a.iii.) The first row is interpreted as titles, and doesn't have to have spaces indicating column separation. Blank rows will be ignored.

VIII.H.4.a.iv.) Each person is represented by one row.

VIII.H.4.a.v.) Each locus is represented by a pair of consecutive columns.

VIII.H.4.a.vi.) Extra columns — e.g. for sample identification — won't hurt because you will always specify which columns you want to work with.

VIII.H.4.a.vii.) If the input includes the ***tab*** character, it will be resolved into spaces on the basis of tab stops arbitrarily far apart. In other words, each column can be either tab-delimited or space-delimited, but not a mixture.

VIII.H.4.b. Subpopulation and Locus Selection

As in **Figure 119** or **Figure 89** a portion of the input file is then displayed on the screen, columns indicated by vertical bands of color and labeled at the top with the letters *a, b, ...* . You are requested to make a selection of columns. There are three possible types of selection that you may make:

*Subpopulation selection:*

VIII.H.4.b.i.) Select a single column to examine a table of the data in that column, and optionally select a subset of the rows for subsequent analysis of any of the three column selection possibilities. Row selection is by entering a phrase or phrases which are used as restriction context. For example, given the sample file in **Figure 119**, selecting column *a* (the first one), and then phrase **D** selects just the rows that have a **D**. Enter two phrases as **D&00** to select the first six rows.

```
Subpopulation designated by which column? (1 or none)
Check independence between which columns? (4 or more)
 Hardy-Weinberg test for which columns? (designate 2)
Choose columns by the letters; space between loci:    cd ef

 a b    c  d    e  f    g  h    i  j

        csf     tpox    tho1    vwf
H  1,   10 12   08 08   06 08   14 18
H  2,   12 12   08 09   07 09   14 20
H  3,   12 12   08 08   07 08   17 18
H  4,   09 11                   17 18
H  8,   11 12           09 09   17 17
H 10,   12 13   08 10   06 07   14 16
```

**Figure 89** Subpopulation and locus selection

After selecting a subpopulation you will be asked to select again. If you wish, you may again select a subpopulation. But eventually you must choose one of the following two locus selection options.

*Note:* This selection option is also useful to examine the collection of symbols, and the number of each, in any given column. After that information is presented in a pop-up window, just press ***enter*** if you wish to keep all the rows.

VIII.H.4.b.ii.) Random subpopulation

For amusement or experiment, you may accept a random subset of rows by entering the character **?** in response to the `Choose columns by the letters` prompt. Given a very large sample of a population that is slightly out of equilibrium (as all of them are), the *p*-value will be nearly 0. The experiment is to see how the *p*-value grows as successively smaller sub-samples are examined.

*Locus selection:*

VIII.H.4.b.iii.) **HWE test** Select a pair columns, comprising one locus, to perform a Hardy-Weinberg check on the data for that locus.

In this case the program then counts the number of people with each pair of alleles, and constructs the genotype incidence triangle for you. You can subsequently edit it as in §VIII.H.1 if you wish.

VIII.H.4.b.iv.) **Independence between loci** Select two or more pairs of columns to perform an independence test among the specified loci. Put a space between pairs, as in the example **Figure 89**.

VIII.H.4.b.v.) **Independence and Y-loci** You may also specify a single column as a group, rather than a pair of columns as illustrated in **Figure 89**. This is appropriate if the column represents a locus on the Y chromosome.

```
Subpopulation designated by which column? (1 or none)
Check independence between which columns? (4 or more)
 Hardy-Weinberg test for which columns? (designate 2)
Choose columns by the letters; space between loci:   c e f

 a       b               c       d       e       f
sample   race            DYS389I S389II  DYS19   DYS390
R1       cauc            2       2       2       2
R2       cauc            2       2       2       2
R3       cauc            2       2       3       2
```

**Figure 90** Selecting Y loci

**Y haplotypes** It is even permitted to group several Y-loci together to be treated as a haplotype that will then be checked for linkage against some other locus or loci. However, in this case you must take special care so that the program doesn't confuse a haplotype specification with an ordinary 2-chromosome genotype as in §VIII.H.4.b.iv. The way to do this is to use a distinct set of allele names for each locus in the haplotype. Moreover, the allele names must be chosen in such a way that when they are all alphabetized or sorted numerically there will be no interleaving of the names from different loci.

VIII.H.4.c. Specify kind of test

In the situation §VIII.H.4.b.iv there is a further possibility as to exactly what is tested. So another prompt allows you to select, for each locus (pair of columns) which if any of the loci should be simultaneously tested for HW equilibrium. The method of choosing is illustrated in **Figure 88**.

There are two possible ways to consider each locus, as discussed in Zaykin et al (1995).

VIII.H.4.c.i.) *as a collection of alleles* — allelic shuffling, discussed in §VIII.G.1.b.i. This way, for each Monte-Carlo trial the alleles will be shuffled without regard to their original pairing. Such a test really tests for independence of loci and Hardy-Weinberg equilibrium at the same time. This is a good choice if the data for this locus appears to be in Hardy-Weinberg equilibrium. But if not, the HW disequilibrium will inevitably result in a low ***p***-value regardless of independence.

For allelic shuffling, include the column letter in the answer to

*Allelic shuffling at which loci?*

VIII.H.4.c.ii.) *as a collection of genotypes* — phenotypic shuffling, discussed in §VIII.G.1.b.ii. In this case, the rows for this locus will be shuffled for each Monte-Carlo trial, but the alleles will not be shuffled between people. Consequently any Hardy-Weinberg disequilibrium will be preserved so the test will be purely a test of independence between loci.

VIII.H.4.d. Summary statistics — *examine tables of expected and observed*

Further, you have a choice of several summary statistics (not Monte-Carlo or an exact test) for allelic combinations between the loci that the program will provide for you:

VIII.H.4.d.i.) odds against observing so many (or so few)

VIII.H.4.d.ii.) observed allelic associations (e.g. # people with A1 and B2)

VIII.H.4.d.iii.) expected numbers of each allelic association

VIII.H.4.d.iv.) excess of observed over expected = observed − expected

VIII.H.4.d.v.) relative excess = (observed−expected) / expected

VIII.H.4.d.vi.) approximate variation from expected, in standard deviations

= (observed−expected)² / max(expected,2)

This tables are only offered in the case of evaluating one locus (HW test) or comparing just two loci due to the difficulty of presenting 3-dimensional and larger tables.

The main purpose of these tables is to help analyze the data further if the Monte-Carlo test gives a small ***p***-value. If there are just a few allelic combinations that are out of kilter, which is often the case, you can find them with these tables. Also, note that these tables count *allelic* associations. There is no option for tables that count *genotypic* associations. Therefore these tables always relate to Monte-Carlo analysis per §VIII.H.4.c.i and are not affected by the choice of shuffling method.

You may examine or review the tables in any order you like from the menu, but as a default behavior the program steps through them for you. As you examine each table you are given the opportunity to add it to the printable report. If you examine a table twice, don't say **y** both times or it will appear twice on the report.

| | ***P*** | ***Q*** | ***R*** |
|---|---|---|---|
| ***P*** | 44 | | |
| ***Q*** | 9 | 3 | |
| ***R*** | 4 | 1 | 1 |

*genotype counts*

To discuss the meanings of the various tests, consider an example showing genotype counts for some 3-allele locus.

| | ***P*** | ***Q*** | ***R*** |
|---|---|---|---|
| ***P*** | 2.9 | | |
| ***Q*** | −4 | 2 | |
| ***R*** | −1.7 | 0.1 | 0.8 |

*excess observed*

The *p*-value is only 2%, which suggests that there may be a problem. I want to find the problem. From exact test menu choose *Examine tables of expected and observed*, under which is the further item §VIII.H.4.d.iv *Excess observed over expected*. The excess (=observed−expected) is at the right.

There are 2.9 more ***PP***'s observed (44) than expected (41.1). Is that a problem? How about the ratio, §VIII.H.4.d.v *Relative excess of observed over expected* (= observed/expected − 1), below.

| | ***P*** | ***Q*** | ***R*** |
|---|---|---|---|
| ***P*** | 0.1 | | |
| ***Q*** | −0.3 | 1.9 | |
| ***R*** | −0.3 | 0.1 | 4.1 |

*relative excess observed*

Now the biggest value corresponds to ***RR***. Which statistic is more relevant?

| | ***P*** | ***Q*** | ***R*** |
|---|---|---|---|
| ***P*** | 3 | | |
| ***Q*** | −6 | 10 | |
| ***R*** | −2 | . | 5 |

*odds against observation*

I think the best spot-check statistic here, the nearly ultimate statistic, is §VIII.H.4.d.i *Odds against observing so many (or so few)*, which I invented for this purpose:

This shows that ***QQ*** is the worst culprit, since it is 10:1 against observing such a large excess of ***QQ*** genotypes by chance from a population in equilibrium. (It is 6:1 against observing *so few* ***PQ***'s. The minus sign doesn't have numerical significance. It's rather a code meaning that the observed number is smaller than expected.)

## VIII.I. *Database similarity test*

**Use:** "Validate" a database.

A reasonable assessment of database validity for a known population is by comparison with another sample from the same population.

This command implements an extension that I developed of Fisher's (original) exact test. Weir (1996 – Genetic Data Analysis II) discusses the use of Fisher's test to compare several population samples for the same locus and (ostensibly) the same race. The result of the test is a *p*-value that represents the chance that such differences as there are would occur by chance if in fact the null hypothesis – the hypothesis that all samples are drawn randomly from the same population – is true. Otherwise put, if the *p*-value is very low, that indicates statistically significant variations in allele frequencies – different populations.

My generalization is to permit simultaneous evaluations of several loci, with only a single *p*-value representing the result of all comparisons. I am not aware of a publication discussing this particular extension, but it is a straightforward probability test.

The raft of papers that pretend to validate databases (genotype population samples) by testing for Hardy-Weinberg equilibrium (§VIII.G) could be improved by using this test instead. At least it says something meaningful.

### VIII.I.1. Outline of use

You have databases for some population for multiple loci, and "reference" databases for the same loci from previous population studies. The reference databases need not be the same for each locus but usually they will be.

The ultimate goal is single statistic (§VIII.I.2.b) which says whether there is a significant difference among the various population samples. For example, if the single overall $p$=0.3, that means that a fat healthy 30% of the time that the conglomeration of the given samples were re-apportioned at random into separate samples, the inter-sample differences would be at least as large as the observed differences among the real data. The implicit conclusion is that since the observed differences would occur quite often by chance, it is believable that they in fact did occur by chance (i.e. by "sampling variation").

Sometimes, the single overall *p*-value does not turn out to be small as hoped, but rather $p$=0. In that case it might be interesting to see if some particular locus or loci are mostly responsible – are problem loci. See §VIII.I.2.d and also §VIII.I.2.c. If the populations are as disparate as different traditional races then probably $p$=0 for every locus individually, but for clearly related populations such as Caucasians and Ashkenazy Jews, or Japanese and Koreans, there may be definite differences but only sporadic ones.

As you proceed with analyses, results of your operations are added to a report which you may afterwards print etc (§V.K)

### VIII.I.2. Operation

The *Database similarity test* repeatedly offers a menu with a set of options of in order to let you add to, subtract from, or analyze a collection of databases to compare.

VIII.I.2.a. *Add a set of databases for another locus*

This option is automatically "chosen" for you when you begin the analysis.

VIII.I.2.a.i.) Choose a locus . The locus list will not include any loci that you have already selected databases for, so usually you can just choose the top on the list, and usually that one is ready to be chosen by simply hitting ***enter***.

VIII.I.2.a.ii.) Select databases. Use the "mass selection" method as described in §XIV.A.3. Since you will typically want to chose similarly named databases for each locus, it is quite helpful to realize that search phrases you previously typed are available from the response memory (§XIV.A.4.a.vi) so need not be retyped and can be recalled from the type-in history with the ***up-arrow***.

The selected databases will be documented on the report.

VIII.I.2.b. *Compute unified p-value across all loci*

This is the bottom-line computation. You can wait until you have accumulated all the databases and loci to perform it, or if a little impatient you may see how it works on just one or a few loci.

Control of the Monte Carlo trials uses the same method as discussed in §VIII.H.2.a.

VIII.I.2.c. *Compute the p-value for one locus*

Select one locus among those for which you have already selected databases (§VIII.I.2.a). Initiate Monte Carlo trials to determine the *p*-value comparing just those databases.

VIII.I.2.d. *Compute the individual p-valueS for each locus*

iterates automatically through all the loci per §VIII.I.2.c. First, the program asks

`How many trials?` and you must answer a number. **1000** may be enough.

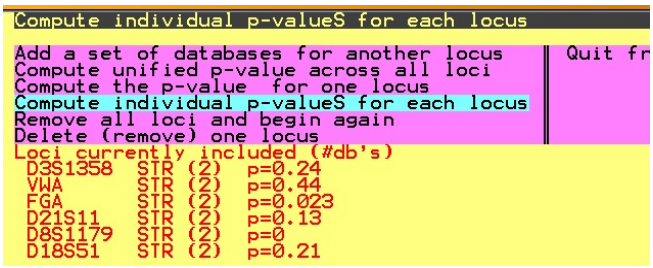



**Figure 91** similarity *p*-values for each locus

VIII.I.2.e. *Remove all loci and begin again*

Begin comparison of new sets of databases

VIII.I.2.f. *Delete (remove) one locus* – in effect, back up a step.

VIII.I.2.g. *Show report* – view and optionally print or save a report of the analysis to date.

VIII.I.2.h. *Quit from the Database similarity test* – which invokes §VIII.I.2.g on the way out the door.

## VIII.J. *Allele report* – rare and common alleles

The *Allele report* command, in the **Population** menu, lists all occurrences of sufficiently rare alleles for all (or a selected subset) of the available databases for one locus at a time. It also enumerates the popular and infrequent alleles:

**D3S1358.** Rare alleles (frequency at most 1/1000 among all 12 databases): 9 11 15·2 15·3 21
common alleles: 15–31% 16–29% 17–21% 18–9% 14–8%
other alleles: 12–0.1% 13–0.5% 19–0.7% 20–0.1%
Allele **9** observed 1/5276

| | | | | |
|---|---|---|---|---|
| **9** | 1/390 | ABI Black | 195 | 97/08/01 9am |

Allele **11** observed 4/5276 = 1/1300, in 3/12 databases.

| | | | | | |
|---|---|---|---|---|---|
| **11** | 1/390 | ABI Black | 195 | 97/08/01 9am | |
| **11** | 1/400 | ABI Caucasian | 200 | 97/08/01 9am | |
| **11** | 2/420 | FSC–Jul99 Black | 210 | 01/07/06 11am | etc. |

## VIII.K. Mutation history

**Use:** Estimate mutation rates and cull likely mutation cases

### VIII.K.1. Overview

This command reviews the casework history to estimate paternal mutation rates. This command is extremely useful for labs wanting to conform to AABB rules, or for research.

The output gives both a summary table of apparent mutation rates – abbreviated form:

| Locus | | est mut'ns per 1000 | cases with 0 or 1 "exclsn" | poss mut'ns |
|---|---|---|---|---|
| PAC425 | Hae | 11 | 1452 | 16 |
| D3S1358 | PCR | 1.8 | 3944 | 7 |
| vWA | PCR | 2.8 | 3627 | 10 |
| FGA | PCR | 5.8 | 3425 | 20 |
| D8S1179 | PCR | 3.8 | 3422 | 13 |

and a detail, showing every suspected mutation, e.g.:

```
(108) Case 12345-10 Residual PI=2,000,000,000 from 13 consistent loci.
Inconsistencies:
   D3S1358 (b)  M=  16  Ch=  18 16  Af=  16 17
(109) Case 12567 Residual PI=100,000,000 from 12 consistent loci. Inconsistencies:
   D3S1358 (c)  M=  15 17  Ch=  17 15  Af=  16
(192) Case 995432 Residual PI=400,000,000,000 from 17 consistent loci.
Inconsistencies:
   D18S51    PCR (b)  M=  Ch=  13 16  Af=  12 15
   D3S1358   PCR (b)           16 17       14 15
   D8S1179   PCR (b)           14 15       11 12
(193) Case 994433 Residual PI=500,000,000 from 16 consistent loci. Inconsistencies:
   TBQ7      Hae (b)  M=  Ch=  4132 1724  Af=  2472 5246
   PAC425    Hae (b)           3920 1523       703  3089
```

**Figure 92** Suspected mutations

which gives the information that you need to refine the summary – such as by excluding cases that are really primer dropout, or that are non-paternity, such as uncle cases.

### VIII.K.2. Preliminaries for mutation analysis

The mutation data is extracted from the table DNAAPPRO (§XVI.B.3), which is a communication table between DNA·VIEW and PATER (for paternity cases computed in DNA·VIEW and reported by PATER) that doubles as an archive of case calculations. All cases analyzed by *Paternity Case* will in the future be available for analysis by *Mutation history*.

VIII.K.2.a.i.) Archive old cases.

However, earlier versions of DNA·VIEW didn't necessarily archive cases to DNAAPPRO if you were not using PATER. In order to make *old* cases available for analysis, now re-run them (*Paternity case*, *calculate paternity/maternity*) in DNA·VIEW. For a moderate number (<100) of old cases, it is reasonably easy using the *batch select* and *batch run* options of *Paternity case*. For a larger number, you may need special help (request by phone or email).

### VIII.K.3. Mutation report

The report includes a fair amount of boilerplate explaining what the various computations represent, and two sections of data – a table of summary statistics, and a list of relevant cases with some details.

VIII.K.3.a.i.) Mutation report summary table (**Figure 93**)

The table consists of one row per locus, and various columns. The numbers in the table count cases for which the given locus was used, and which also satisfy one or another additional criterion:

» `Total cases; all` gives the total number of cases (paternity or maternity) for which a given locus was used. Hence this counts all cases, inclusion or exclusion.

» `Total cases; =0` means the number of cases for which every locus is consistent with paternity. This number figures to be a slight underestimate of the number of true paternities tested with this locus.

» `Total cases; <=1` means the number of cases that had 0 or 1 loci inconsistent with paternity. (So-called "exclusion" loci.) At least as a first approximation, this number estimates the number of true paternities that were tested with this locus.

» `Total cases; <=2` is the somewhat larger number which also includes cases with 2 "exclusions".

» `Possible mutation; =1` is the number of cases that had a single inconsistency and that inconsistency was in this locus. As a first approximation, this number is the number of mutations for this locus.

» `Possible mutation; =2` is the number of cases that had a two inconsistencies, one of them in this locus. These cases should be examined individually as possible additional mutations.

| | | Est mutn per1000 | Total Cases =0 | <=1 | <=2 | <=3 | all | Poss Mutn =1 | =2 | =3 | >3 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| pYNH24 | Hae | 1.7 | 10162 | 10278 | 11192 | 14359 | 14916 | 17 | 593 | 3092 | 514 |
| EFD52 | Hae | 3.5 | 8933 | 9052 | 9809 | 12456 | 12978 | 32 | 420 | 2579 | 479 |
| pH30 | Hae | 32 | 442 | 538 | 569 | 616 | 720 | 17 | 19 | 32 | 78 |
| D3S1358 | PCR | 1.8 | 3822 | 3918 | 3944 | 4107 | 5331 | 7 | 6 | 36 | 661 |
| vWA | PCR | 2.8 | 3532 | 3627 | 3653 | 3797 | 5019 | 10 | 5 | 47 | 727 |
| FGA | PCR | 5.8 | 3331 | 3425 | 3443 | 3565 | 4721 | 20 | 2 | 48 | 865 |
| D8S1179 | PCR | 3.8 | 3329 | 3422 | 3442 | 3563 | 4731 | 13 | 5 | 44 | 755 |
| D21S11 | PCR | | 3345 | 3440 | 3460 | 3587 | 4760 | . | 2 | 48 | 816 |
| D18S51 | PCR | 3.3 | 3270 | 3363 | 3382 | 3506 | 4659 | 11 | 7 | 62 | 884 |
| D5S818 | PCR | 0.82 | 3555 | 3651 | 3677 | 3819 | 5037 | 3 | 5 | 26 | 603 |
| D13S317 | PCR | 1.6 | 3544 | 3640 | 3667 | 3811 | 5030 | 6 | 5 | 35 | 707 |
| D7S820 | PCR | 2.2 | 3997 | 4096 | 4128 | 4302 | 5575 | 9 | 3 | 56 | 756 |
| D16S539 | PCR | 4.3 | 854 | 939 | 961 | 1030 | 1213 | 4 | 2 | 21 | 98 |
| TH01 | PCR | | 823 | 907 | 928 | 995 | 1180 | . | . | 19 | 81 |
| TPOX | PCR | | 862 | 946 | 968 | 1039 | 1228 | . | 4 | 16 | 79 |
| CSF1PO | PCR | 3.3 | 820 | 904 | 926 | 993 | 1174 | 3 | 6 | 14 | 96 |

**Figure 93** Mutation History summary statistics

» **`Estimated mutations per 1000`**

This is the important statistic. It is computed on the basis of paternity (or maternity) cases that have one inconsistent locus (a so-called "exclusion"). It is the ratio

(`Possible mutation; =1`) / (`Total cases; <=1`)

This estimate can be refined by reviewing the "suspicious cases."

VIII.K.3.a.ii.) Suspicious case table (**13**)

All suspected mutations are listed. Cases are included if

- » there were exactly 1 or 2 or 3 inconsistent loci, and
- » the residual PI>100.

Only the inconsistent loci are shown. Grossly illegal data (base pairs<0) are removed first. However, some PCR types may be shown as a ?. If so, then the locus is re-listed ("again:") with the base pair sizes.

Review the instances of possible mutation. This table is intended to be useful for sorting them out, but obviously the best determination requires researching other records and finally, making some sort of judgement.

## VIII.L. Racial "Discrimination"

Two commands/facilities consider racial origin in the light of DNA data.

» *Racial Distances* (§VIII.M) is a population genetic research idea. It attacks questions like, "How useful is DNA to distinguish races? How disparate are the races?"

» *Racial Estimation* (§VIII.N) is an investigative tool that analyzes a particular DNA profile.

For the theoretical underpinning of the calculations, see Brenner (1997c, 1998, 2006).

## VIII.M. *Racial distances* – compare many races

A DNA profile will have a different probability of occurrence in different racial or ethnic groups. Usually the group in which it is most probable is the actual group of origin. On average how much less probable is the profile from a "wrong" group than from the right one?

### VIII.M.1. Uses

The answer to this question can be interpreted in several useful ways:

VIII.M.1.a. typical error from using the wrong population

VIII.M.1.b. a new measure of racial distance

VIII.M.1.c. prediction of the ability to guess the race of a stain donor

### VIII.M.2. Background

The DNA·VIEW *Racial distances* command (**Research ideas** menu) uses available population databases

Racial distance Using 15 loci Identifiler

Typical (geometric mean) likelihood ratio supporting the true origin (row) of a DNA profile, versus an alternative origin (column).

| | Iraqi | Korean | Caucasian | Egygin (Mongol) | Brahmin Indian |
|---|---|---|---|---|---|
| Iraqi | 1 | 13 | 2.2 | 5.6 | 900 |
| Korean | 12 | 1 | 200 | 1.6 | 5000 |
| Caucasian | 2.4 | 200 | 1 | 300 | 3000 |
| Egygin(Mongol) | 19 | 2.7 | 500 | 1 | 500e6 |
| Brahmin Indian | 700 | 20000 | 20000 | 100e6 | 1 |

Calculation uses population studies for all available common loci betweeneach two populations.

**Figure 94** Distance chart between races

to estimate the distances among a set of races that the user specifies. Of course the answer depends on which loci are considered; any desired set of loci may be specified.

### VIII.M.3. Operation

VIII.M.3.a. Choose the loci to consider

You are first offered the "multiplex" menu (§IX.B.7) in order to select a set of loci. Note that selecting a single locus is among the possible choices.

VIII.M.3.b. First selection races to compare

From the chart, which shows all the races for which data on some of the selected loci is available (the number in parentheses is the number of available loci), choose any set of races by typing the race letters.

To consider all the mentioned races, just ***enter***.

VIII.M.3.c. All pair-wise racial distance are then calculated using the formulas described in Brenner (1997c) and (1998). Where fewer than the maximum number of loci are available for a comparisons between two loci, the distance determined is scaled up so that all numbers in the table are approximately comparable.

VIII.M.3.d. The result report is displayed on the screen and may be printed (via *Print*).

## VIII.N. *Racial estimation*

improved The *Racial estimation* command (in the **Casework** menu) computes the likelihood to see a given DNA profiles in each of several races. Comparing these likelihoods gives the likelihood ratios supporting each race compared to the others. Also, the command permits calculating all the profiles of a case at once, rather than just one individual. The command is also available in the *Immigration/Kinship* menu.

### VIII.N.1. Initiate operation

VIII.N.1.a. (as a main menu command)

VIII.N.1.a.i.) Choose **Casework** ▸ *Racial estimation*.

VIII.N.1.a.ii.) `What collection of people? (Person/Case/Worklist)` **`Case`**

VIII.N.1.a.iii.) Enter a case number or person number, or select a worklist as the case may be

VIII.N.1.b. (as an *Immigration/Kinship* option)

VIII.N.1.b.i.) Choose **Casework** ▸ *Automatic Kinship* (or other *Case* command).

VIII.N.1.b.ii.) *Select case* – choose the case

VIII.N.1.b.iii.) Choose *Immigration/Kinship* ▸ *Racial Estimate*

### VIII.N.2. Tentatively choose races to compare

A chart lists all races for which allele frequency databases are defined as defaults for at least part of the profile(s). The numbers in parentheses are the numbers of loci for which default databases are specified for a given race. The suggested default response is a list of those race letters for which nearly complete (according to a heuristic measure) data is available. `Races of possible interest?` **`cEkqR`**

VIII.N.2.a. database "availability"

For purposes of the *Racial estimate* command, the databases that are considered are those that are marked as "default" (§VIII.C.2.b, *database, per race*).

VIII.N.2.b. Choose any selection of races.

Type in the race letters of interest then ***enter***, or just ***enter*** to accept the suggested list.

### VIII.N.3. Consider racial mixtures?

There are two possible kinds of racial mixture – one parent of each racenew, or a blended ethnic history. **Yes** for both kinds of analysis to be included in the result table; **no** for neither, just the "pure" races.

### VIII.N.4. Choose races to compare


**Figure 95** Choose races to compare

There is no guarantee that the loci are the same when the numbers are the same for two different races, nor that a larger number of loci includes a smaller number as a subset. Therefore, the program may offer a menu (**Figure 95**) showing all combinations of races that can be compared with the same loci, and the number of loci for each such combination.

### VIII.N.5. Result

Case 0110 ancient Mongols
Relative support for each racial origin

| | W-S10 from Keyser 2009 | D-degraded. male | B-female | C-"European" male |
|---|---|---|---|---|
| Egygin(Mongol) | 1600 | 84 | 110 | 3.2 |
| Iraqi | 6700 | 41 | 53 | 3 |
| Korean | 16000 | 50 | 1 | 1.6 |
| Caucasian | 500 | 41 | 4 | 1 |
| Brahmin Indian | 1 | 1 | 2.1 | 4.7 |

new The result is displayed on the screen and can be printed (*Print*) or filed (**File** menu) and conveniently imported into Excel (since it's tab-delimited).

Calculations are made for the selected races, and also for mixtures if requested (see above). Only the several most plausible mixtures, not all of them, are included in the chart.

Note that numbers are comparable within each column but not between columns.

# IX. MISCELLANEOUS OPERATIONS

## IX.A. *Maintenance*



**Figure 96** *Maintenance* options

The *Maintenance* command brings up the menu shown in **Figure 96**, whereupon you may choose from among the listed options, which are described individually below.

### IX.A.1. *Locus parameters & preferences*

Locus/probe maintenance is a large topic in DNA·VIEW. Also, the facilities for locus maintenance are accessible not only from within the *Maintenance* command, but also any time the probe/locus list appears. For this reason it is convenient to put the description of the facilities in a section (§IX.B below) by itself.

### IX.A.2. *Locus report*

DNA·VIEW creates a report listing the loci and all locus parameters, as illustrated in **Figure 97**, which can be printed using *Print*. A legend at the bottom of the parameters describes the parameter columns that are present.

```
[127] D3S1358   3p         STR      0=66 r=4                              m=0.2
[70]  VWA       12p13.3    STR      0=83 r=4 y=199 a=147                  m=0.3
[14]  FGA       4q         STR     0=146 r=4 y=302 a=215     s=7.14      m=0.29
[90]  Amelogenin           STR     0=101 r=6             P=2
[153] Penta D   21q        STR     0=368 r=5                             m=0.1
[154] Penta E   15q        STR     0=148 r=5                             m=0.14
[8]   TBQ7      D10S28     Hae III            δ=3 σ=0.7 g=0.2 r=2 l=2.5 m=0.07
[6]   pYNH24    D2S44      Hae III            δ=3 σ=0.7 g=0.2 r=2 l=2.5 m=0.09

Columns are:
 PCR parameters
  0=Size of allele type #0, base pairs
  r=Repeat unit, base pairs
  y=When multiplexed, maximum size
  a=When multiplexed, minimum size
  P=Notation (0=STR,DQa  1=PM  2=Amelo  3=SNP)
  s=Typical stutter (in %)
 Single locus parameters
  δ=Match window (±), % of molecular size
  σ=Interassay coeff of variation, % of molecular size
  g=Gjertson's åg, % of molecular size
  r=Inter-read match window (±), % of molecular size
  l=Inter-lane match window (±), % of molecular size
 Miscellaneous parameters
  m=Paternal mutation rate:  rate of false exclusions (%)
```

**Figure 97** Locus report

A further section of the report documents the Allele Resolution Filter (obsolete) definitions.

**IX.A.3.** *locus list by chromosome*

```
Str sNp Poly, Other, All loci?
                        (S,N,P,O,A) S
```

```
D5    [84]   CSF1PO     5q33-34     STR
      [128]  D5S818                 STR
D6    [68]   SE33        D6S        STR
      [71]   F13A1      6p25-24     STR
      [134]  DHFRP2                 STR
      [149]  DRB1       6p21.3      STR
      [155]  Penta F    6q          STR
D7    [130]  D7S820     7q11        STR
      [131]  IL-6       7p15-p21    STR
      [151]  Penta B    7q          STR
D8    [88]   LPL        8p22        STR
      [132]  D8S306                 STR
      [133]  D8S639                 STR
      [139]  D8S1179                STR
      [150]  Penta A    8p          STR
      [163]  D8S320                 STR
      [193]  D8S1132                STR
```

**Figure 98** (partial) locus list by chromosome

### IX.A.4. Races

The 52 lower and upper case letters, **a-z**, **A-Z**, are used in coding races. You may attach any spelling (i.e. race name) you like to each one. See **Figure 99**.

In addition, the ***dash*** (**-**) is used instead of a race letter coding children (since their race is irrelevant for calculations, and their alleles shouldn't go into any data base). The dash may also be used to code any other person who, for whatever reason, will be omitted from all data bases.

§XIV.A.9.a.iii has more information on entering race codes.

IX.A.4.a. *race codings (change)*

The list of races may be edited from this *Maintenance* option, but it is often more convenient method to access the list by answering **?** as a race code any place a race code is to be supplied — e.g. from *Case* or from *Worklist*.


**Figure 99** race codings

To change the race spelling associated with one of the race letters:

- From the green menu of races, put the red bounce bar on the desired item – for example by typing the race letter.
- Select the item by hitting ***tab*** (*not* ***enter***!).
- Type the new name into the answer box, then ***enter***.

IX.A.4.b. *race codings (list)*

Either by selecting this option, or after using the previous one, a report is prepared listing all the race codes and their corresponding names. It is available via *Print*.

### IX.A.5. Readers

Some DNA·VIEW operations require the user to select a user identity from the list of *readers*. Each reader is a 3-digit number and a name.

Two numbers are special:

» 000 is the "student" user. DNA data provided by user 000 is ignored for computation if other data (same lane, same locus) is available. However, if the student's data is the only available data, it is used.

» 099 is assumed to be a mechanical and hence infallible user. This number can be associated with data imported from another computer (Genotyper for example). Except for user 099, data that is entered only once is marked with an asterisk (*) on reports warning that it may be unreliable.

To modify the list of readers,

IX.A.5.a. obtain the list of readers

Select *readers* by typing type **read** ***Space*** from the *Maintenance* menu. Or, the list appears as part of various input operations such as *Type in a Read* (§IV.D.4). Therefore you can add or change a reader on-the-fly.

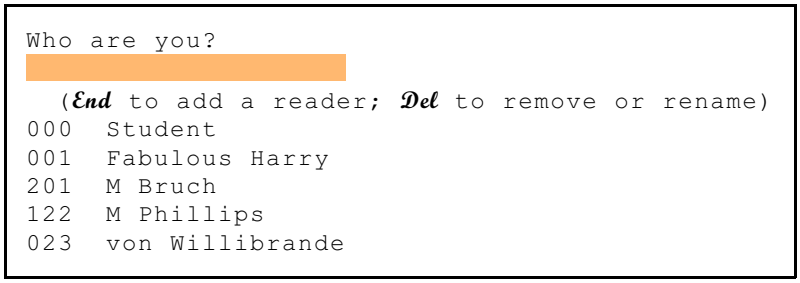



**Figure 100** enrolling a reader

IX.A.5.b. add a reader

Strike the ***end*** key. You are then prompted for the new reader number, with a new sequential number suggested as a default choice

```
Number of the new reader 7
```

***Enter*** to accept the number, or enter the number of your choice. At the next prompt, enter any name

```
Name for 007 Bond, James Bond
```

IX.A.5.c. change a reader name

Two ways: Either ***end*** as if to add a reader (§IX.A.5.b, and then give number of the reader to change. *Or* use the ***del*** key method explained in §IX.A.5.d.

IX.A.5.d. delete or rename a reader

Move the red bounce bar to the reader to delete, and press ***del***. The prompt

```
Revise name, or delete the name to delete the reader
007  Bond, James Bond
```

To delete: Type ***space*** (which will clear the name) ***enter***.

To rename: Type the new name, then ***enter***.

IX.A.5.e. create a list of readers

To create a report listing the readers, obtain the list of readers through *Maintenance*. When you finish with maintenance operations, a list is created that can be printed with *Print*.

**IX.A.6. Worklist configuration/formats (add or list)**

**IX.A.7. standard ladder/molecular weight marker (report, add)**

Worklist configurations and standards ladders are necessary only if DNA·VIEW is used for allele sizing. They are unnecessary if DNA profiles are imported from a Genotyper file for example, with allele "calls" already made. Therefore these obsolescent sections have been removed. Consult a pre-2007 version of the manual if interested.

**IX.A.8.** *Network preferences (for Paradox)*

Installation issue. See §XV.D.

**IX.A.9.** *document Paradox table*

This option may be useful for programmers.

**IX.A.10.** *rebuild Paradox index*

See §XI.F.1.

**IX.A.11.** *report systemwide switch/parameter settings*

Prepare a report describing the state of your system — warning you of any unusual options in effect, etc. The report includes:

**[Environment]** Directories where various files are stored. Network system? PATER present? **[Printers]** Printer type. page format **[Options]** Case number format. Menu style. Decimal character. Report header information. Default date? PCR format. Numlock state. Off ladder allele policy. **[Parentage/Automatic mixture Options]** settings. Are posting options consistent? Mutation switch reasonable? **[Loci & databases]** Active locus list and order. Default database list. Loci without default. **[Special parameters]** Importing defaults. Kinship prior.

**IX.A.12.** *record switch/parameter settings*

The current settings for most DNA·VIEW parameters – locus information, options choices including *Case Options* such as probability calculation method, database defaults – are saved, with your initials. Subsequently every time the program starts it checks all the settings and warns of any discrepancies:

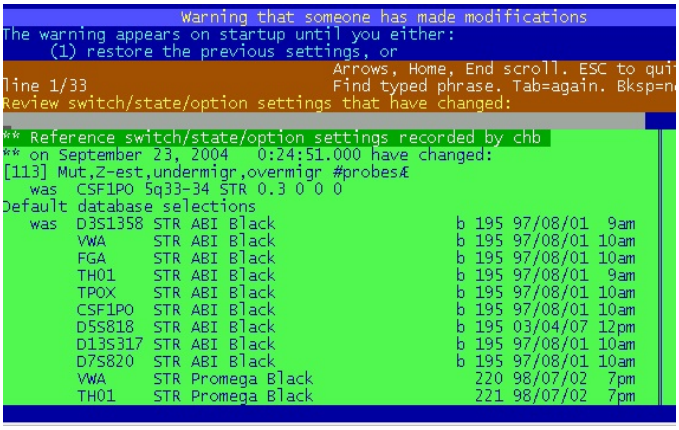



**Figure 101** Discrepancy report, current vs. standard settings

To get rid of the warning, either

IX.A.12.a. Restore all the old settings, using the warning report as a guide, or

IX.A.12.b. Record the new settings.

IX.A.12.c. Disable the feature.

Invoke *record switch/parameter settings*.
***Click*** UPDATE (CHANGE) OR REMOVE.
What is your name? (Your name)
What action? TURN OFF THIS FEATURE.

As an alternative or adjunct security feature you may consider passwords (§IX.A.15).

**IX.A.13.** *compare switch/parameter settings*

looks for any switches that have changed compared to the recorded values. For any ones that have changed, it shows you the *old, recorded* value, which is the information that you need in order to change it back. (If you want to see the new, current values, use *report systemwide switch/parameter settings* (§IX.A.11).

If there are no differences then *Compare Switch* makes no response. If there are differences, then they are shown on the screen and the screen is frozen until you hit a key from the keyboard.

*Compare Switch* becomes active only after the first time you execute *Record Switch*. Thereafter it executes automatically every time you start DNA·VIEW.

### IX.A.14. *rewrite (compress) files* – conserve disk space

Sometimes some DNA·VIEW files accumulate unnecessary dead space. Invoking *rewrite files* displays a table of wasted space ("slack"). Select a file from the menu of files and ***enter*** to rewrite the file and recover the space. Or abort (***ctrl-C***) to do nothing.

Compressing is generally harmless except for the time that may be involved (compressing a 100Mbyte file on a network might take 5-10 minutes).

**Caveat**: If you have a networked DNA·VIEW system, every user but one (including PATER users) must sign off before you use this tool because you can't compress a file while it is being shared.[15]

A list is shown of all DNA·VIEW files. For each file, the actual disk space consumed (SIZE) and the amount of wasted space (SLACK) is shown. The default selection is the one with the largest amount of slack. Select a file to compress it. You will be asked to wait — even minutes for a file of several megabytes — during the compression.

Interrupting compression might cause a minor problem. Erasing any files found on the hard disk with the extension `.SFD`, such as `DNAFREQ.SFD`, will cure the problem.

### IX.A.15. Password access to critical DNA·VIEW functions

*Make or modify the password permissions* is an optional security feature to prevent unauthorized changes to critical DNA·VIEW parameters and settings by requiring a password. Alternatively or in conjuction you can use the *Record switch/parameter settings mechanism* (§IX.A.12) which warns if any settings are changed.

The passwords are stored in a special file. If the file has some password records, then various DNA·VIEW operations require password entry. If the file is empty or non-existent, then password barriers are not used.

There are three security levels.

| security level | description | privilige |
|---|---|---|
| 1 (highest) | supervisor | any operation, including delete databases or cases, update to new DNA·VIEW system, modify locus names, record switch settings |
| 10 | user | casework, change some locus parameters |
| 20 | student | casework |

The password maintenance screens allow adding or removing passwords, and are self-explanatory.

[15] File #29 is not shared; it is private to each station. So you can compress it anytime.

## IX.B. Probe/locus maintenance

DNA·VIEW has hundreds of loci already included including all commonly used forensic loci as of this writing (**Figure 156**, **Figure 157** and **Figure 158**, §XVI.A.2). The list is uniform across all users in the sense that everyone should use the same line numbers — for example, VWA is locus [70]. This is to facilitate sharing of databases among users. For the same reason, some probes, such as pYNH24, are listed several times representing the various commonly used restriction enzymes.

At the same time, the list is completely user-maintainable. You can freely change the spelling (e.g. VWF instead of VWA if you prefer), the values of the various numeric parameters associated with any locus, or even to add a new locus. However, to maintain world-wide uniformity *please check with DNA·VIEW international headquarters before adding a new locus*, and then use the line number that is issued for your new locus.

Exception: Lines [100]–[119] are reserved and available for "Site use" — anything you like, but intended for usage that you expect will not be of interest to the user community.

Locus maintenance is allowed from every screen that shows the probe/locus list. There are many incidental situations in which this happens, such as when the user needs to choose a locus to *Type in a read*.

### IX.B.1. Locus maintenance

There are several useful actions to take with the locus list, including selection, reordering, adding, hiding, and setting properties.

IX.B.1.a. **Select a locus**

Move the bounce-bar to a locus in the locus list and OK or ***enter*** to select the locus.

IX.B.1.b. FULL LIST / SHORT LIST

These buttons toggles between a concise locus list that includes only "active" loci, and an extended list that includes all loci. The extended list also shows locus number (in the DNA·VIEW list), and a comment field (not user-changeable) of various information such as alternate names, Genebank information, PCR parameter possibilities.

If the desired locus is not found in the list, it may be necessary to add it (§IX.B.1.e) but more likely it is merely hidden (inactive). To check if it is in the list but hidden look at the FULL LIST. Then, because of the select-context mode of DNA·VIEW menus, typing almost any variation or part of the locus name will bring it into view.

IX.B.1.c. (DE)ACTIVATE

In order to keep the locus list less cluttered, the (DE)ACTIVATE button lets you hide those majority of probes/loci that are not of interest. Hidden means the locus is not visible from the SHORT LIST. It is visible but indented one character on the FULL LIST (§IX.B.1.b).

Activation and deactivation are also possible wholesale using the multiplex mechanism (§IX.B.1.d.i).

IX.B.1.d. **Locus ordering** – the REORDER buttons

It is convenient to arrange the locus list with commoner loci toward the top, and also to have loci in the same order as they occur in reference documents. Therefore the user may specify the preferred order of loci. This order determines:

» the order of loci in the locus list

» the order in which they occur in reports, such as a paternity or kinship calculation

» the next locus provided as a default suggestion by *Database similarity test* (§VIII.I.2.a.i) or *Type in a Read* (§IV.D.4)

» the next loci introduced by the **#** key in *DNA odds* (§V.G), *Y haplotype matching LR* (§V.H) or *DNA exclusion* (§V.I)

There are two methods for ordering loci, wholesale and one-at-a-time.

IX.B.1.d.i.) Wholesale locus ordering

REORDER: MPLX pops up a list of multiplexes (§IX.B.7)

» Select a multiplex from the list and

PUT THESE AT TOP to activate and position the loci of the multiplex in standard order at the top of the locus list;

ALPHABETIZE AT TOP to activate and position the loci of the multiplex in alphabetized order at the top of the locus list;

UNORDER to deactivate the loci of the multiplex and move them to the bottom, non-priority part of the list.

» With no multiplex selected (no bouncebar – try the ____ button), ALPHABETIZE ALL puts all active loci in alphabetic order.

IX.B.1.d.ii.) Single locus ordering

Highlight a locus and

» ↕ and ↓ to adjust the position of a locus within the list.

» ↓↓ to remove a locus from the priority list.

IX.B.1.e. **Add a locus**

It is unlikely that you will need to add a new locus, as all the usual identification loci are already incorporated in DNA·VIEW. Please see §XVI.A.2. If you don't see a locus that might be expected, possibly it is present but temporarily hidden.

Therefore, before adding a locus,

IX.B.1.e.i.) Check if the locus is already present, but inactive, per §IX.B.1.b. If it is, you only need to confirm that the various parameters (i.e. "properties") are ok (§IX.B.3.f).

IX.B.1.e.ii.) Determine the proper line number for the new locus. See page 313, 314.

IX.B.1.e.iii.) The NEW button opens a dialogue to add a new locus.

Type the line number for the new locus, ***Enter***.

IX.B.1.e.iv.) PROPERTIES (with the new locus line number highlighted) to enter locus maintenance for the new number at the point of enzyme selection. Select **SNP** (or **PCR**, etc) as a pseudo-"enzyme".

IX.B.1.e.v.) Do each of the relevant maintenance operations, definitely including *edit locus name* and *assign enzyme*.

IX.B.1.e.vi.) Add probe/locus (keystroke instructions)

Here are the steps, keystroke by keystroke, for adding a new PCR locus D17S1290:

Select [**Housekeeping**], *Maintenance* from the command menu.

Select *locus parameters and preferences*

Type **D17S1290** to search if the locus already exists. If the locus was already defined at the time of the last DNA·VIEW update, this will find it. But assume not. Inquire from DNA·VIEW for the correct new line number. Assume it is 418.

IX.B.1.e.vii.) Add a new line if necessary

NEW. What number do you want to add? **418** ***enter***. (Now the locus list will include [418], and line 418 will be highlighted in red.)

PROPERTIES (This selects line 418 for locus maintenance, moves to the locus maintenance §IX.B.2) screen, and asks for the restriction enzyme.)

IX.B.1.e.viii.) Specify the enzyme: If presented with the menu of (pseuedo-)enzymes, select *STR* ***enter***.

IX.B.1.e.ix.) **Change the name** — Select *edit the locus name* from the locus maintenance menu, and ***enter***. Type **D17S1290** ***space space*** **17q** ***enter***.

IX.B.1.e.x.) **Set the PCR parameters** — Select *PCR Parameters* from the locus maintenance menu, and ***enter***. Type **102** ***enter*** **4** ***enter enter enter*** **0** ***enter*** **0** ***enter*** **1** ***enter***. (Assuming primers per Yamamoto, base pair size = 102 + 4 × allele#.)

IX.B.1.e.xi.) If you know a mutation rate, select *Locus parameters (mutation rate ...)*. If the mutation rate is 3/1000, type **0.3** ***enter enter enter enter***. (That's because 3/1000 = 0.3%.)

IX.B.1.e.xii.) *window size* can be left as all 0. The other menu items are irrelevant for STR's or are obsolete.

CLOSE returns to the *locus* menu.

IX.B.1.f. **Change locus parameters ("PROPERTIES")**

Highlight a locus click PROPERTIES to enter the *locus maintenance* screen (§IX.B.2) for the highlighted locus.

## IX.B.2. Locus maintenance screen

The locus maintenance screen (**Figure 102**) will:

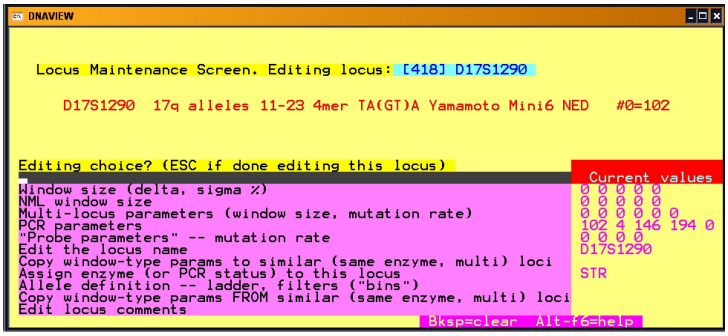



**Figure 102** Locus maintenance screen

IX.B.2.a. show the full comment (which may be as long as 4 lines).

The comment is built in and not user-editable. It includes such information as the name of the locus that officially (per DNA·VIEW world-wide definition) belongs on the line, Genebank information if I have looked it up, PCR parameters for various commercial versions of the locus (the size of allele #0 depends on the primer, although the repeat amount of course does not change), and perhaps more.

IX.B.2.b. present a menu of options

for modifying parameters and other editing functions for the currently highlighted locus. Skipping over obsolescent options (e.g. RFLP), the menu includes

IX.B.2.b.i.) *PCR parameters* — size of allele #0, repeat amount, size range for multiplexing. The PCR parameters are discussed in further detail in §IX.B.3.

IX.B.2.b.ii.) *Locus parameters* — mutation rate, default sizes for bands beyond the ladder region. Details in §IX.B.3.f.

IX.B.2.b.iii.) *Edit the locus name* — permits spelling changes, or introducing new loci

IX.B.2.b.iv.) *Assign enzyme or PCR status* — to establish or change the enzyme associated with this locus

IX.B.2.c. changing parameter values

If you CANCEL parameter setting the operation is aborted and parameter values are not changed.

Note that fractional parameter values are specified in %.

## IX.B.3. *PCR parameters* modified

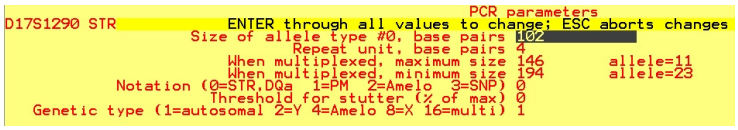



**Figure 103** PCR parameters

Users normally do not need to be concerned with this option which allows controlling details about how loci are handled and understood by the program. Normally new loci are automatically installed, with appropriate parameters, when a newer program version is installed.

DNA·VIEW displays and accepts PCR alleles in standard notation, i.e. letter or repeat number. Internally though – and mostly invisible to the user – the data is represented as a number (e.g. base pairs). The *PCR parameter* screen lets you edit the definition and notation. Provided the information is supplied consistently input and output from DNA·VIEW will occur in the expected way.

The parameters are shown in **Figure 103**.

IX.B.3.a. Conversion to base pairs

The first two PCR parameters are the numbers to convert from allele number to base pairs.

Referring to the examples of the last section, the relationship between 146 bases and allele 11 is defined by the formula, for the case of D17S1290:

$$146 = 102 + 11 \cdot 4$$

Thus the essential parameters are 102, which can be thought of as the size that allele #0 would be, if there were such an allele, and 4, which is the repeat amount.

IX.B.3.b. Repeat amounts and fictitious repeat amounts

IX.B.3.b.i.) If the *repeat unit* is 0 then the allele is just a number of base pairs, as for RFLP loci.

IX.B.3.b.ii.) For STR loci use the actual repeat amount – 4, 5, 2 etc. as the case may be.

IX.B.3.b.iii.) For Amelogenin use **6**. However, if you have already established other values and created some data using them, do not change the values.

IX.B.3.b.iv.) For polymarkers (including DQA1), I recommend using the fictitious values of **100** and **10** for the conversion parameters (per §IX.B.3.a). These values will give an acceptable display appearance. However, if you have already established other values and created some data using them, do not change the values.

IX.B.3.b.v.) For SNP's use the fictitious values of **100** for `size of allele #0` and **1** for `repeat amount`.

For each of the above categories, see also §IX.B.3.d and §.IX.B.6.a

IX.B.3.c. Multiplexing sizes

The third and fourth parameters, minimum and maximum size, are only necessary for purposes of importing data such as GeneScan data where there may be several STR's multiplexed in a single color. The "multiplex" parameters can be specified when needed during the import process (§IX.B.3.c)

IX.B.3.d. Notation

The possible values for the `Notation style` parameter are defined as follows:

**0** for STR, DQa or RFLP. Alleles are numbers, such as `12` or `1.2` or `5662`.
If *repeat unit* > 0, then the normal display would be the conventional repeat number. For example using the above parameters a 198 bp TH01 allele displays as `9.3`. Format variations are possible (*Option Pcr notation style*, §IX.F.28).

**1** for polymarkers or equine loci. Alleles are letters `A B C D ... Z`. (However, the equine allele `O` is sometimes shown on output as `ò` when the letter `o` would mean a null allele.)

**2** for Amelogenin. Alleles are named `X Y`.

**3** for SNP's. Alleles are named **G**, **A**, **C**, **T**.

IX.B.3.e. SNP's

For the conversion parameters, see §IX.B.3.b.v. Select the value **3** for `Notation`, which corresponds to alleles named G, A, C, and T.

Assign to SNPs the pseudo-enzyme SNP, which is [25] on the list.

IX.B.3.f. Genetic type

The genetic and other rules vary according to chromosome etc. Therefore the genetic category of loci is specified, using the coding

**1** autosomal
**2** Y-chromosome
**4** Sex marker, e.g. Amelogenin
**8** X-chromosome
**16** multi-locus probe

### **IX.B.4.** *Locus parameters (mutation, rare allele rates)*

Select a probe and you can set certain parameters for that probe. The parameters are

IX.B.4.a. *Paternal mutation rate* μ*: rate of false exclusions (%)*

μ is the percent of true fathers who would apparently be excluded by reason of mutation by this probe system. The PI will then be computed as described in §XIII.B.6.

If the mutation rate is specified as 0, the default value (§V.C.3.g, V.C.3.h) applies.

IX.B.4.b. *rare allele rate (only for Screening) (%)*

This special feature is to cater to things like a historical designation of the TH01 "10" allele as "R" (=rare), although in fact it is not "rare" in the usual DNA·VIEW sense of being nearly new. In that case, use this parameter to specify, per locus, a minimum % frequency for such pseudo-rare alleles.

By default, *Disaster Screening* (§VII.D) treats off-ladder alleles (if imported as "rare" – see §VII.B.2.b.ii) as new alleles and calculates their frequency according to the *Count observed allele* (§V.C.4.a) and *Minimum #* (§V.C.4.b) *Case options*.

The frequency assumed by the *Screening* search will be the largest of the frequencies obtained by considering each of the three parameters.

### IX.B.5. Additional locus topics

IX.B.5.a. Locus name format

The first ten characters of the probe name are shown on all menus, and are generally the locus name. The second ten characters can be used for chromosomal location name.

The only significance of the two parts is that on a PATER report (assuming that you have the PATER software as well), the parts are shown separated by /.

IX.B.5.b. To change a locus, use the method described at §IX.B.2.b.iii.

### IX.B.6. Locus status and Restriction enzymes

A locus can be a STR, or a SNP, or a POLYmarker, or can be assayed by restriction enzyme and a probe. In the latter case, an enzyme – user specified – is associated with the locus. In the other cases, the status of STR, SNP, POLY etc. is accommodated by same mechanism in the program, as if it were a (pseudo-)enzyme.

**Figure 160**, §XVI.A.3, page 315 is the standard list of pseudo-enzymes and ·restriction enzymes assigned so far. The list is user-configurable, but other than personal preferences for spelling it is unlikely to be necessary to change it. The first three characters appear in database lists.

IX.B.6.a. Suggested pseudo-enzymes

IX.B.6.a.i.) STR loci — It is best to associate pseudo-enzyme #22 only with STR loci. To this end it makes sense to change the spelling from "PCR" to "**STR**".

The reason is that "STR mutation model" – the (modified) stepwise mutation model – applies to loci so categorized.

IX.B.6.a.ii.) Polymarkers — For polymarkers, use instead #17 **POLY**.

IX.B.6.a.iii.) SNP — For SNP's, use #25 **SNP**.

### IX.B.7. Multiplexes

From DNA·VIEW's point of view a multiplex simply means a particular set of loci in a particular order.

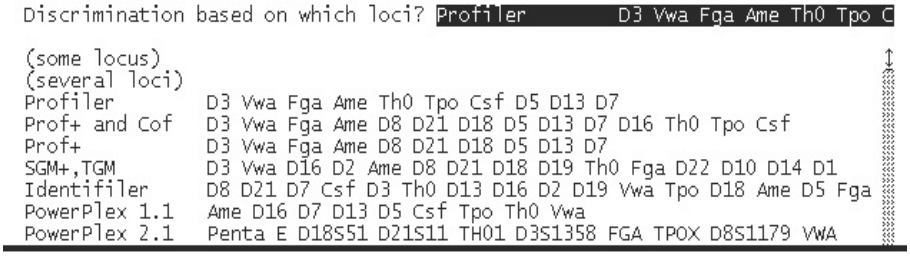



**Figure 104** Multiplex selection

IX.B.7.a. Uses

DNA·VIEW recognizes sets of loci in several ways –

IX.B.7.a.i.) to adjust and maintain the locus list

» locus order (§IX.B.1.d.i)

» which loci are active (§IX.B.1.c)

IX.B.7.a.ii.) in *Kinship simulation* (§VI.G.4.b) to say which loci are simulated

IX.B.7.a.iii.) *Racial distance* (§VIII.M)

IX.B.7.b. Selection

A large number of multiplexes are predefined. When the occasion arises to select a set of loci, these are always included in the selection menu. In addition, there are two special menu items:

IX.B.7.b.i.) *(some locus)* – lets you pick one locus. For example, you might want to do a one-locus simulation in order to evaluate the locus.

IX.B.7.b.ii.) *(several loci)* – lets you pick any set of loci by choosing them one at a time. Although there is no way for the user to add new "multiplexes" to the list, this option ensures generality in that you can do a simulation or racial database comparison for any combination of loci.

Select loci successively via OK and ADD ANOTHER until DONE.

## IX.C. *Worklist Sizes*


This command (in the **Examine data** menu) creates a profile report for a selected worklist.

## IX.D. Statistics

**Use:** ① Check problems reported during database compilation. ② One way (*Compare* may be preferable) to determine child sizes in a paternity case if using DNA·VIEW Abridged (otherwise *Paternity case* takes care of it automatically).

You can get a summary of all the sizings for an individual. If there is data for more than one probe, you choose which one you want to see.

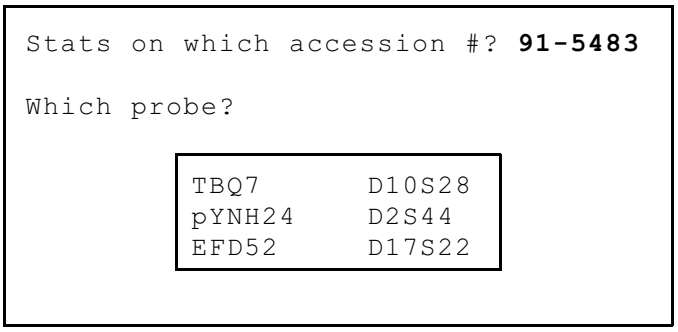



**Figure 106** *Statistics*; *same person* choice

A preliminary list then shows all worklists, reads, and lanes associated with the individual, whether alone or in co-electrophoresis, for the selected probe. If the number of bands is other than 2, there will be a notation next to the lane number — °, -, +, or * — indicating 0, 1, 3, or more bands.

You may eliminate some of the rows from the preliminary report by listing row numbers to exclude in response to the *Ignore sequence numbers?* prompt. So by excluding the reads for all but one lane, you can get the intra-assay statistics for that lane.

A further level of winnowing is offered when dealing with an accession number, such as a control, for whom there is a large amount of data. Both to speed up the analysis and because you may only be interested in recent measurements, you might be asked *Start from?*, with a choice box containing a list of all worklists. Only data from the worklist record you select or a later one will be gathered for the statistical summary.

The summary shows the various allele sizes (molecular sizes as determined by interpolation) available for the individual, and a statistical summary consisting of mean, range, and standard deviation. There are several possibilities and options for the list of measurements:

### IX.D.1. *Intra-assay* versus *Inter-assay*

IX.D.1.a. Intra-assay statistics

If there is only one lane, which has been read several times, then the various readings of the lane are shown (1 reading per row and 1 column per band), and the statistics are intra-assay — that is, they tell how much variation occurs from read to read of the same image.

```
Statistics for accession #89-02502      Probe pS194 D7S104
SEQ  Lane Read #  Read date    Reader Worklist,config & date
  1   16    87 1989/09/03 12h    173  89/08/29 c065 » #1a pS194
  2   16    88 1989/09/03 12h    173  89/08/29 c065 » #1a pS194
         Ignore which SEQ's?
   7445   7594
   7449   7586

   7447   7590  MEAN
      4      8  RANGE
    2.8    5.7  σ
  0.04%  0.08%  RELATIVE σ
```

Showing two readings of the same run for subject 89-02502, as well as the mean, range, standard deviation, and

**Figure 107** Intra-assay statistics

**Figure 106** is an example.

The resultant report can be *Print*ed.

## IX.E. Browse

The **Browse** program lets you examine lists of data and delete a variety of objects, and also provides access (also available through other menus) to change various switches and parameters. The possibilities are listed in **Figure 108**. Deleting generally also includes renaming.

*Worklists*
*Lanes*
*Options*

**Figure 108** *Browse* menu

### IX.E.1. Change name or comment

You can also use this menu to change the **comment** attached to most kinds of object. The general method is

» Select the object (worklist or configuration) as if to delete it

» but don't confirm the deletion:

```
Ok to delete? n
```

» then answer the prompts with the new comment, and also worklist date.

### IX.E.2. Lane deletion

Select *Lane* from the menu of **Browse** options and you are first given a list of reads to choose from. Each read starts with a number. Probably you know what number you want because you took it from the **Compare** report or from the **Statistics** preliminary report. Select the appropriate read.

A list is then presented showing, for each lane in the read, brief identification and, in parentheses, the number of bands measured. As you select the lanes in turn, the number in parentheses goes to (0). No file changes are made yet, though. When you finish, selecting no lane, you are asked for confirmation for your changes (*Commit changes to file?*). Answer **y** and the lane data is deleted from this read.

### IX.E.3. Read deletion

Select the **Browse** option *Read*, then select the read(s) to delete from the list. You must confirm each deletion (*OK to delete?*); then the read may be expunged.

Note: Reads can also be deleted with the *Worklist* command, or from any worklist list (§V.J.1.d.i).

### IX.E.4. Worklist, Configuration, and Standards ladder deletion

These are similar to Read deletion, except that there is a hierarchy of data:

i) Standards ladders are referenced by

ii) Configurations, which are referenced by

iii) Worklists, which are referenced by

iv) Reads.

To delete a configuration but leave intact a worklist depending on it would cause internal chaos. Therefore, whenever you attempt to delete any of these items, the program first attempts to delete any objects lower in the hierarchy that refer to it.

Example: You request deletion of Configuration x. Worklists xw and xn use configuration x.

You are prompted *OK to delete worklist xw*? Suppose you say **y**, but there is a reading, xwa, of worklist xw.

Then you are asked *OK to delete read xwa*? If you now say **y**, the following cascade of deletions occurs:

read xwa — DELETED!!

worklist xw — DELETED!!

Next, you are prompted *OK to delete worklist xn*? If you answer **no**, then the result is the messages:

worklist xn — NOT DELETED!!

configuration x — NOT DELETED!!

In summary:

- You can't delete anything you shouldn't.
- DNA·VIEW will warn you of any lower level objects, and guide you through deleting them.

Note: Worklists can also be deleted with the *Worklist* command, or from any worklist list (§V.J.1.d.i).

## IX.F. *Options*

This command is a miscellany of functionalities that you can specify to control program operation.

### IX.F.1. *Box drawing characters*

Three alternative drawing choices are available for box borders (e.g. when DNA·VIEW puts a box around a result in an *immigration/kinship* report:

IX.F.1.a. The box drawing characters ┌─┐ └─┘ │ ‖ are attractive if available but some national display or printing modes don't have them and the result is very unattractive. In that case choose one of these alternative:

IX.F.1.b. Symbols **+ - | >** as an approximate way to draw box borders, or

IX.F.1.c. *(none)* – leave the borders off. This is an improved alternative to §IX.F.8.c.

### IX.F.2. *Case # format: 09/00123*

new

This option lets you set three parameters governing case number appearance (format).

DNA·VIEW (and PATER) case numbers may be any integer from 1 up to about 2100000000 (that's 10 digits, and the exact limitation is 4-byte computer integers, i.e. case#<$2^{31}$). This allows for example a 4-digit year and 6 more digits of sequence number within the year.

For convenience of input and viewing appearance the case number can be broken into two parts with any character you wish separating them.

IX.F.2.a. The case number format parameters are

Y: `leading ("year") digits?` (typically choices are 2 or 4)

**Y** controls two things: (a) leading 0 padding on display of a case number, (b) the meaning of the special case number "year" input buttons per §V.B.28.a.iii. **Note** that if you want to type in the year you can use as many digits as you wish, not even necessarily relating to a year. For example 12345/12345 can be legal case number.

N: `trailing (sequence) digit positions?` **`5`**

**N** controls (a) 0-padding after the separator on display of a case number (i.e. `09/00123` rather than `09/123`), (b) minimum number of digit positions for the sequence part when you use a special input key **`L T`** or **`N`** to prefix a year code without typing the year.

S: `Separator character(s). (First one is for output; all for input.)` **`/.-`**

The first separator is the main one, which the program will use for display of case numbers. The others are just extra options that you can use for typing in case numbers.

» **Display** appearance is determined mostly by the parameter N and the first S. The full integer case number is written. If there are *more* than N digits, then the first separator is placed before the final N digits. If the result is *fewer* than Y digits (but at least 1 digit) before the separator, then additional `0`'s are padded to the left.

There may be *more* than Y digits before the separator.

» **Input**. When entering a case number there are several possibilities:

- Type in as many digits as you wish, e.g. **`200901234`**

- Type some (prefix) digits, any of the separators, then more ("sequence" or suffix) digits, e.g. **2009.1234**. If the suffix has fewer than N digits additional 0's will be inserted.
- Type a suffix (that is, the "sequence" digits) then (instead of ***enter***) one of the special keys **T L N** to create a prefix consisting of the last Y digits of **T**his, **L**ast, or **N**ext year.

  The suffix digits will be padded to (at least) N digits, even if there is no separator character.

Examples (Assume this year = 2009):

| Parameters | | | Some input possibilities | Case number | |
|---|---|---|---|---|---|
| Y | N | S | | internal number | display form |
| 0 | 0 | (none) | **123** ***enter*** *or* **123 T** | 123 | |
| 2 | | | **90123** ***enter*** *or* **0123 T** | 90123 | 090123 |
| 4 | 4 | | **123T** | 20090123 | |
| 2 | 5 | / | **123N** *or* **10/123** | 1000123 | 10/00123 |
| | | 0 | **99.432** *or* **99*0432** | 9900432 | 99*00432 |

**IX.F.3.** *Command menu style: FLAT/2 levels*

If you are an experienced user, set this parameter to **FLAT** ("expert") for a "flat" main menu with lots of items. It's not so clear, but requires fewer keystrokes to get around.

When learning familiarity, you might prefer the hierarchical, 2-level menu.

**IX.F.4.** *Compare report style*

The Compare report will perform in various alternative ways depending on the way you set these options.

The older, *long format*, is retained mainly for compatibility.

The *short format* tends to be neater and less cluttered. First, the marker lanes are not included. Second, lanes with no data are not included.

Third, new information is included: In addition to the digitizations from the different sizings, an average is also shown.

Fourth, the sizing information (the averages) are optionally written to a Paradox table.

Fifth, compared to the longer format the information is presented in transposed format — each different reader is a column, not a row. Each different band gets a row.

**IX.F.5.** *Convert accession codes: (none) / WTC / Katrina*

The "WTC" or "Katrina"settings cause accession codes to be displayed in a format consistent with World Trade Center or Katrina identification usage.

**IX.F.6.** *Date order: 3/18/2008*

Nominate whether dates are displayed American (month-day-year), European (day-month-year), or Asian (year-month-day) style.

**IX.F.7.** *Decimal marker: . (dot)*

For international compatibility, you may select dot or comma.

**IX.F.8.** *DNA·VIEW character translation*

IX.F.8.a. Symbol sets

Accented characters sometimes fail to print correctly because of the printer using a "symbol set" different from the symbol set used on the display monitor. The display monitor symbol set is called "PC-8", and has for example the symbols **ü** and **é** as ASCII codes 129 and 130. If the printer is also operating with the PC-8 symbol set, then no translation is necessary.

However, there are also other symbol sets — especially used with languages that have many accented characters — and they differ from PC-8 with respect to the symbols that correspond to the ASCII codes, especially the upper half of the ASCII table from 128–255. For example, the HP Roman 8 symbol set includes extra accented characters, omits the line drawing characters, and the symbols **ü** and **é** are ASCII codes 207 and 197.

IX.F.8.b. Translation mechanism

Therefore, in order to allow these and other characters to print correctly or nearly so on a printer that is not using the PC-8 symbol set, a translation mechanism is provided. This option provides a mechanism whereby the numeric ASCII code for each screen character can be translated into a different numeric code when printing.

The mechanism provides a menu of the higher ASCII codes, from 128–255. Select a code from the menu, then respond to the prompt to give the new ASCII value. When done with selections, make the empty selection and only then will the new translation table be filed.

While you can assign any translation codes you like, the program has some intelligence and knows what the likely translation is based on your choice of language (see §IX.F.18). If a character is currently listed in the table as untranslated, the likely translation will be suggested as a default.

IX.F.8.c. Box-drawing characters

Depending on the printer settings, those characters that make elegant borders on the screen may look like garbage on the printed page. You can get rid of them by specifying a translation to 0. This doesn't actually introduce 0 characters (nulls) into the output; it simply suppresses the character. Deprecated – easier to use §IX.F.1.

**IX.F.9.** *DNA·VIEW Printer*

Change the kind of printer used for reports (not graphics) — the printer that DNA·VIEW will assume it's talking to when you *Print*. The main options are

» *Laser* to print directly to a Laserjet, Deskjet, or compatible printer;

» *PrintFil* to print in cooperation with the PrintFil software. See §XV.D.3.a for more details.

To specify the kind of printer for purposes of graphics (e.g. printing images from *Plot*, §VIII.E), you have to edit the CGI.CFG file (XV.D.6). But see *METAFIL*, §VIII.E.1.b.v.

**IX.F.10.** *DNA·VIEW Printer port*

Possibilities:

» *(Display on screen)* defers the choice until each time you print (see §V.K.1),

» *print via PrintFil* will, if the PrintFil (see §XV.D.3.a) software is installed, send the image to PrintFil for processing and printing,

» *LPT1*, *LPT2* print via (probably virtual versions of) these traditional ports. To print to USB, choose either of these and see §XV.D.2.a.v.

**IX.F.11.** *DNA·VIEW report title line*

With this option you can change the report title of the printed reports. It is possible to create a fancy title with varying fonts or sizes by inserting printer control codes – e.g. PCL for a LaserJet or Deskjet printer. As part of these codes you will need to insert the non-typeable character ***Esc***. For this purpose you can type *{* as a surrogate, which will be translated to ***Esc***.

**IX.F.12.** *DNA·VIEW report includes: [Version, Workstation]*

For better tracking and documentation of the reports that get printed by DNA·VIEW, you may elect that the DNA·VIEW version number and/or the network name (see Network, §XV.D) of the workstation from which the report is printed be included on the first page of each report.

**IX.F.13.** *DNA·VIEW report epilogue*

The *epilogue* is a technical facility. If for example DNA·VIEW is sharing the printer with other users and needs to send a special printer sequence after each use of the printer, this is the place to put that sequence.

**IX.F.14.** *Default date?*

If you choose **y** then you can enter new accession numbers faster because ***Enter*** will accept today's date as a default. On the other hand, if you are serious about the accession date — perhaps it is part of your documentation — then choose **n** here to minimize errors on input.

**IX.F.15.** *Graphics detail scaling*

If your printer is a deskjet some of the graphic output looks better if you set this parameter to 0.5. Otherwise, it should be 1.

**IX.F.16.** *Interpolation parameters*

Using an allelic ladder, molecular sizes are usually determined according to three flanking markers. Otherwise, eight markers are used for a globally fitting curve.

You can experiment with other choices than 3 and 8 using this option.

**IX.F.17.** *Keyboard*

The *Keyboard* choice lets the user choose among various national keyboards. There are a variety of special keyboard drivers, not guaranteed to work ideally. To switch from one choice to another it may be necessary to exit from the program, especially when to simply remove a driver via the choice *(none)*.

**IX.F.18.** *Language*

Some of the menus, explanations, and report paragraphs have multi-lingual options. Choose which language you prefer.

**IX.F.19.** *Locus name style like: D16S539 STR*

Four formatting choices for locus name display are offered:

(a) The bare locus name, such as `D16S539`, or:

(b) Include the chromosomal information, and/or

(c) Include the category such as `STR`.

**IX.F.20.** *Locus parameters*

This option is equivalent to *Maintenance* option of the same name described in §IX.B.

**IX.F.21.** *Message display time*

Sometimes DNA·VIEW pops up a message for a limited time (maybe 1 or 3 seconds by default depending on the message). Set this value to 10 seconds to give yourself lots of time to read (anyway ***space*** when the message appears will hurry it along), or 0 for the default times.

**IX.F.22.** *Minimum frequency parameters*

Same as *Options from "Case"* (§V.C)

**IX.F.23.** *Name tag style: NAME/DATE*

Toggle whether the name tag block – the list of people in a case – displays an accession date, or the name of the person.

**IX.F.24.** *Numlock on/off*

Reverse the default state of the “numlock” key.

**IX.F.25.** *Off-ladder allele policy: DISCARD/RARE*

(revised – 2014)

When importing DNA profiles (§X.A – using e.g. *Genotyper import*), what happens when an illegal notation (such as "?" or "OL" or "OL13.2") is encountered. Three choices, which roughly corresponding to ignoring 2, 0, and 1 allele at the locus:

» *Discard genotype if it includes "ol", "?" or other garbage* means that the allele and it's companion (i.e the genotype for the locus for that person) are ignored and not imported.

» *Treat "ol", "?" etc as a rare allele (but "OL13" as 13)* means that the allele will be treated as a rare allele not found in any database. Rare alleles will match one another, however.

» *Ignore "OL", "?" etc (as if blank); treat "OL13" as 13*. The illegal notation is ignored and the other allele (if any) is imported. Most DNA·VIEW calculations treat the single allele as a homozygous type, though sometimes a null allele is allowed for also.

**IX.F.26.** *Options from "Case"*

This is an alternative way to access the same options that are accessible as *Options* from the *Paternity/Automatic mixture* or *Automatic Kinship* sub-menu (discussed in §V.C).

**IX.F.27.** *PCR allele binning? NO/YES*

This **obsolescent** option controls whether “allele resolution filters” are active. If it is set to **`yes`**, then all sizes are resolved, if possible, before being used for casework, creating databases, etc. If it is set to **`no`**, then raw sizes as stored are used.

**IX.F.28.** *PCR notation style*

Using this option from the **Options** submenu, you can choose how PCR sizes will be formatted for output. PCR sizes are shown as base pairs (optionally), and (also) as an estimated allele number and discrepancy.

IX.F.28.a. Round/Truncate

IX.F.28.a.i.) `Truncate size to estimate allele number, offset always positive: "17.15"`

An allele is shown as a positive (or zero) discrepancy from the next lower integer allele. For example, the common TH01 variant allele will print as 9·3 even though it is closer to 10 than to 9. This option is consistent with standard usage in the literature.

IX.F.28.a.ii.) `Round the sizes to estimate the allele number, offset may be negative: "18.-1"` (obsolete – do not use)

IX.F.28.b. Short/Long notation

Choose the "short" format (e.g. `20.9`). "Long" is obsolete.

IX.F.28.c. Omit/Show odd base pairs

If the size is an exact number of repeats, you can choose to display (i.e. `18.0`) or to omit (just `18`) displaying the 0 odd base pairs. The second way is probably more readable. The first alternative is included mainly in case you will be passing the sizes along to another computer program (i.e. using the *Compare* report).

IX.F.28.d. decimals

In case the allele is represented, for whatever reason, as a non-integer number of base pairs, you can show 0, 1, or 2 decimal digits.

IX.F.28.e. separator – (period is universally standard)

To display a fractional repeat amount, several choices are possible: 9·3 or 9.3 or 9,3.

**IX.F.29.** *page format*

This controls how various reports use the paper. If the printer is a laserJet, even the type size can vary in order to fit the report better on the paper. You specify

IX.F.29.a. *Normal type, chars/inch, (usu 12)* for the standard type size.

IX.F.29.b. *Maximum chars/inch, (rec 20)* permits a smaller type to be used to avoid line wrapping, such as for the "RAW FRAGMENTS" section of the paternity report.

IX.F.29.c. *Left margin columns (rec >3)* is the left margin in "normal" columns.

IX.F.29.d. *Column positions printable (US<87, A4<84)* again are in units of the "normal" type. The numbers in parentheses correspond to 0 right margin, so at least 3 fewer is recommended.

IX.F.29.e. *Lines/page (US<69, A4<74)*. Again, the given figures will result in no bottom margin, so subtract a few lines. Alternatively, you may specify an essentially infinite page such as 9999 lines to defeat DNA·VIEW's pagination attempts and page headings. The printer will then start a new page when necessary.

IX.F.29.f. *Use Natural page breaks, or Full pages to save paper?* **N**atural breaks means avoid breaking a logical section across pages. **F**ull page means conserve paper, but harder to read.

**IX.F.30. Paradox** *directory*

Best practice and normal installation is to specify **`pdox\`** as the location where that Paradox tables are kept. This relative notation denotes a subfolder of the installation folder for DNA·VIEW. Using relative notation ensures that no change will be needed if DNA·VIEW is migrated to a new location.

**IX.F.31.** *PATER character translation*

This option controls the character codes that are used for PATER reports, and doesn't affect DNA·VIEW at all. Operation is as described in §IX.F.8.

**IX.F.32.** *PATER signature line*

**IX.F.33.** *PATER Logo*

These options are only useful if you are using the PATER program to generate reports. They allow you to specify some of the boilerplate on the PATER report.

**IX.F.34.** *Plot line thickness*

This controls the thickness, in pixels, of lines in data base plots (§VIII.E). A value of 1 gives a narrow line, 100 or 200 gives a very bold one, 2000 looks rather amusing like advertising copy.

**IX.F.35.** *RFLP: disabled*

Some menu options that are useful only for RFLP work are hidden; switch to *enabled* to see them.

**IX.F.36.** *Window style*

This option defines the way that DNA·VIEW interprets the window size parameter delta (δ) for probability computations.

For discrete allele systems either δ=0 or at most δ is very small. In either case the alternatives for windows size interpretation probably have exactly the same result.

*Recommended window style* is the best choice.

## IX.G. *Profile Data*

The *Profile Data* command, in the **Examine data** menu, is a quality control tool to check for inconsistent or incomplete data sets.

Examine and compare the DNA profiles for the individuals on one or several worklists. The result is a report showing

IX.G.1.a. discrepancies – any samples-loci for which there is a different DNA typing result on two different worklists;

IX.G.1.b. duplicate assays – a table showing which samples-loci have data from at least two worklists, and those with missing data.

The report is tab-delimited and therefore is perhaps most easily visualized by importing it into a spreadsheet program such as Excel.

| **Discrepancy section** | 2 typing conflicts | Kin Lab B | Kin Lab C |
|---|---|---|---|
| One row per discrepant sample-locus<br>One column per worklist | 1001-01059 M 702044 D3S1358 | 15 16 | 14 17 |
| | 1001-01060 C 702044 D3S1358 | 15 16 | 12 |

| **Duplicate assay / missing data section** | sample | D3S1358 | FGA | D21S11 |
|---|---|---|---|---|
| One row per sample<br>One column per locus<br>key:<br>! = discrepancy (very bad)<br>– = only one assay<br>(blank) = no assay<br>* = two or more assays (good) | 1001-01059 M 702044 | ! | * | – |
| | 1001-01060 C 702044 | ! | * | – |
| | 1001-01061 D 702044 | * | | – |
| | 1001-01062 E 702044 | * | * | – |
| | 1001-01063 A 702044 | * | * | – |

## IX.H. Compare

**Use:** ① Check problems reported during database compilation. ②Determine average sizes in a paternity case if using DNA·VIEW Abridged (otherwise *Paternity case* takes care of it automatically) and if the case falls on a single gel.

**Compare** prepares a report showing the molecular weight determinations for a worklist. If there are several reads of the worklist, the various weight determinations will be shown in columns for comparison.

The first step in **Compare** is to choose a worklist. If the lane identification data for the worklist has not previously been supplied, you'll be given the opportunity to fill it in now.

For the worklist, all known reads are presented in a list. You may trim the list by answering as indicated.

If the interpolation calculations haven't previously been performed, they will be done now. This takes 1–4 seconds per read of a worklist, depending on the computer.

The report format is one paragraph per lane, and one line per read within the paragraph. The paragraph is labeled with the lane number and accession numbers. In case of a standards lane, the reference sizes of the ladder rungs are also presented.[16]

An alternative format — controlled by **Options**, *Compare Style* — omits ladder lanes and optionally files sizes to a Paradox table, among various differences. See the discussion of **Compare** options (IX.F.4).

## IX.I. Directory

**Directory** will give you a listing of all accession #'s that appear on any worklist in the system, and for each accession number the listing will show for which loci that number has been digitized.

**Directory** can be useful

- » to cross-check worklists (V.J.3) against reads, to make sure all worklists have been read.
- » to see which people have only been run with part of the probe battery.
- » to determine probe usage for royalty payments.

[16] The DNA·VIEW interpolation algorithm uses a global curve fitting method. Therefore, the size determination for a given digitization may not be exactly the same as the reference value for the putative allele.

## IX.J. Quality Control

You can tell DNA·VIEW that certain accession numbers represent samples used for quality control. Presumably there are quality control samples routinely run on the same gels as ordinary casework. If the size values for the controls fall outside of expected bounds, there may be a technical problem in running the gel. If far out of bounds, maybe there's a mixup.

Defining the lists of quality control samples and creating the expected size ranges is performed by the **Quality Control** command. The expected size range can be defined in either of two ways:

i) based on historical data from sizes in the DNA·VIEW files. This is the designed method. The historical data is compiled into the appropriate statistics using *recompute*, described below.

ii) based on sizes and ranges typed in by the user. The statistics can be typed in using *edit stats*, described below. This method is supplied as an adjunct to the Codis export (§X.F) facility. It also might be useful when DNA·VIEW is first installed and there is no historical data.

The quality control data, defined here, is used for checking (monitoring) by the programs **Read** and **Import/Export** *Codis: export size in CMF format*.

The **Quality Control** command provides the following options:

**IX.J.1.** *add control*

For each probe, you can designate as many accession numbers as you like. DNA·VIEW will then monitor (§V.A.10) their appearance on each reading with that probe of any gel.

The same accession number can of course be enrolled as a control for two or more different probes. Note that there is nothing to stop you from using the same accession number even if the physical samples that are used as controls are different for two different probes. The possible advantage of this trick is to have identical rosters for two different probes. If the rosters are identical, then you can use one copy of the roster (§V.J.3) for both probes.

**IX.J.2.** *disenroll control*

You can remove them from the list.

**IX.J.3.** *edit stats*

The sub-menu option *edit stats* within **Quality Controls** is used to manually change or enter the reference parameters associated with a quality control sample.

01-00562 pYNH24 D2S44 Hae III
04-00562 pYNH24 D2S44 Hae III
01-00562 TBQ7 D10S28 Hae III
04-00562 D2S1338 PCR

Edit stats of which control? **04-00562 D2S1338 PCR**

**Figure 109** Select quality control to edit

**QC Stats for 04-00562 D2S1338 PCR**

| Size | Std dev (bp) | Coeff Var |
|---|---|---|
| 11 | 0.1 | 0.03% |
| 12 | 0.2 | 0.07% |

***ENTER* after each field. *END* or *ESC* when done**

**Figure 110** Edit Quality Control statistics

After you select this option, you are presented with the list of controls-loci (**Figure 108**). Select any one of them and you are presented with the statistics for that locus (**Figure 110**). You can manually enter/ change the sizes and standard deviations (e.g. to correspond to the official CODIS values). The Coefficient of Variation (std dev / size) is recomputed as you change each changeable field.

***Enter*** after entering the new number for each field. ***End*** when finished to return to the **Quality Control** sub-menu. At that point **edit stats** will conveniently be highlighted as the default choice in case you want next

to edit the statistics for another quality control. Moreover, when you re-enter the **edit stats** screen, the next quality control in order will cleverly be highlighted so that you merely need to type the sequence ***end enter enter*** to finish editing one quality control sample and begin editing the next one.

IX.J.3.a. Effect of modifying QC parameters

When you edit the QC statistics as shown above, the typed-in values *replace* the values (if any) that had been computed by the **recompute** sub-menu option. This might be the range report after editing as above:

Quality Control Standards

| Sample | Probe | Date Range | Size | StdDev | N | ±2 s.d. | ±3 s.d. |
|---|---|---|---|---|---|---|---|
| 01-00562 | pYNH24 D2S44 Hae III | 91/09/19-98/04/18 | 1529 | 1 % | 14 | 1500-1558 | 1486-1572 |
| | | | 4005 | 0.5 % | 17 | 3975-4045 | 3955-4065 |
| 04-00562 | pYNH24 D2S44 Hae III | 92/11/20-97/04/28 | 1995 | 1 % | 3 | 1955-2035 | 1935-2055 |
| | | | 5153 | 0.43% | 3 | 5109-5197 | 5087-5219 |
| 01-00562 | TBQ7 D10S28 Hae III | 91/09/19-98/04/18 | 998 | 0.21% | 2 | 993-1002 | 991-1004 |
| | | | 1541 | 0.48% | 2 | 1526-1556 | 1518-1563 |
| 04-00562 | TBQ7 D10S28 Hae III | | 1992 | 1.1 % | | 1950-2034 | 1929-2055 |
| | | | 7110 | 0.7 % | | 7010-7210 | 6960-7260 |

IX.J.3.b. Purposes of QC stats

The QC statistics serve two purposes, which are potentially in conflict:

i) They are used by **Read** to prepare the QC comparisons in the Lane Report. For this purpose we normally want the historical values from this laboratory, as obtained by **recompute**.

ii) They are used by CODIS export (§X.F) as parameters in the "elliptic fit" formula. For this purpose we need to use the national consensus values, typed in using **edit stats**.

One way to avoid conflict is to have two versions of each control, as illustrated above. Continue to use the control called 01-562 for the paternity work, but use 04-562 for the CODIS worklists.

When you use the **recompute** option to recompute the laboratory values for paternity work, you must then either:

a) **recompute** only one locus-probe at a time, to avoid recomputing and overwriting the CODIS values, or

b) type in the CODIS values again after doing the recompute.

(A third, optimally convenient option may be added in the future.)

IX.J.3.c. **Note**: vs *recompute*

*Edit stats* is a manual alternative to *recompute*. At least as regards any particular QC, you would do one or the other, but not both.

**IX.J.4.** *list*

For each designated probe/accession number combination, DNA·VIEW keeps track of statistics on recent gel runs. Select *list* to view a table like **Figure 111** of the current statistics and to prepare it for printing.

```
Quality Control Samples

Size ±Std Dev
pS194   91-05307
pYNH24  01-00011   3087 ±16                91/10/01 - 92/07/15
pYNH24  01-00009
TBQ7    01-00009   1346 ±18   3883 ±33     91/10/01 - 92/07/15
EFD52   01-00020   1269 ±11   4103 ±39     91/10/01 - 92/07/15
```

**Figure 111** *Quality Control* summary list

**IX.J.5.** *print*

Same as **Print** — send the last report to the printer.

**IX.J.6.** *range report*

```
Quality Control Standards

Probe     Sample        Date Range     Size   StdDev  N   ±2 s.d.      ±3 s.d.

pYNH24   01-00011   91/10/01-92/07/15 3087    0.53%  57  3055-3120    3038-3136
TBQ7     01-00009   91/10/01-92/07/15 1346    1.4 %  76  1309-1383    1291-1401
                                      3883    0.85%  72  3817-3949    3783-3983
EFD52    01-00020   91/10/01-92/07/15 1269    0.9 %  34  1246-1292    1235-1303
                                      4103    0.94%  34  4026-4180    3987-4219
```

**Figure 112** *Quality Control* range report

**Figure 112** shows an alternate format summarizing the quality controls which is obtained by selecting the menu item *range report*. Note that the range report includes the number of measurements, **N**, which there wasn't room for on the summary list report.

**IX.J.7.** *recompute*

After you *add control*, and from time to time, you have to *recompute* in order to create and save the statistics. The statistics will be based only on worklists on or after one that you pick. Therefore you have the option either to recompute everything at once, or to recompute each control singly.

The statistics maintained are the average size calculated for each band, and the interassay standard deviation, in base pairs, of the set of calculated sizes.

Ignored readings: If there are homozygous and heterozygous readings on file for the same control and probe, the homozygous ones are ignored. Readings with more than two bands are ignored. Also ignored are lanes labeled as a mixed sample, as are all lanes with a label in the second position of the worklist roster (i.e. even if the first position is blank).

If no valid readings are found for some samples, a warning report is displayed and the old values are not changed for those samples.

IX.J.7.a. **Note**: vs *edit stats*

> *Edit stats* is a manual alternative to *recompute*. At least as regards any particular QC, you would do one or the other, but not both.

**IX.J.8.** *show details*

As a side effect, *recompute* saves the individual band sizes lane by lane for each control. It is very useful to look at this list if the statistics happen to be suspicious. If there are one or two aberrant numbers, you can quickly find them and note the worklist and lane from which they come. This information, coupled with **Compare** or **Flash** and **Browse** (delete lanes) makes it easy to clean up the data.

# X. IMPORT/EXPORT

This chapter describes the various options of the *Import/Export* command. Since there are about twenty options it is convenient to group them into categories.

## X.A. *Import/Export* – import DNA profile data

DNA profiles can easily be imported using files similar to those produced by the ABI Genotyper (§X.A.4) or GeneMapper (§X.A.5) or Hitachi STRCALL software, but with quite a lot of extra flexibility. The same file can even be used to cause DNA·VIEW cases (*Paternity / Crime / Automatic Kinship Case*) to be created at the time of import.

The input is treated as a collection of "Reads" – one per locus – which are associated with a worklist. The data thus becomes available for casework, for building databases (§VIII.C.4), etc.

### X.A.1. Importing steps – preliminary

X.A.1.a. Create the table of alleles & other information outside of DNA·VIEW. The ABI Genotyper or GeneMapper or Hitachi STRCALL software automatically create data in an acceptable format. It is also quite easy to prepare (or modify) the data manually in Excel per §X.A.4.

X.A.1.b. Tab-delimited text file

X.A.1.b.i.) DNA·VIEW can't directly import an Excel file. Therefore save the file as text. You can do this directly from the GeneMapper or Genotyper software, or you can use Excel to convert.

Save as type: **Text (tab delimited) (*.txt)**

X.A.1.b.ii.) Save anywhere you like, but the folder `\DNAVIEW\IMPORT` is convenient

X.A.1.b.iii.) Save as: Any convenient name, but realize that long names or names with blanks will appear a little odd when you look for them in DNA·VIEW (§X.A.2.b.ii).

### X.A.2. Importing steps – in DNA·VIEW

For keystroke-level detail, see the tutorial §IV.B.1.

X.A.2.a. Select the *Import/Export* command, *DNA profile data import*, then option *Genotyper Import* or *GeneMapper Import* or *Hitachi STRCALL import*.

X.A.2.b. Choose a file to import

X.A.2.b.i.) *Type in the name of the subdirectory (=folder)* where the file(s) to import reside.

For example: `C:\DNAVIEW\IMPORT` (which can be abbreviated to `IMPORT`). *Don't* type the file name at this point!

The directory/folder selection is persistent; you won't need to type it again unless you want to change the directory that you use.

X.A.2.b.ii.) *Select a file* to import from the menu of files.

X.A.2.c. Create (NEW) (or rarely choose[17]) worklist to import into.

---

[17] If you are re-importing a genotype file after having corrected some genotype information, re-select the same worklist and the same "reader" as before so that the new genotype information will over-write the old.

However, if the *roster* information has been changed, then you need to delete the old worklist (or at least the roster information in it) before re-importing against it.

If the roster information of the new genotype file has no conflicts with the previously created worklist then the additional data can be imported against the same worklist. For example, additional loci can be added this way. Please note §X.A.2.d though.

If the new genotype file includes new lanes, higher-numbered or otherwise not included among any lanes previously defined on the stored worklist then you may add the new data to an old worklist.

X.A.2.c.i.) Date. ***Enter*** or OK to accept today's date; otherwise EDIT for a custom date.

X.A.2.c.ii.) `Worklist # or brief ID`. The file name will be suggested as a worklist name comment. Change it to a descriptive phrase if you wish.

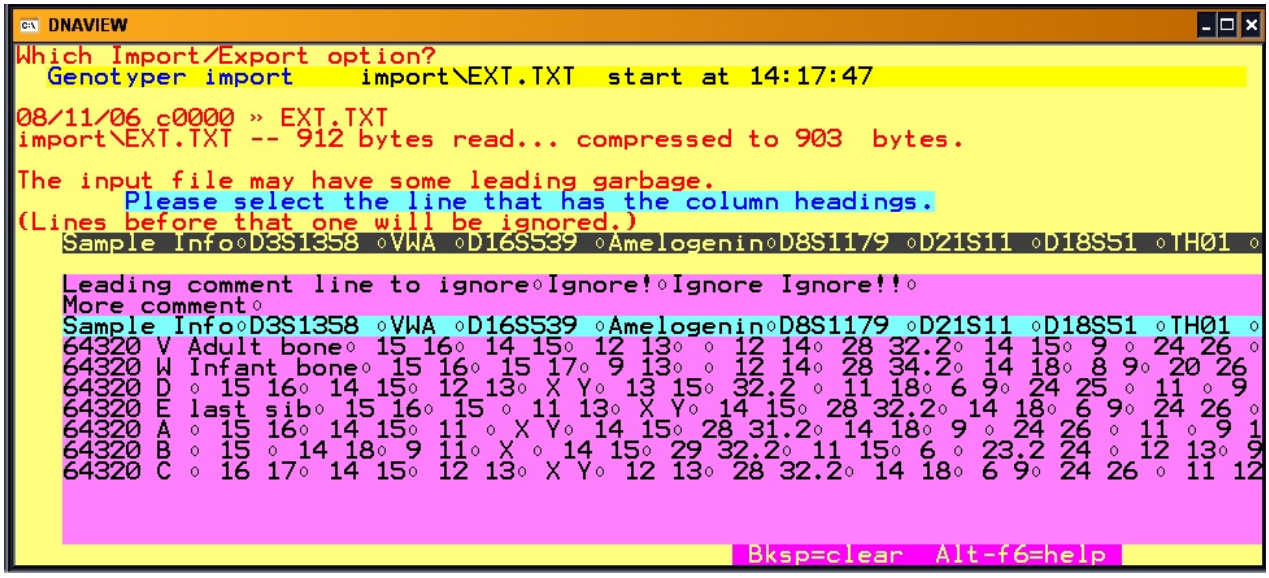
X.A.2.c.iii.) `Ok so far?` NEXT>>

X.A.2.d. *Choose a reader*

Reader number **099** is appropriate choice when the allele file is software-generated. This choice is interpreted by *Paternity (etc) Case* to mean the data is infallible and doesn't require a duplicate entry. However, you may use any reader number. If you use the same reader number twice for the same worklist, the second import will supplant the first one. The second and subsequent import with different readers will be compared and any differences are mentioned in a *Case* report.

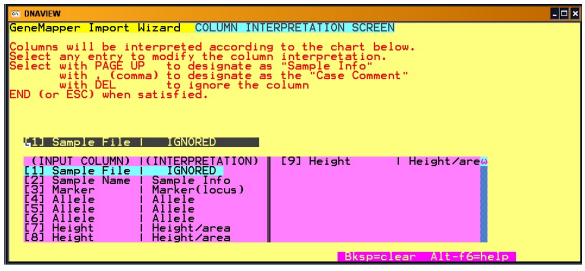
X.A.2.e. Find title row

If the first row of the file looks like it has the column headings, then this step is skipped. If not, then a




wizard screen as illustrated here is presented. Select the line which you wish DNA·VIEW to use as the column heading line. Any preceding lines are ignored.

X.A.2.f. *Genemapper import* only – Column interpretation screen




Depending on how it is configured, GeneMapper™ can produce some variation of the output columns. DNA·VIEW guesses how the GeneMapper columns correspond to information that DNA·VIEW is interested in. To guarantee complete flexibility, a *column interpretation screen* allows the user to over-ride any incorrect guesses.

The critical (recognized and imported columns) are

X.A.2.f.i.) Sample Info – means the column containing DNA·VIEW sample names in any of the various allowed formats (§X.A.8). (If there is a column named "Sample Name" or only one column named "Sample" anything, DNA·VIEW guesses that it is the Sample Info column. Otherwise, DNA·VIEW requires the user to choose.)

X.A.2.f.ii.) Comment (optional). See §X.A.5.a.ii.

X.A.2.f.iii.) Marker – the column with locus names. The word "Category" is recognized as an alternative.

X.A.2.f.iv.) Allele – a column or columns with the allele sizes. The word "Peak" is permitted as an alternative to "Allele" for the heading of the columns containing the locus names. "Allele 1", "Allele 2" etc are ok. If you were wondering, no, it is *not* ok to call some of them Allele and some Peak!

X.A.2.f.v.) Height – optional column or columns with peak heights. If present, they must be equi-numerous with Allele columns.

***Esc*** if all column interpretations are ok; select any row to change the interpretation for that row. This

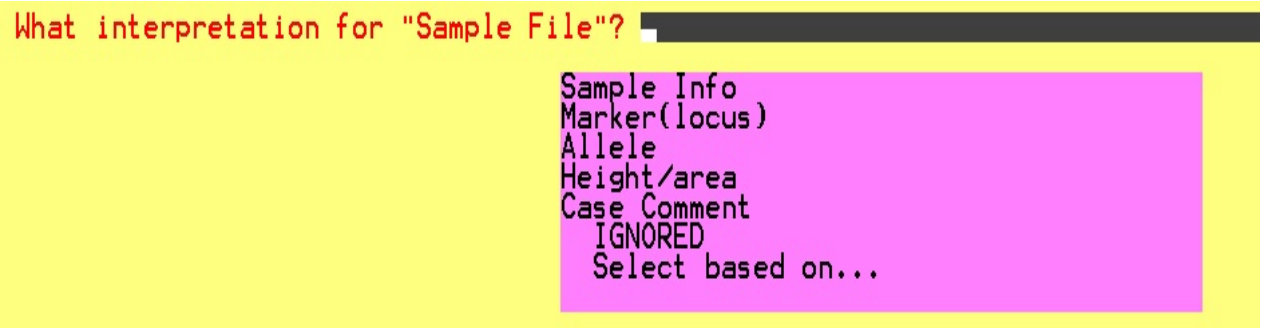



**Figure 115** Define GeneMapper column meaning

brings up a special dialog box (**Figure 114**) from which you may define the column as you wish, regardless of the title on the column.

» **Column filter** – One special option, `Select base on ...`, allows you to use a column as a *filter*. The causes a pop-up to list choices in that column, and you may type a phrase indicating the choices (that is, the rows) that you wish to accept. *Example*: If you have population studies for several different races, you could select one race at a time for import.

X.A.2.g. Locus/input field interpretation screen

A vertically-split menu shows the column headings from the import document on the left, and DNA·VIEW's guesses as to the intended meaning on the right (**Figure 115**). You may modify

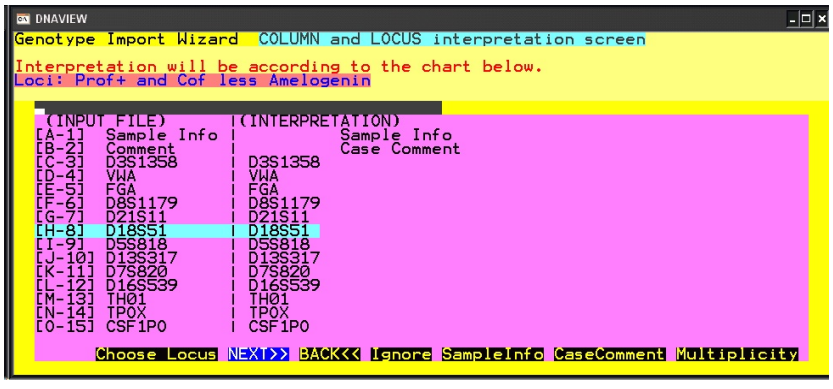



**Figure 116** Locus interpretation screen

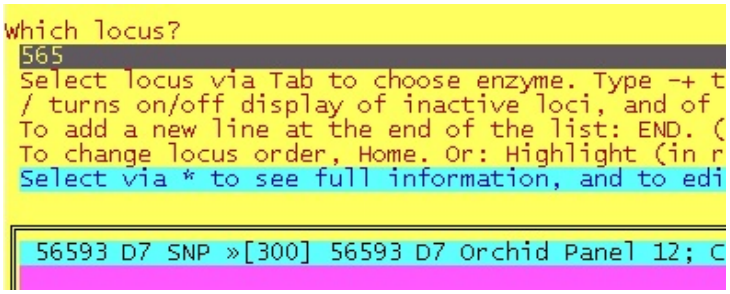
the guesses if there are any problems, using the buttons:

» NEXT>> if all ok.

» If any locus is misinterpreted or marked as AMBIGUOUS, select it and then select the correct locus from the subsequent locus menu.

» If a locus is marked as IGNORED but you do want to import it, try the same procedure.

» If you cannot immediately find the desired locus in the locus list, maybe you need to click FULL LIST to show the full locus list.




X.A.2.g.i.) Shortcut keys: Move the red bar to a row to re-interpret (i.e. ***single left click***), and

» If the wrong column (or no column) is recognized as `Sample info`, click SAMPLE INFO at the right one.

» To omit a column, use IGNORE.

X.A.2.h. Create cases in DNA·VIEW

If the Sample Info (or Specimen ID) field (§X.A.7.d) includes case+role (§X.A.8.b) specifications of people, the import process may (if there is any new case, and case parameters genre or computation race per §X.A.7.e.ii are not supplied for it) need to create case and people definitions.

If so, the screen shown in **Figure 117** shows the default suggestions for three parameters that the program needs to build a case: kind of case, race list, and starting accession number. If the suggested values are what you want, simply answer ***Enter*** or **y**. If you need to change any of them, answer **n** and respond to the prompts that follow.

X.A.2.h.i.) `What kind of cases?`

*Paternity*
*Maternity*
*Kinship*
*Crime*

Select from the menu choices – paternity, etc.

» The differences among the choices are:

1. the different role lists (see **Figure 154**, page 311), but they have no effect on computation except

2. The first two kinds of cases include the *calculate paternity/maternity* option §V.B.7 which recognizes M,C,F, role-letter combinations for paternity computation. (**Note** that M is the mother

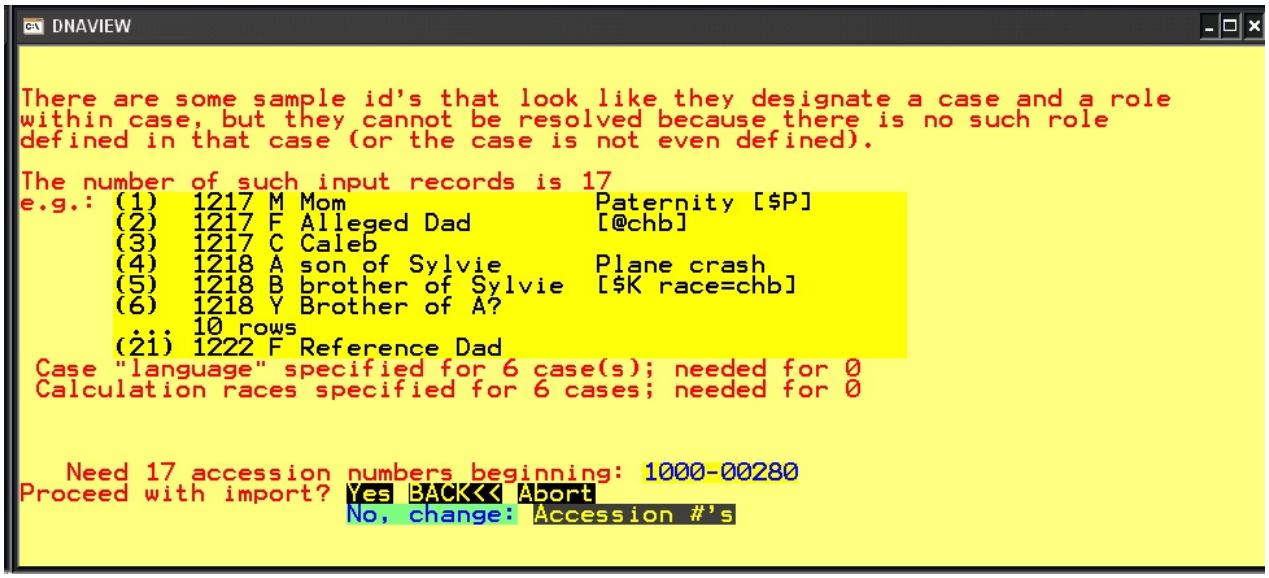



**Figure 119** *Genotyper/GeneMapper import* case definition

for a paternity case, but the *father* for a maternity case – §XVI.A.1.a.)

» The kind of case can easily be changed (§V.B.15), so don't worry if the import file contains several different kinds of case. For now just make the plurality selection.

X.A.2.h.ii.) `Specify computation race(s) to associate with the cases` **`cb`**

Choose one or several race letters appropriate for the plurality of the cases being imported. It will be easy to change the choice case-by-case later (§V.B.13).

☞ Note that either or both of the above two questions will be skipped if the input doesn't imply creation of any new cases, or if all the new cases have comments which tell what the case kind (or "language") and computation races should be per §X.A.7.e.ii.

X.A.2.h.iii.) `Generate 10 accession numbers beginning` **`1000-90345`**

Just ***enter***.

Unless the user included sample numbers as part of the Sample Info (or Specimen ID) field, computer-generated sequence numbers will be supplied. You may ignore these numbers and will never need them to refer to samples – the case+role notation (§XIV.A.9.a.iii) is always available instead – but for historical reasons the numbers must exist internally.

To avoid any conflicts, allow the program to increment its counter sequentially forever, starting in the 1000-... range, and never manually assign any number 10yy-sssss. That should not be a problem because DNA was only discovered long after the Norman Conquest.

X.A.2.i. The data is imported.

» A log of the importing is appended to the import report.

» Quality control (i.e. K562) size checking is implemented, just as with *Read*.

### X.A.3. File formats

Input files for *Genotyper/GeneMapper/Hitachi StarCall Import* contain substantially the same data in different arrangements. The following sections present the various formats, then discuss the common features

### X.A.4. *Genotyper import* file format

| Sample Info | Case comment | D3S1358 1 | D3S1358 2 | vWA | | FGA | AMEL |
|---|---|---|---|---|---|---|---|
| LADDER | | 12 | 13 | 11 | 12 | 18, 19 | XY |
| 1-564 | | 14 | 15 | 17 | 18 | 23 24 | XX |
| A blank or comment row like this is effectively ignored | | | | | | | |
| 2000-1814 | | 14 | 14 | 17 | 18 | 21 21 | X |
| 200007067 C | Bahama routine | 15 | 15 | 16 | 19 | 20, 21 | XY |
| 200007067 D | paternity | 17 | 18 | 15 | 19 | 21 21 | XY |
| 200007067 F | case | 14 | 15 | 15 | 19 | 19 21 | XY |
| 200008905M1 | Two alleged | 18 | 18 | 15 | 16 | 22 24 | XX |
| 200008905AF1 | daddies | 15 | 18 | 16 | 17 | 22 22 | XY |

**Figure 120** Genotyper import file format

**Figure 119** is an example of the *Genotyper import* version of the import format illustrating some of the permissible variations.

Each row represents one sample – a person/mixture/etc including a measurement of the DNA profile for that sample.

X.A.4.a. Build an input file.

A file like **Figure 119** can be built using the ABI Genotyper software

X.A.4.a.i.) In the Genotyper software, fill in the Sample Info according to the ideas of **Figure 123**.

X.A.4.a.ii.) Use the "Make allele table" option (Apple-5), then select the Table tab to export it

» as tab-delimited text if possible. If not,

» as an Excel table. Then open it in Excel and save it as tab-delimited text.

X.A.4.a.iii.) If necessary move it to the PC where DNA·VIEW is.

X.A.4.b. Proceed as discussed under §X.A.1

**X.A.5.** *GeneMapper import*

The first step of the import wizard will help you ignore these first few lines of the file.
The next step decides on the interpretation of the column headings – e.g. "Sample Name" is DNA·VIEW's "Sample Info"
The next step is the locus name interpretation.

| Sample File | Sample Name | User Defined | Marker | Allele | Allele | Height | Height |
|---|---|---|---|---|---|---|---|
| A01_3030 | 050243_M | Raskolnikov | D8S1179 | 12 | 13 | 1510 | 1622 |
| A01_3030 | 050243_M | | D21S11 | 29 | 31.2 | 2000 | 1700 |
| A01_3030 | 050243_M | | D7S820 | 7 | 13 | | |
| A01_3030 | 050243_M | | CSF1PO | 10 | 13 | | |
| | 050243_C_Chip | Murder [lang=C] | D8S1179 | 8 | 9 | | |
| | | | D21S11 | 24 | 24.2 | | |
| | | | D7S820 | 6 | 7 | | |
| | | | CSF1PO | 6 | 7 | | |
| A03_3034 | 050243 F | Case [r=cbj] | D8S1179 | 14 | | | |
| A03_3034 | | | D21S11 | 30 | | | |
| A03_3034 | | | D7S820 | 8 | 11 | | |
| A03_3034 | | | CSF1PO | 11 | | | |
| A04_3035 | 050244 A | | D8S1179 | 12 | 14 | | |
| A04_3035 | | | D21S11 | 28 | | | |
| A04_3035 | | | D7S820 | 10 | | | |
| A04_3035 | | | CSF1PO | 10 | 14 | | |

**Figure 121** *Genemapper Import* example file

The GeneMapper file has about the same essential information as the Genotyper Allele Table, but in a more verbose format as is illustrate by the example file of **Figure 121**. (This file is included as part of a DNA·VIEW installation or update with the name `\dnaview\examples\gmapper.txt`.

X.A.5.a. GeneMapper format comments

X.A.5.a.i.) Sample Info

» One column, usually called **Sample Info** or **Sample Name**, contains the specimen/person identifier for DNA·VIEW. Most efficient is to configure this information in the ABI software at "collection time", but it can also be added by manual edition in Excel after the file is exported from the ABI software. §X.A.2.g.i explains how the input wizard allows any name for this column.

» The specimen id can either be replicated for every row of DNA data for that specimen, or, you can leave a cell blank to indicate a continuation of the data above.

» Any name is acceptable for the column as the DNA·VIEW input wizard allows you to choose (§X.A.2.f).

X.A.5.a.ii.) Comment/specifications

» Optional. A column usually called **Comment** or **User Defined** (the latter name is easier to arrange with GeneMapper[18]) as discussed in §X.A.7.e.

X.A.5.a.iii.) Genotypes

» Each specimen/person takes multiple rows

» The genotype for one locus can be on one line – with several "allele" columns – as illustrated, or, alternatively, on several lines with the alleles arrayed vertically.

X.A.5.a.iv.) (Peak) height/area

If the optional peak height or area information is provided, then the data and columns provided must be conformal with the allele data and columns.

Whether you actually supply peak height or peak area doesn't matter to DNA·VIEW.

X.A.5.b. Build an input file.

A file like **Figure 121** can be built using the ABI Genotyper software

X.A.5.b.i.) In the GeneMapper or Genotyper software, fill in the Sample Info according to the ideas of **Figure 123**.

X.A.5.b.ii.) Save in the appropriate format

X.A.5.c. Proceed as discussed under §X.A.1

**X.A.6.** *Hitachi STRCALL Import*

The *Import/Export* option *Hitachi STRCALL import* is substantially the same as the *Genotyper Import* (§X.A.1, page 226), except for slight differences in the acceptable file format.

| | | Population | Penta E | | D18S51 | | D21S11 | |
|---|---|---|---|---|---|---|---|---|
| Lane No. | Specimen No. | Group | 1 | 2 | 1 | 2 | 1 | 2 |
| Lane2 | 12345 af | | 7 | 12 | 14 | 14 | 29 | 33.2 |
| Lane3 | 1492-1 | | 7 | 12 | 10 | 14 | 29 | 30 |
| Lane5 | 1492-2 | | 13 | 14 | 16 | 20 | 28 | 31.2 |
| | | | 13 | 14 | | | | |
| Lane6 | 1492-3 | | 5 | 12 | 17 | 18 | 28 | 30 |

**Figure 122** Hitachi STRCALL input file format

X.A.6.a. Preparation

X.A.6.a.i.) Create an Excel table of alleles using the STRCALL software.

X.A.6.a.ii.) Export the "Merged Landscape" page from Excel as discussed in §X.A.1.

X.A.6.b. Input file format

[18] GeneMapper allows you to create a *comment* column but beware! It won't be output. You're better off selecting *User Defined* for the column in which you put the case comment and parameter information. And DNA·VIEW will look for that column as well.

The input file is a tab-delimited text version of Hitachi's "Merged Landscape", as illustrated in **Figure 122**.

X.A.6.b.i.) Column headings

The column headings are arranged in a two-row format

» The **locus names** (`Penta E, D18S51`, etc.) are on the first line, and each of them is assumed to apply to one (or more) columns to its right. The allele numbers, 1 and 2 in the second row, are essentially ignored.

» The key word **`Lane No.`** in the second row designates the column containing the lane numbers.

» The key word **`Specimen`** in the second row designates a column with patient id information.

X.A.6.b.ii.) Data

» Each row of data must have a lane number such as **`Lane2`** or **`Lane 2`**. (Unlabelled lanes can be, according to Hitachi, either alternate assays of the same sample, or continuation lines containing extra alleles. On an Excel spreadsheet the two possibilities are distinguished by colors, but since DNA·VIEW has no way to tell which is which the line is ignored. Hand checking is therefore advisable.)

» The *Specimen Id* information follows the same format and rules of interpretation as explained in **Figure 123**, page 236.

X.A.6.c. Proceed as discussed under §X.A.1

### X.A.7. File format details

X.A.7.a. Excel vs. Tab-delimited text

It depicts an Excel table, but since it must be converted to Tab-delimited text before it can be imported to DNA·VIEW, the vertical lines really correspond to tab characters.

The allowable format is quite flexible.

X.A.7.b. Genotypes

The alleles for a locus can be entered

X.A.7.b.i.) in several columns, one allele per column the way the ABI Genotyper software does it, as exemplified in the figure by D3S1358 and VWA.

To cater to mixtures, the number of columns is not limited.

X.A.7.b.ii.) in one column

» separated by ***space*** such as `23 24`. They can also be separated by comma space, as `18, 19`. (Comma alone doesn't work.) Note FGA in **Figure 119**. Any number of alleles are allowed.

» The ***space*** is not needed for SNP's or other systems with 1-letter names like Amelogenin

An allele name like **`<10`** or **`>22`** is translated to 1 or 99.

X.A.7.b.iii.) **Illegal alleles; important note:** The treatment of an allele notation such as **`OL`** depends on the *OFFLADDER Options* policy per §IX.F.25.

X.A.7.c. Locus column headings

X.A.7.c.i.) Locus names can be abbreviated. The program will consult the list of active loci to guess the intention, and in any event the user has an opportunity during import to clarify any mis-guesses.

X.A.7.c.ii.) A blank column heading is assumed to apply to the same locus as its left-hand neighbor. (Note vWA.)

X.A.7.c.iii.) If not blank, a continuation column must abbreviate the name the same way, except for a space-number suffix (such as the " 1" and " 2" after D3S1358), which is ignored.

X.A.7.c.iv.) If columns for the same locus occur *non*-consecutively, then they are taken to represent two different assays. Both are imported and any inconsistencies would come to light when the data is used for a calculation.

X.A.7.d. Sample Info column

A column (usually) with the title "Sample Info" is used to enter roster information (§X.A.8).

X.A.7.e. Comment column (optional)

The optional comment column is for a comment that applies to the case as a whole. (Any comments specific to a single sample should be put at the end of the Sample Info cell, as a "name or comment".)

If (as is the usual situation) the case occupies several rows, you may if you wish spread the case comment among the rows as on **Figure 121**.. When DNA·VIEW imports it will string together all the different comment fragments as if they were put in a single cell and separated by spaces. (But if a fragment is duplicated – as is likely when using GeneMapper software – only one instance will be imported.)

A comment cell can contain two kinds of information – a case comment, and case parameters for the type and computation races of the case. Example

| Sample Info | Comment | Marker | ... |
|---|---|---|---|
| 91023403F_General_Lopez | Lopez incest study [genre=P races=dch] | D3... | |

X.A.7.e.i.) The case comment (**Lopez incest study**), which

» appears and can be searched on in the DNA·VIEW *Paternity/Automatics Kinship ... Case* command

» Is placed by PATER at the top of the report.

X.A.7.e.ii.) The **case parameters** in brackets – for example **[genre=P races=dch]** are codes to indicate the language and computation races (1 or more 1-letter race codes) for the case. There are two possible case parameters:

» the type (or "language" or genre) of case (i.e. **P** for paternity, **M**, **C**, **K** for maternity, crime, or kinship). Example: **genre=P**. Acceptable alternatives to the keyword **genre**= include **kind=, language=, type=** or **$** (without **=**);

» the computation races – **races=** or **@** (without **=**) then up to six race letters. Also, you may shorten the keyword as much as you like so **r=chb** is an acceptable race list specification.

For futher examples and hints note **Figure 121**. **Raskolnikov murder case [language=c races=cbj]** means a *crime case*, computation races **cbj**, with a case comment **Raskolnikov murder case**. Same for **Raskolnikov murder case [ $C @cbj ]** or even **Raskolnikov [ @cbj ] murder [k=C ] case**

It may be most efficient to spell out in the input file only the *exceptional* case parameters. If most cases are the same, no need to label them because there are other ways to specify these parameters:

» If omitted, the default values for the worklist/project are used – which the user may edit at the time of input.

» Anyway the case parameters can be adjusted later, case-by-case, within the *Paternity (etc) Case* program.

### X.A.8. Defining the roster

The allowable **Sample Info** entries can be summarized by the notation:

{*person-id*}{*case + role letter* {*description*}},

where { } represent optional items. That is, the above notation means that any of the following options are acceptable:

*person-id*

*person-id* *case+role*

*person-id* *case+role* *description*

*case+role*

*case+role* *description*

X.A.8.a. *person-id* – means a DNA·VIEW accession number (person number) with a dash in it, such as `2004-101`,

X.A.8.b. *case + role letter* – means a case number followed by a role letter (upper or lower case ok). An intervening ***space*** (or ***underscore*** – §X.A.8.d) is optional; either `4767C` or `4767 C` is ok. *After* the role letter though please do put a space or underscore to separate it from any following description.

X.A.8.b.i.) role letter substitutions

In addition to the standard DNA·VIEW 1-letter roles such as `C, D, E`, ... for the first few children in a paternity case, you may use `C1, C2, C3`, ... . Similarly you may put `F1, F2`, ... instead of `F, G`, ..., and so on. Also, `AF` is acceptable instead of `F`.

X.A.8.c. *description* – Any text following the *case+role letter* is considered as a description for the person. This can be the person's name or any sample documentation. Up to 40 characters are accepted.

| style | **Sample Info (or Name) (or Specimen)** | **Remarks** |
|---|---|---|
| person/patient number | `1-562`<br>`2000-1813` | As usual, leading 0's after the dash (–) may be omitted. |
| role-within-case (DNA·VIEW style) | `4767 C`<br>`4767D`<br>`4767 f Henry`<br>`2003-00123 4767 M Sophia` | i.e. first child of case 4767.<br>Upper or lower case ok.<br>Space(s) between case # and role are optional.<br>C=C1, D=C2 etc.<br>f=f1=af=af1, g=f2=af2, etc. |
| role-within-case (various acceptable styles) | `48905 M1`<br>`48905 AF1`<br>`48905 af2 Accused man`<br>`48905_af2_Accused_man`<br>etc. | ***Underscore*** is same as ***space*** |

The way to label quality control samples is to use their DNA·VIEW accession number code, such as `1-562`.

**Figure 123** Example allowable ID formats for roster import

**`2003-00123 4767 M Sophia`** includes all three options, and means that this is person number 2003-00123, that case 4767 with a role **M** will be created if it does not exist already, this number and the name "Sophia" will be assigned to that role.

**`4767 f Henry`** uses the person number of case 4767 role F if it exists; otherwise the computer creates the role (and case if necessary) and assigns a sequential accession number.

**`2000-1813`** associates this person number with the DNA data. The number is not automatically inserted in any case, though it may of course already belong to a case.

X.A.8.d. Using underscores ( _ ) for ***space***

The GeneMapper software doesn't allow the user to type in spaces[19] – that's why so there are so many underscores ( _ ) in sample codes (e.g. the **Sample File** field in **Figure 121**). Therefore, DNA·VIEW accepts the underscore as a ***space*** for purposes of the `Sample Info`.

### X.A.9. DNA·VIEW and Osiris

(from version 33.07 – 2014)

Profile data can be exported from Osiris and imported into DNA·VIEW.

Add an export option to Osiris as explained below which will create data files including RFU values acceptable to DNA·VIEW for import using the "Genotype" import option.

The DNA·VIEW "Genotyper" file format (§X.A.4) can include, in addition to allele sizes, also rfu (and other) information.

X.A.9.a. What to export from Osiris

X.A.9.a.i.) Export anything that might be an allele

[19] GeneMapper™ (and Genotyper as well?) disallows the following symbols: `\ / : * " < > | ? ' space`. Therefore presumably `! # $ % & ( ) [ ] { } + - . , ; = @ ^ _ ~` are permitted.

For DNA·VIEW mixture calculation, data should be exported from Osiris with minimal filtering. A low detection threshold such as 35rfu is desirable. Signals that look like stutter should be exported allowing DNA·VIEW to make decisions.

X.A.9.a.ii.) But do use Osiris' clever artifact diagnostics

» I think it is proper to allow Osiris to filter out (not export) signals for which Osiris' base-pair estimate is not close to an integer, as representing trash rather than true alleles. Osiris uses the word "residual" to mean the error between estimated base pair size and the nearest integer.

» Similarly, Osiris' parameter Fit<0.98 (possibly even Fit<0.99 from limited experience) is probably diagnostic of illegitimate data.

Osiris version 2.3 seems to allow automatic filtering based on a residual threshold, but require manual editing to utilize the Fit data.

X.A.9.b. Osiris "Lab settings" configuration

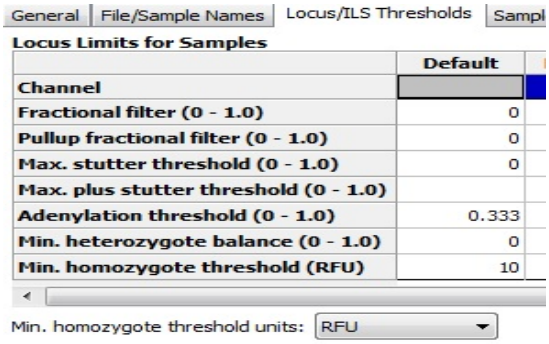
Create customized Osiris lab settings operating procedures (in Osiris: Tools ▸ Lab settings) by modifying standard Osiris operating procedures (NEW... ▸ **Select an operating procedure to copy**).

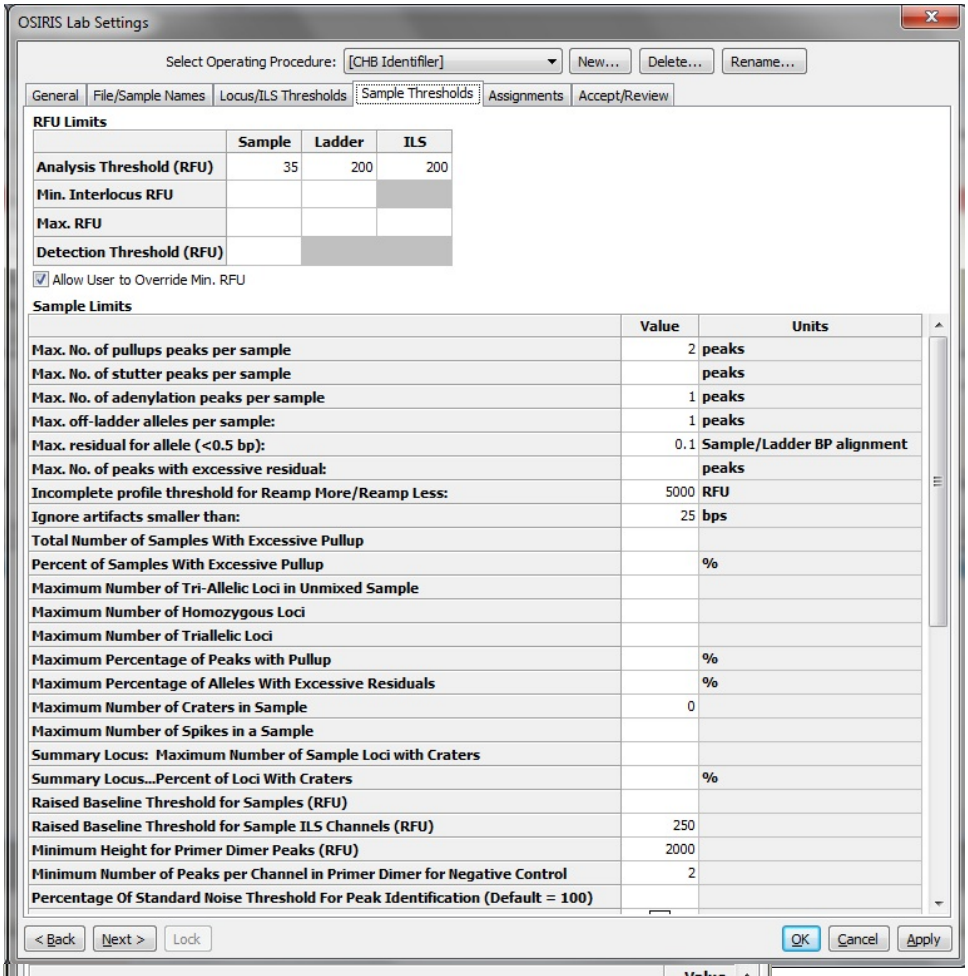
The following screen shots show my parameter choices. Note that the choice **fsa** vs. **hid** is part of the operating procedure (in the **General** tab). To switch between these formats I manually edit the operating procedure.

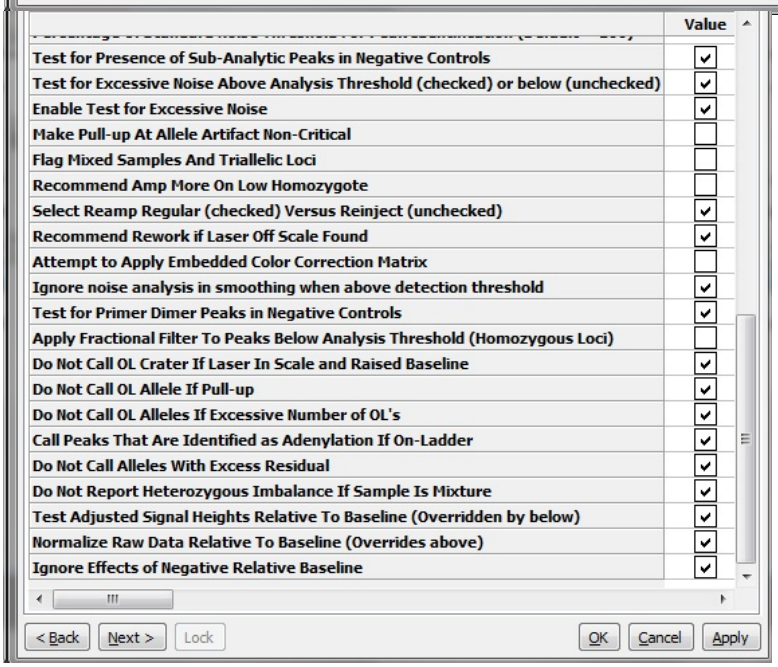
X.A.9.b.i.) Fractional filter

» For a mixture, Osiris' fractional filter must be 0 or unused so DNA·VIEW will see even relatively small peaks.

» For mixture reference profiles 0 is ok (and will be preferable for a post-33.08 version which will use the stutter clues)

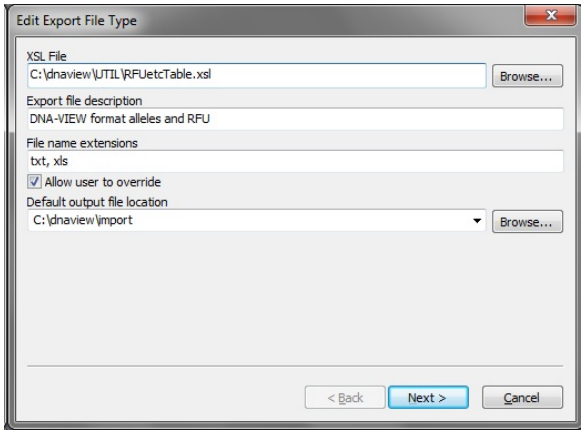



OSIRIS Lab Settings
Select Operating Procedure: [CHB Identifiler]
New...
Delete...
Rename...
General
File/Sample Names
Locus/ILS Thresholds
Sample Thresholds
Assignments
Accept/Review
RFU Limits
Sample
Ladder
ILS
Analysis Threshold (RFU) 35 200 200
Min. Interlocus RFU
Max. RFU
Detection Threshold (RFU)
Allow User to Override Min. RFU
Sample Limits
Value
Units
Max. No. of pullups peaks per sample 2 peaks
Max. No. of stutter peaks per sample peaks
Max. No. of adenylation peaks per sample 1 peaks
Max. off-ladder alleles per sample: 1 peaks
Max. residual for allele (<0.5 bp): 0.1 Sample/Ladder BP alignment
Max. No. of peaks with excessive residual: peaks
Incomplete profile threshold for Reamp More/Reamp Less: 5000 RFU
Ignore artifacts smaller than: 25 bps
Total Number of Samples With Excessive Pullup
Percent of Samples With Excessive Pullup %
Maximum Number of Tri-Allelic Loci in Unmixed Sample
Maximum Number of Homozygous Loci
Maximum Number of Triallelic Loci
Maximum Percentage of Peaks with Pullup %
Maximum Percentage of Alleles With Excessive Residuals %
Maximum Number of Craters in Sample 0
Maximum Number of Spikes in a Sample
Summary Locus: Maximum Number of Sample Loci with Craters
Summary Locus...Percent of Loci With Craters %
Raised Baseline Threshold for Samples (RFU)
Raised Baseline Threshold for Sample ILS Channels (RFU) 250
Minimum Height for Primer Dimer Peaks (RFU) 2000
Minimum Number of Peaks per Channel in Primer Dimer for Negative Control 2
Percentage Of Standard Noise Threshold For Peak Identification (Default = 100)
< Back
Next >
Lock
OK
Cancel
Apply


Value
Test for Presence of Sub-Analytic Peaks in Negative Controls
Test for Excessive Noise Above Analysis Threshold (checked) or below (unchecked)
Enable Test for Excessive Noise
Make Pull-up At Allele Artifact Non-Critical
Flag Mixed Samples And Triallelic Loci
Recommend Amp More On Low Homozygote
Select Reamp Regular (checked) Versus Reinject (unchecked)
Recommend Rework if Laser Off Scale Found
Attempt to Apply Embedded Color Correction Matrix
Ignore noise analysis in smoothing when above detection threshold
Test for Primer Dimer Peaks in Negative Controls
Apply Fractional Filter To Peaks Below Analysis Threshold (Homozygous Loci)
Do Not Call OL Crater If Laser In Scale and Raised Baseline
Do Not Call OL Allele If Pull-up
Do Not Call OL Alleles If Excessive Number of OL's
Call Peaks That Are Identified as Adenylation If On-Ladder
Do Not Call Alleles With Excess Residual
Do Not Report Heterozygous Imbalance If Sample Is Mixture
Test Adjusted Signal Heights Relative To Baseline (Overridden by below)
Normalize Raw Data Relative To Baseline (Overrides above)
Ignore Effects of Negative Relative Baseline
< Back
Next >
Lock
OK
Cancel
Apply

X.A.9.c. Osiris export for DNA·VIEW

X.A.9.c.i.) **DNAVIEW\util\RFUetcTable.xsl** is a custom DNA·VIEW export template for Osiris as discussed in Appendix E of the Osiris documentation.




X.A.9.c.ii.) Osiris "Export file type" configuration

The Osiris manual, "Export Setup Tutorial" explains the procedure. Briefly:

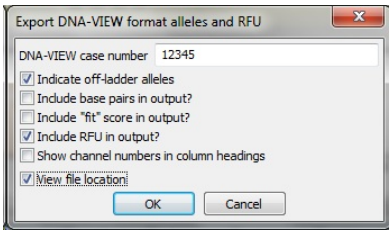
In Osiris, **Tools** ▸ **Export file settings** ▸ NEW... and follow the screen shot at right.

The remaining screens should fill correctly automatically. Now you have a new export menu item in Osiris: **File** ▸ **Export** ▸ **DNA-VIEW format alleles and RFU ...**

X.A.9.d. Exporting data – preparation in Osiris

X.A.9.d.i.) Open the ***.oer** file, review the data and delete (uncheck) noise such as bad Fit that won't be automatically filtered.

X.A.9.d.ii.) **File** ▸ **Export** ▸ **DNA-VIEW format alleles and RFU ...**




» Check **RFU** to provide DNA·VIEW with intensity information.

» The **base pairs** and **"fit"** boxes are for future development; DNA·VIEW doesn't consider that data at present.

» The case number – whatever you type which can include DNA·VIEW *Case number formatting* if you use it – will be included in the output file.

» Role letters aren't included in the output so you have to add them manually; hence the check mark in **View file location**.

X.A.9.d.iii.) Manually editing the output file

Open the exported text file in a text editor or (safest) in Excel.

» Wherever you see `(role?)`, substitute a suitable role letter. Don't bother for controls or ladders which are of no interest to DNA·VIEW and are not part of a case. (It is unnecessary though harmless to delete uninteresting lines.)

» Add any comment including language and race codes for the case in the first (`Comment`) column (per §X.A.7.e). Be sure not to add or delete any tab characters. (That's the reason Excel is safest).

X.A.9.e. Import into DNA·VIEW

X.A.9.e.i.) Choose whatever treatment of off-ladder alleles is appropriate (§IX.F.25) – probably `Treat as a rare allele`.

X.A.9.e.ii.) *Import/Export* ▸ *Genotyper import* per §X.A.1.

## X.B. *Import/Export – database*s

This section discusses entering "databases" – population data on allele frequencies – into DNA·VIEW. See also the discussion "Ways to make a database" (§VIII.C.3) about alternatives such as typing the population data in and creating a database by compiling DNA·VIEW data.

### X.B.1. *Allele frequency (or count) population data import*

(revised – 2014) allows flexible possibilities for importing population data such as is found in journals. A typical example:

Table 1

Allelic frequencies and forensic parameters of 17 STR loci from six provinces of Argentina

| | D3S1358 | TH01 | D21S11 | D18S51 | PENTA E | D5S818 | D13S317 |
|---|---|---|---|---|---|---|---|
| | (N=639) | (N=639) | (N=639) | (N=639) | (N=639) | (N=639) | (N=639) |
| 2.2 | - | - | - | - | - | - | - |
| 5 | - | - | - | - | 0.035 | - | - |
| 6 | - | 0.242 | - | - | - | - | - |
| 7 | - | 0.246 | - | - | 0.125 | 0.025 | - |
| 8 | - | 0.102 | - | 0.001 | 0.015 | 0.008 | 0.122 |
| 9 | - | 0.161 | - | - | 0.008 | 0.04 | 0.115 |
| 9.3 | - | 0.235 | - | - | - | - | - |
| 10 | - | 0.013 | - | 0.01 | 0.089 | 0.059 | 0.081 |
| 11 | 0.001 | 0.001 | - | 0.013 | 0.112 | 0.392 | 0.257 |
| 12 | 0.001 | - | - | 0.135 | 0.193 | 0.314 | 0.267 |

• • •

**Figure 129** Population database suitable for importing

X.B.1.a. Input file format

The file may be conveniently created in Excel, but for DNA·VIEW's purposes export it as tab-delimited or comma-delimited (`.csv`).

X.B.1.a.i.) Each column may be either

» data for a locus, preferably titled by the locus name

» allele sizes for one, some, or all of the loci

» a race or population designation

» or ignored

X.B.1.a.ii.) Rows are either

» The title row (D3S1358 etc)

» data – alleles and the frequencies or, preferably, counts (=number of observations)

» or ignored

X.B.1.b. Operation

Importing the file and creating population databases proceeds by stepping through the import wizard.

X.B.1.b.i.) **pdf import**. To import from a pdf document (sometimes data comes from a journal and the article is only available in pdf format):

» Open the document in Adobe. ***ctrl-a***, ***ctrl-c*** to select and copy the *entire* document.

» Open Word or similar and paste the clipboard.

» Cut away everything except the table with the data.

» If necessary introduce tabs to restore alignment of any mis-aligned columns.

» Save the file as plain text.

X.B.1.b.ii.) **Preliminaries**. These preparatory steps are not required but may make the importing more efficient and less confusing.

» Trim the import file of most of the superfluous rows.

» Decide which race code(s) you will use and be sure the race names (§IX.A.4) are defined.

X.B.1.b.iii.) `Site name for these databases?`

By default the data is assumed to be generated by your own lab, in which case your world-wide unique DNA·VIEW `Site` name will be the first part of the database name. See **Figure 71**. That's why the prompt refers to `Site`. In another situation I might put a journal reference such as `JFS 44(6)` as the "`Site`".

When you return to this prompt after finishing the import of a data file, you may ***ctrl-c*** to quit to the main menu. Or, you can enter the same or a different `Site` for the next input data file.

X.B.1.b.iv.) Select the file to import as in §X.A.2.b.

» If you have just imported a file you may meet the time-saving prompt

`Reuse same import\Italy.txt?`

Answer **`yes`** to continue at step §X.B.1.b.vi, with the garbage rows removed.

» If the file is comma-delimited (`.csv`) format, when asked `Convert comma to tab?` answer **`y`**.

X.B.1.b.v.) **Edit and accept the file**. A sample of the file is shown on the screen and a dialogue permits you to eliminate a few extra rows.

» `Does it look like the right file?` **`y`**

» `Enter [line numbers] to remove` **`1 2 4 60 61`**

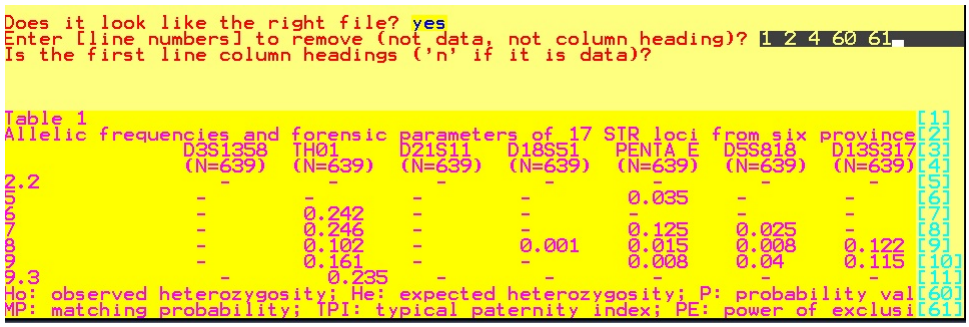



**Figure 130** Enter [line numbers] to remove

Type in the numbers of any lines from the file sample displayed on the screen that are unnecessary to the import – comments, ancillary statistics – using the bracketed numbers on the right side of the screen.

You are given repeated chances at this step so it's good enough to eliminate a few lines each time (but recall §X.B.1.b.ii).

» `Is the first line column headings?` **`y`**

It's best to retain a row of `column headings` – the locus names – at the top. Aside from that one row, every other row must be data.

X.B.1.b.vi.) DNA·VIEW recognizes the columns. If all has gone well, you will have a screen with vertical colored stripes representing the columns of the data file. The letters `a b c d ...` designate the columns. You have two options.

» To select a subset of the rows corresponding to some race, type the single column letter of the

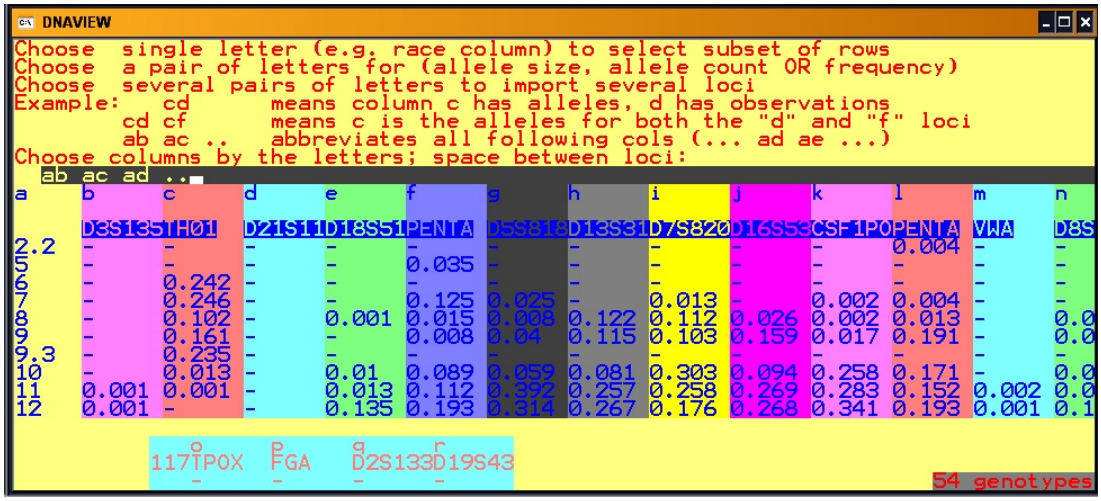



**Figure 131** Selecting desired columns for database import

column containing the race codes and proceed per §X.B.2.e.

» To import one or more databases, type one or more pairs of letters, spacing between the pairs as shown in **Figure 131**. Each pair designates two columns with the allele sizes, and the allele frequency or count, for a locus, such as **`ab ac ad ...`**(the ellipsis dots are permitted and signal that the same pattern should continue through all the loci. The ellipsis pattern **`ab cd ef ...`** is also recognized.)

***Enter*** to proceed.

X.B.1.b.vii.) **Locus interpretation screen** is the same as §X.A.2.g.

X.B.1.c. Import of population data one locus at a time (Operation).

One locus at a time you now import the population databases. There are several screens for each locus.

X.B.1.c.i.) **Scale the counts**. DNA·VIEW wants allele counts, not frequencies. Therefore if the input data is in frequencies it's necessary to multiply it by the appropriate scaling factor. The program requests the number of *people*; then it multiplies by double that number to account for the pairing of chromosomes.

Most published allele population studies have the frequencies and a claimed but frequently inaccurate number for the number *n* of people typed. The program gives assistance in guessing the accurate number for each locus.

If the import file has counts rather than frequencies, this step is skipped.

X.B.1.c.ii.) **Select the race, file the database** exactly as per §X.B.2.i and §VIII.C.5.e.

---

**X.B.2.** *Genotype table of population sample. (Pair of columns=genotype for a locus)*

creates DNA·VIEW allele frequency population databases from tab-delimited text files such as those produced by Genotyper (after exporting the Excel file as tab-delimited text).

This method is the preferred method if possible because it creates the database from the raw data. Consequently

» the allele counts are sure to be accurate ( published information often is not);

» the import program can find and report near-duplicate samples.

| a | b | c | d | e | f | g |
|---|---|---|---|---|---|---|
| Sample Info | D3S1358 | D3S1358 | vWA | vWA | FGA | FGA |
| WI 1 | 15 | 16 | 14 | 15 | 19 | 21 |
| WI 2 | 17 | 18 | 17 | 17 | 19 | 25 |
| WI 3 | 16 | 17 | 18 | 18 | 20 | 23 |
| BL 4 | 15 | 16 | 15 | 20 | 23 | 26 |
| etc . . . | | | | | | |

DNA·VIEW's display of the Genotyper-format import file adds the "a b c ..." headings and the shading.

**Figure 132** File format for importing genotypes

Choose *Import* from the main menu, then select *Genotype table of population sample*. The importing steps are then as follows:

X.B.2.a. *Specify the Site name* which will be associated with the database created from the imported data. When the data is published I use a publication reference, sometimes as long as **`FSI138(2003)p134`**.

X.B.2.b. *Specify the subdirectory* where the file(s) reside.

X.B.2.c. *Select a file* to import, and choose to *retrieve* it. There is some flexibility permitted in the input file format but roughly it should look like **Figure 132**.

X.B.2.d. An iterative dialogue allows you to remove some irrelevant comment lines from the file. Besides the data rows (one per person), it is ok (in fact best) to retain a row of column headings.

X.B.2.e. (optional) *Specify the rows to import* – the FILTER button.

X.B.2.e.i.) Subset of rows by ethnic category

One of the columns, for example column ***a***, may be a category description for the person/genotype in the row. At any rate, click FILTER then select column ***a*** and you are given the option to choose which categories to keep, using the "Multiple selection" method, §XIV.A.3. Note that the method conveniently permits

» Selecting one race (or any arbitrary category)

To select only those rows of **Figure 132** containing a "WI" code as part of the first column, specify the context **`WI`** in the pop-up window and ACCEPT. Repeated subselections are permitted to narrow the choices further.

» Selecting several races or categories – Two phrases can be entered together vis: **`WI&BL`**

» Dropping an unwanted sample or set of samples – the REMOVE button.

To eliminate for example sample `DB2003-01811-xyz` it might be sufficient to REMOVE **`01811`**.

X.B.2.e.ii.) **Random subset of rows** (useful only for demonstration) – the RANDOM button.

Click RANDOM then give a number in response to the prompt

```
Select how many random people?
```

X.B.2.f. *Specify the columns to import*. Designate (by letter codes, e.g. **de**) pairs of columns to import as a database. The program later makes a guess as to the locus based on the column headings if present, which the user can confirm or correct.

X.B.2.f.i.) Two formats are allowed:

» Select pairs of columns, one pair for two alleles for each locus – such as **bc de fg**.

» Select a sample id column plus pairs of columns for loci – **a bc fg de**.

X.B.2.f.ii.) Alternatives for convenience:

» **Ellipsis abbreviates** remaining loci: Typing e.g. **bc de ...** is shortcut for the remaining column pairs.

» **Interactive menu** selection of loci: ***Enter***, with no text, to select column pairs for import from a menu. Each selection from the menu represents two consecutive columns, so this method does not allow designating a "sample id" column. Re-selecting the same column removes the previous selection (correcting a mistake). Type ***Esc*** when finished selecting to continue to the next step.

X.B.2.g. *Locus identification*

A locus-identification chart/menu is displayed (like **Figure 139**) with DNA·VIEW's guesses as to the loci named in the column headings. Check the chart, and correct any misunderstandings using the same procedure as in §XIII.C.4.

X.B.2.h. Sample id's

If any column was designed as sample ID (by virtue of an unpaired letter in §X.B.2.f.i), then any duplicate sample id's will be reported, and cause import to be aborted. Also, DNA·VIEW generates internal sample numbers from the sample ID's which can later be used by *List and search database profiles* (§X.E.1) to regenerate the multiple-locus genotypes from several databases.

X.B.2.i. (optional) Check for duplicate or similar profiles.

```
 Selected genotypes are 7 locus profiles.
Shall I check for similar genotypes? y
 1 pair of specimens match at 7 / 7 loci.
row   Seq#  #D3S D3S D16 D16 AM AM THO1 THO1 TPO TPO CSF CSF D7S D7S
[115] 5273  15   18  11  12  X  X  7    9    8   9   10  12  9   10
[128] 4704  15   18  11  12  X  X  7    9    8   9   10  12  9   10

 15 pair of specimens match at 5 / 7 loci.
 151 pair of specimens match at 4 / 7 loci.
 1222 pair of specimens match at 3 / 7 loci.
 4750 pair of specimens match at 2 / 7 loci.
 8793 pair of specimens match at 1 / 7 loci.
No duplicate specimen numbers.
```

**Figure 133** Checking import database for duplicates

See **Figure 133**.

X.B.2.j. Iteratively per locus, create a database.

X.B.2.j.i.) Choose the race letter to be associated with the database.

X.B.2.j.ii.) File the database (or skip it) as explained in §VIII.C.5.e.

## X.C.

*Import/Export* – sharing databases

Two complementary facilities to allow users to swap databases are described below.

### X.C.1. *DNA·VIEW Frequency Database Import* (transfer between users)

The *Import/Export* option *DNA·VIEW frequency database import* lets you integrate additional, DNA·VIEW supplied databases into your list of databases.

The new databases may be supplied on a disk, with a filename such as `DNAFREQ.SF`, but in any case with the extension `.SF`. The file can either be the `\dnaview\dnafreq.sf` file of a DNA·VIEW system, or can be a file created by the complementary *Export DNA·VIEW databases* (§X.C.2) facility.

You can copy the file to your hard disk for faster operation if you like, but be sure not to overwrite the `DNAFREQ.SF` file that contains your current list. Either copy it to another subdirectory, or change the name, e.g. to `NEWFREQ.SF`. The extension must be `SF`.

Also, you can Import from your Demo system (§XI.E) to populate an empty Production system list of data bases.

X.C.1.a. Select file from which to import

The command first queries for the path and name of the new file.

X.C.1.b. Mass selection of databases to import

Then you are given the opportunity to limit the list of possible databases to import using the "mass selection" method as described in §XIV.A.3. It is often convenient to ***enter***, temporarily selecting all of them; you will later get the chance to narrow the selection.

X.C.1.c. The import process iterates through the remaining choices one at a time.

X.C.1.c.i.) Each new database that is identical to an old one is skipped.

X.C.1.c.ii.) Otherwise, the directory entry for the new database is shown and you are asked `Import?`.

X.C.1.c.iii.) **n** if you don't want it. Then you will again be presented with the remaining list of databases to import, so you can narrow that list systematically. For example – offer & rejection:

```
I can import: (# 30 / 996 )            Import it? n
D3S1358 STR Canada CFS East Indian    167 01/09/21 1 pm
```

Helpful offer:

```
Limit databases again if desired
Accept lines containing ~
 966 databases
```

To reject similar database quickly:

```
Accept lines containing ~CFS                    enter
 958 databases
```

X.C.1.d. **y** to (probably) import the new database

The imported database can either be added to your collection, or can update a database already there. You are presented with a list of candidates for replacement — same probe and same race(s).

» Press ***End*** to make a new entry, or

» select one of the old entries to replace it.

In the case of replacing an old entry, the program will display both the new database and the candidate for replacement side-by-side, and ask for further confirmation before making the replacement, as shown in **Figure 79**, page 165.

After completing the import, you may wish to use *Database ▸ Set default database, per race* (§VIII.C.2.b, page 160) to check that the default race selections are as intended.

### X.C.2. *Export DNA·VIEW databases* (transfer between users)

*Export DNA·VIEW databases* is the inverse of *DNA·VIEW frequency data import* (§X.B).

You are first asked

`Subdirectory for the DNAFREQ.SF file to export to?`

You may choose for example **`A:`**, if you have a good diskette in drive A.

`File name:` Usually choose *(new file)* from the menu.

Then `File name?` Supply a DOS file *prefix* (no dot); the system will add the `.SF` suffix.

The rest of the operation is similar to importing – multiple selection (§XIV.A.3) to get the approximate list of databases to transfer, then prompting for each possible choice.

## X.D. Import/Export – GeneScan & Pharmacia

This chapter on options for importing data by migration amount and having DNA·VIEW compute the size and allele calls has been removed for space. If you are interested older manuals are available on-line.

## X.E. *Import/Export* – miscellaneous options

### X.E.1. *List and search database profiles*

This **Import/Export** option includes several possibilities.

X.E.1.a. List database profiles

Produce a list of phenotypes obtained from one or several databases. If the databases each contain substantially the same people typed in different probes then the list will show the multiple-locus phenotype for each person. *It is only possible with DNA·VIEW-compiled databases* (§VIII.C.5, §VIII.C.7).

X.E.1.b. Search for matching profiles

You may also ask the program to make a special search for partly or completely similar profiles. You may

X.E.1.b.i.) search the entire database for **duplicates** (or, if RFLP, near-duplicates),

X.E.1.b.ii.) search the entire database against one or **a few specific profiles**. This search can be either for full-profile match, or for half-match (one allele at each locus, such as parent and child).

The specific profiles can be

» profiles previously entered into DNA·VIEW, specified by accession ID, AND/OR

» profiles that you may type in.

X.E.1.c. Operation

X.E.1.c.i.) Select a collection of (DNA·VIEW-compiled) databases. To be sensible, they should be different loci for essentially the same collection of people. They are selected using the database "mass selection" method described in §XIV.A.3. Note the possibility to use the α symbol (***alt-a***) to specify default databases.

X.E.1.c.ii.) `Find matches, Report phenotype list, or Both (F, R, B)?` **F**

» **F** If your goal is simply to find matches, you may not want to include the possibly voluminous list of all database profiles in the report.

| ID # | D8S1179 | D21S11 | TH01 | D16S539 | D2S1338 | D19S433 | VWA |
|---|---|---|---|---|---|---|---|
| U 114808 | | 28 28 | | | | | 17 14 |
| 99-00814 | 15 12 | 31 30 | | 10 9 | | 14 12.2 | 16 15 |
| F 50243 | 14 14 | 30 30 | 8 7 | 11 11 | 22 17 | 14 14 | 18 15 |
| M 36508 | 15 14 | 31.2 30 | 7 6 | 12 11 | | | 19 15 |
| C 36508 | | | 11 7 | 11 10 | | | |
| F 36508 | | | 7 6 | 11 9 | | | 19 15 |
| 99-37731 | | | 9.3 6 | 12 11 | | | 18 18 |
| 99-37732 | | | 9.3 6 | 12 11 | | | 18 17 |
| 99-37733 | | | 6 6 | 12 12 | | | 18 17 |
| 100-00001 | 14 12 | 34.2 32.2 | 7 6 | 12 11 | | | 18 17 |

**Figure 134** Database profile report

» **R** Skip the remaining dialogue; create the profile report.

» **B** Create a report with both matches and all the database profiles. The report has the matching report first, then the database profile list.

X.E.1.d. The Matching List

X.E.1.d.i.) `Minimum number of loci to match?` **6**

The point is that some people might be typed in only a few loci. If two people match completely with respect to the selected databases, but one or both of them was only typed in one of the loci, do you still want that match included on the list? If so, answer **1**. More commonly the answer will be **2** or **3** for RFLP and **6** or more for STR data.

This option does *not* mean that some non-matching loci may be ignored. Rather, it is a way to ignore incomplete profiles in order to avoid potentially having a large number of uninteresting matches reported.

X.E.1.d.ii.) `Do you want to include some extra profiles?`

**n** All the phenotypes in the database are compared with one another. Thus, for a database of 1500 people, about 1,000,000 comparisons. The searching is done in a very fast and efficient manner. If you choose this option, the search begins.

**y** In addition, search the database for a few specific profiles that you specify. For a 1500 person database, several thousand comparisons. If you choose this option, there are more questions:

X.E.1.d.iii.) `Match 1 allele or 2 alleles per locus?` **2**

Two possible search modalities:

**1** for a "Paternity search" — people sharing one allele at each locus. If you choose this option the search will **only database vs. entered profiles** – there is no database vs. database paternity search.

**2** "Full search" looks for people matching both alleles at each locus. It compares all profiles in the databases with one another, as well with any extra profiles.

X.E.1.d.iv.) `Accession # to match?`

Profiles that already exist in DNA·VIEW can easily be chosen using their accession # or role+case (per §XIV.A.9.a.iii). Choose as many, one at a time, as you wish.

Simply ***enter*** accepting the "year" number of 0 number to finish.

X.E.1.d.v.) `Would you like to type in some profiles?` **`n`**

This is less convenient, but gives you the chance to search for anything and also to edit/modify and profiles specified in the previous step.

The editing format is two lines per person, one column per locus (because there are two sizes at each locus). Example:

```
                  [0] YNH          [1] TBQ
[0] 93-12345  [0]  1234           2345
[1] 93-12345  [1]     0           3000
[2] Person 1  [2]  1234           2345
[2] Person 1  [3]  2000           3000
```

will find anyone in the databases that matches (2345 3000) in TBQ, and matches either homozygous 1234 or (1234 2000) in YNH. Data entry is using the numeric editor (XIV.B.2.b). Allele notation is allowed. ***Ctrl-e*** when finished.

X.E.1.d.vi.) Example of a matching list

Matching pairs are listed with a score (0.0 = exact match). Example:

```
             [0]      [1]        [2]       [3]      [4]      [5]
    ID #     D8S1179 D21S11     TH01      D16S539 D2S1338 VWA
Average std dev diff/band=0.0 Maximum=0.0
 U 185       15 13   33.2 32.2  9.3 7    14 11    25 17    16   15
 F 185               33.2 32.2  9.3 7    14 11    25 17

Average std dev diff/band=0.0 Maximum=0.0
 1001-02599  15 14   30.2 29    9.3 8    11  9             19   15
 2007-83838  15 14                                         19   15
```

For RFLP searches there are some additional options and parameters. Find an archived old version of this manual if you are interested.

X.E.1.e. Database searching and convicted offenders or missing persons

Three possible methods:

X.E.1.e.i.) A high-powered searching facility *Search NDIS for Relatives* that allows a database of unlimited size and implements not only direct searching but also kinship searching is under development, and will be an extra-cost option.

As currently envisioned it's use is limited to searching and it is not intended for cataloguing offenders nor offender list maintenance.

X.E.1.e.ii.) The *Screen disaster matches* command can be used in the same way but limited to about 100,000 offenders.

X.E.1.e.iii.) The searching method described above for DNA·VIEW compiled databases can be used to maintain offender lists and search for direct matches, for a limited number of offenders. The protocol would involve setting up appropriate DNA·VIEW databases, updating them as necessary, and using the above *List and search database profiles* (§X.E.1) facility to search. The database maintenance protocol is laid out in detail in §VIII.C.4.

The search then proceeds. *Print* or **File** ▸ *Save as* the result report.

### **X.E.2.** *LIMS import of case & person definitions*

Import file specifications are on the web at http://dna-view.com/interface.htm. Accepts text files defining cases and people.

### **X.E.3.** *Case and accession data import*

This is an old and unused facility. See an older manual for documentation. Contact the DNA·VIEW author for newer and more comprehensive approaches to DNA·VIEW – LIMS integration.

## **X.F.** *CODIS: export size data in CMF format*

This rarely referenced section has been removed to save paper. Consult a pre-2007 manuals, available on-line, if interested.

## X.G. Integration of DNA•VIEW within LIMS system

Some of the features of DNA·VIEW can be controlled by a script, which permits using DNA·VIEW as an engine under the control of a LIMS systems. The LIMS creates certain files – a genotyper table with DNA profiles, a script file instructing DNA·VIEW to do certain operations such as read in the DNA data, make paternity calculations, kinship calculations, or kinship simulations. Eventually DNA·VIEW signals completion and stops. Then the results can be collected by the LIMS system and integrated into its database.

As of now establishing the LIMS–DNA·VIEW link is a custom task; i.e. some consulting is involved.

# XI. PROGRAM VALIDATION & MAINTENANCE

## XI.A. Program Validation

Program validation means assuring that the program does what it should. Some aspects of validation:

§XI.A.1 lists some test suites built into DNA·VIEW.

§XI.A.2 lists facilities in DNA·VIEW to document standard settings and detect accidental tampering.

§XI.A.3 suggests a protocol for validation by checking user cases.

§VI.F.8 can be helpful to compare against a published case.

§XI.A.4 discusses database validation.

### XI.A.1. Test suites in DNA·VIEW

The tests in this section were mainly written for the program developer, but are available for the user to run also. They validate by comparing operation of the program against canned results, or against a calculation using a different algorithm. In that way they test whether a program error has been introduced.

XI.A.1.a. Paternity and maternity case calculations (§V.B.30)

**Casework** ▸ *Parentage Case* ▸ (CALCULATE button) *parentage test examples*

Runs a suite (or selected example) of one-locus calculations covering examples of maternity, paternity, duo, trio, mutation, and silent alleles.

XI.A.1.b. Kinship test examples (§VI.K.9)

**Casework** ▸ *Prototype Kinship* ▸ *run test cases; check program*

Derives the formulas for a suite of kinship problem and compares them with canned results.

XI.A.1.c. Mixture formula testing

The DNA·VIEW Mixture Solution – the LR method one (§V.D) – includes two completely different algorithms for deriving the LR formula, so they can be used to test one another. There is a set of canned examples, and you can also test your own examples.

XI.A.1.c.i.) Mixture test suite (§V.D.9)

**Casework** ▸ *Automatic mixture* ▸ (CALCULATE button) *mixture examples*

XI.A.1.c.ii.) Roll your own

Open any of your mixture cases and proceed to the *Locus mixture computation screen* (?) for some locus. Set θ=0 and observe the LR formula and value. Then set θ=0.00001, which will trigger the

alternative, θ-sensitive, algorithm. The formula will look a little different because of the incidence of all the T's (which represent θ), but with θ so small the numerical LR should be identical.

XI.A.1.d. Kinship simulation test case

This one may a little inconvenient for users, as it assumes the reference database for race **c** is the JFS 44(6) CODIS Caucasian Profiler+ & Cofiler population data. It also may be unimportant.

**Casework** ▸ *Open Case* ▸ select any case ▸ *simulation menu* ▸ (SIMULATE button) *Run test case*

### XI.A.2. Facilities for documenting DNA·VIEW settings

XI.A.2.a. Report parameter etc. settings in DNA·VIEW

including default databases and mutation rates, use §IX.A.11, **Housekeeping** ▸ *Maintenance* ▸ *report systemwide switch/parameter settings*.

XI.A.2.b. Warn of changed parameter settings.

Under **Housekeeping** ▸ *Maintenance*, see §IX.A.13 *compare* and §IX.A.12 *record switch/parameter settings*.

### XI.A.3. Protocol for casework validation

Many labs like to check after a software update that the new version will give expected results, or at least to see and understand any differences from the previous version. For that purpose it's convenient to have a handful of test cases. I suggest choosing from the lab's actual casework some cases covering a variety of situations that are part of the lab's normal work, i.e. paternity including inclusion, exclusion, mutation, etc.; kinship; mixture.

XI.A.3.a.i.) Tagging the validation cases

I give each validation case a comment (§V.B.11 *edit case comment*) including a distinctive code such as `$v`. This (arbitrary) code is convenient in conjunction with *Open case*. As per §V.B.28.a.ii, it makes it quick to find all of them and to open one at a time for testing.

XI.A.3.a.ii.) Record the kinship scenario

For kinship validation cases, especially if the kinship scenario is complicated use the "pick list" (§VI.F.7.p *Scenario from/to pick list*) to save it. Include the case number and perhaps `$v` as comment in the scenario. That makes it easier to find the scenario when using the FETCH FROM PICKLIST button (§VI.F.1.a.i).

XI.A.3.a.iii.) Record the right answer

In a comment field – either the case comment or, if a kinship case as a comment in the pick list scenario – make a note of the calculation conditions (what reference databases) and the desired answer (LR= for the various races calculated).

Once the validations cases are nicely set up, checking five or ten cases takes only a few minutes.

### XI.A.4. Database validation

One possible and possibly useful meaning of database validation is supplied by the *Database similarity test* of §VIII.I. More traditional but of more dubious value is checking for Hardy-Weinberg equilibrium etc – see §VIII.G *Exact Tests for Independence*.

## XI.B. *Update*

The *Update* command is used to complete installation (or possibly repair) of a new (or repaired) version of DNA·VIEW. In those circumstances *Update* starts automatically; no need for the user to invoke it. It does though ask permission to run:

`Ok to proceed with software update?` answer `YES`.

Improvements and modifications to DNA·VIEW consist of an update installer CD, or download. After you run the installer (as explained in §XV.A, Installation Appendix) DNA·VIEW will only present a reduced menu of commands until the *Update* process is permitted to run. *Update* makes some data file changes so that your data files will become compatible with the new DNA·VIEW program release.

(The update files have been copied into the \DNAVIEW\UPDATES *directory* by INSTALL as part of the update installation. Once the *Update* has been performed those files are no longer necessary.)

## XI.C. Program Errors

Despite the greatest care in preparation and testing, programs inevitably have errors. We need your help with both of these kinds:

### XI.C.1. Wrong answers

These can come from algorithmic errors in the programs, or from errors in the tables. Our only possible protection against these is vigilance. Spot check the output for plausibility. Be especially watchful when you get a new version of the program.

### XI.C.2. Fatal errors

These are error conditions detected by the program itself, and which prevent it from continuing.

Conceivably you can generate one by an unanticipated data entry error — maybe a fractional case number.

In such a case the program will stop with a message including wording like `VALUE ERROR`, `SYNTAX ERROR`, or `DISK FULL`. It will also print a fax form.

XI.C.2.a. Reporting errors

Please report errors, both to help yourself and because this is one way that DNA·VIEW is in a constant state of improvement.

There are two good ways to report an error –

XI.C.2.a.i.) Send or fax the fax. Please include a contact phone number. At the time of the error the program will wait at a special prompt; sometimes I need to ask you to try some experiments at the point of this prompt to further diagnose the problem. Therefore, if it is convenient for you to leave the computer in its exact state and contact me immediately, please do so.

XI.C.2.a.ii.) Email the DNAERROR.SF file (from the `DNAVIEW\` main folder). This file contains all the information that the fax does *provided* that you have not answered yet affirmatively to the sign-off prompt `Have you sent off the error report from May 6 yet?` The point is that when you answer **y** the error file is erased and the diagnostic information is therefore lost.

XI.C.2.b. Recovering from errors

Here are some procedures that will sometimes get you past such a problem:

XI.C.2.b.i.) For some errors the error report will include a suggested remedy. Try it.

XI.C.2.b.ii.) Turn the computer off and back on again. Sometimes a computer gets into a strange state.

XI.C.2.b.iii.) Try looking at the data in a different way. If you can determine that some part of it is peculiar, try deleting that part.

### XI.C.3. Network lock

Occasionally a networked DNA·VIEW system runs into a Paradox interlock problem. The symptom is either a red **Waiting for table** box on the screen that won't go away, or an error starting DNA·VIEW that refers to Paradox.

The cure is to sign everybody off of DNA·VIEW and PATER, invoke the **Repair** tool (§XI.G) from the desktop icon, and choose the *Fix* option.

## XI.D. DNA·VIEW Files

### XI.D.1. Data files

XI.D.1.a. DNACFG.SF contains vital configuration information.

In case of an individualized network configuration, in addition to the master DNACFG.SF on the (shared) server, there may also be a local copy for an individual workstation. This allows the separate workstations to refer to different printers, and to maintain different values of certain preserved settings such as the last *DNA odds* calculation.

In addition, there may be a copy of this file in the `UPDATES\` folder. That file is only needed temporarily during update to a new DNA·VIEW (or PATER) version.

XI.D.1.b. DNAMAINT.SF contains configuration information mainly about loci and probes.

XI.D.1.c. DNAREADS.SF, DNARDIR.SF DNAMEMB.SF DNAMDIR.SF have DNA profile data and organizational information that links the profiles to worklists and ultimately to people (i.e. accession numbers).

XI.D.1.d. DNAERROR.SF is a diagnostic record for program development/maintenance. See §XI.C.2.a.ii.

XI.D.1.e. The `PDOX` folder contains groups of similarly named files that constitute Paradox "tables." The tables are broadly of two categories:

XI.D.1.e.i.) PDIR, CDIR, CASEDIR, NAMEDIR, DNAAPPRO define DNA·VIEW cases, vital data.

XI.D.1.e.ii.) RDIR and PEMB exist only for efficiency and can always be reconstructed (§XI.G.2) as they essentially duplicate information found in files of §XI.D.1.b.

XI.D.1.f. DNAFREQ.SF is a self-contained file with all of your available allele frequency population databases, including information about which database is to be used as a default for calculations for each race.

### XI.D.2. Security, Backing up data

All of the above files change and grow as you use the system. Therefore they need to be backed up on a regular basis.

Computers disks are very reliable, but not 100%. It is therefore 100% that some day your hard disk will fail. Since it would probably be extremely awkward to lose DNA·VIEW and all the accumulated data, it should be mandatory laboratory policy to back up the files on a regular basis.

Backing up data is also useful as a way to archive old frequency databases. Possibly you update the frequency databases from time to time as more samples accumulate in the system. However, sometimes an old case comes to court and the necessity potentially arises to reproduce the calculations or provide the databases that were in use when the case was originally reported. In such a pinch the backup tapes should provide the solution.

XI.D.2.a. Network

The easiest method is to install DNA·VIEW on a network drive if you have a network. That way the network administrator automatically backs everything up for you.

Note that you don't have to be running the networked version of DNA·VIEW to use this method. A single copy version of DNA·VIEW can be installed on the network and so long as it is never accessed from two machines at once it will never know the difference.

If you have a networked DNA·VIEW system, then in addition to the files on the network drive there are a few files on the local hard drive of each client machine, usually in C:\DNAVIEW. To ensure that this directory is also backed up, it would be a good idea to copy it to the network drive from time to time. If for example the local machine is called **Wolf**, the local drive is C:, and the network drive is F:, you might copy the folder **C:\dnaview** to **F:\dnaview\wolf**.

These files don't have much in the way of valuable current data, so it would be enough to so the above once or occasionally.

XI.D.2.b. Backup tape

If you have to back up yourself, get a backup device – tape or CD writer – and establish a backup routine. With about a dozen sets of backup tapes you can do the following:

- Daily: Backup at the end of the day. Save these tapes all week. The daily backup can be "incremental" — i.e. only copying files that have changed. (The backup software that comes with the backup unit can do this automatically.)
- Friday: "Full" (not incremental) backup. Save the Friday tapes all month.
- Monthly: Save the last Friday tape of each month to the end of the quarter.
- Quarterly: Save the last monthly tape of each quarter.

A common practice is to keep some of the tapes off-site to protect against fire.

### XI.D.3. Program files

XI.D.3.a. The file DNAVIEW.BAT starts up DNA·VIEW. It is invoked by the desktop icon or from the Start Menu.

XI.D.3.b. The files DNATREE*.SF and DNAVIEW.WS are programs and are replaced by each new DNA·VIEW version.

XI.D.3.c. The files in \DNAVIEW\APLII and \DNAVIEW\GSS are also program files that don't change.

XI.D.3.d. The files in \DNAVIEW\UPDATES are only used temporarily, by the *Update* command, when a new version is installed.

## XI.E. PATER or DEMO

From the **Leave menu**, choose *PATER* or *DNA·VIEW Demo ↔ Production* to switch quickly to PATER (if it is installed), or to switch to the **DEMO**nstration system (obsolete) if it exists.

XI.E.1.a. Behavior from the Demonstration system

From the Demonstration system, you can similarly switch back to Production DNA·VIEW.

## XI.F. *tools* (requires supervision)

The *tools* command is a door to some special facilities, mostly accessed by the function keys. The ones not mentioned here are used only under supervision in order to make emergency bug fixes.

When through with *tools*, ***F3*** to return to the COMMAND menu.

### XI.F.1. Reindex

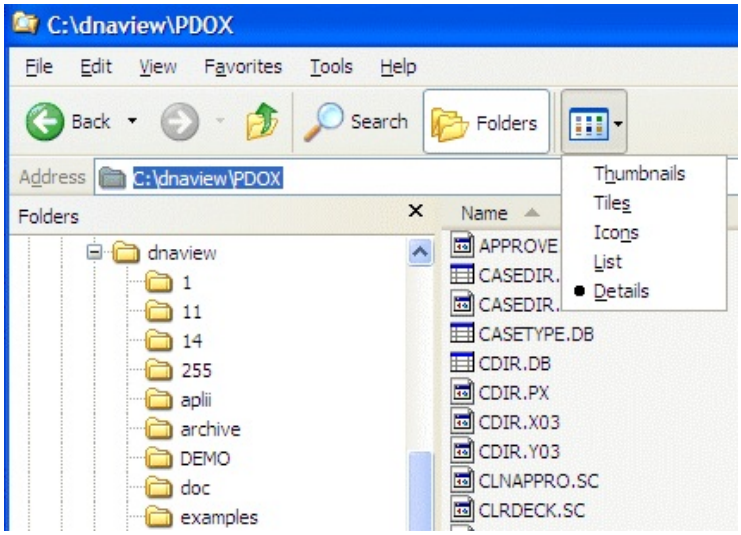
***f6***

This **tool** is also accessible from the command menu: **Housekeeping** ▸ *Maintenance* ▸ *Rebuild Paradox index.*

For maintenance purposes it may be necessary or desirable to erase and then rebuild indexes for some or all of the Paradox tables (§XVI.B).

XI.F.1.a. Normally the erasing part is done automatically by *Reindex*. Occasionally though there appears an error message like

```
CDIR IS indexed
```




in which case you need to erase the index files manually. In that case, read the following paragraph.

The Paradox tables are kept in a `PDOX\` folder within the DNA·VIEW folder – i.e. typically in `DNAVIEW\PDOX`. A Paradox table consists of a handful of related files, such as `CDIR.DB, CDIR.PX, CDIR.X03, CDIR.Y03`. The "`DB`" file has the actual vital data. The other files are "index" files that exist only for efficiency and can be reconstructed from the `DB` file. Therefore, you can safely erase the index files. To do so, sign off DNA·VIEW, use the Windows Explorer ("My Computer") and browse to `DNAVIEW\PDOX`. Click on the "view" icon (see figure) and select Details from the flyout. Click on **Name** to alphabetize the file names. Now it is easy to find and delete just the index files for any given table.

**When to do:** If the error report ERROR HISTORY includes

XI.F.1.b. `Mget`, but TUTILITY (see §XI.G.2.a) failed to find a problem with PEMB.DB, try rebuilding the index for PEMB.

XI.F.1.c. `Cdir`, but TUTILITY failed to find a problem with CDIR.DB, try rebuilding the index for CDIR.

*Reindex* to rebuild one or several Paradox "index" files. *There must be no other DNA·VIEW or PATER users signed on.*

Several kinds of items populate the menu that appears.

XI.F.1.c.i.) `Must reindex!`

If the bounce-bar sits on a line such as `Must index! RDIR`, hit ***Enter*** to permit recreation of a missing index.

XI.F.1.c.ii.) `Reindex`

Additionally, any index can be rebuilt by selecting a line such as `Reindex CDIR`.

Note: Needless rebuilding is harmless, except that it make take some minutes and must be allowed to complete once initiated. (But if e.g. a power failure crashes the computer during rebuilding, there is no disaster – just rebuild later.)

XI.F.1.c.iii.) `Change Paradox library from: pdox\`

This option provides a way to introduce or change the drive letter for the folder containing Paradox tables. Normally thought the best choice is `pdox\`, without a drive letter and without the preceding parts of the path.

XI.F.1.c.iv.) *Change sort order from: ASCII*

Don't change the sort order unless you understand Paradox. Changing the sort order necessitates rebuilding all indices.

### XI.F.2. Push_Me

**Select useful tools** ***f1***

Press ***f1***, which is labeled **`Push_Me`** to obtain the set of user-semi-friendly and interesting tools which are described below:

### XI.F.3. Repair

Sometimes files that sit in one place on the disk for too long start to rot. Occasional use of **FileCompress** (§X.K.6) will probably prevent this happening. However, if it does happen you will get an error with the error message FILE DATA ERROR, indicating that some of the data in a file is corrupt.

The cure in that case is to use **Repair**, which will wipe out the corrupted portions of the file. The corrupt data is irretrievably lost, but usually that's not serious, and removing it at least enables DNA·VIEW to operate without further trouble.

To invoke **Repair**, go to tools and type

**`Repair`** *nn Enter*

where *nn* is a number that designates the file to repair. You will need to contact DNA·VIEW support, with the fax error message, to determine the right value for *nn*.

In cases of extreme damage, DNA·VIEW may abort during **Repair** — you may be dumped out of DNA·VIEW, or the computer may even freeze. However, restart DNA·VIEW and restart **Repair**, and it will delete the last bad spot and continue.

## XI.G. Repair Paradox problems

The **DnaView** desktop folder has an icon called **Repair** that runs any of several operations that can be helpful to resolve corruption problems that occasionally occur (e.g. by unceremonious shutdown of DNA·VIEW or PATER such as by a power failure) with the Paradox tables that DNA·VIEW and PATER use for some kinds of data.

There are certain files that occasionally become corrupt and thereby prevent proper operation of DNA·VIEW, but which can easily be repaired. **Repair** is part of the solution.

### XI.G.1. Reasons for trying Repair include

» the strange error message from DNA·VIEW *Internal error no 226*.

» Data posted from *Paternity Case* to PATER sometimes fails to show up.

» Data that was there yesterday seems to be missing today.

» The program begins an operation and never completes (but usually can be interrupted with ***Ctrl-break***).

» advice from the program author

XI.G.1.a. Don't try **Repair** while there are any DNA·VIEW or PATER users.

### XI.G.2. Repair operation.

Double-click on the **Repair** icon. Options include:

Q. *Quit* – exit from the **Repair** utility

F. *Fix* – Remove temporary Paradox locking files, which Paradox should have deleted but didn't. It can never hurt to do this provided there are no DNA·VIEW or PATER users on the system.

P. *erase Pemb* – Erase a certain expendable DNA·VIEW table, which will then automatically be reconstructed when DNA·VIEW next starts.

R. *erase Rdir* – similar

D. *erase DNAAPPRO* – similar, except that this table is not re-constructible. The data, which is historical case data useful for eventual study of mutation rates, will be lost.

The DNAAPPRO file is a communication file between DNA·VIEW and PATER. If data fails to reach PATER from DNA·VIEW as it should, it may be because of corruption in DNAAPPRO. If so, resetting it with this option is appropriate.

The drawbacks to erasing DNAAPPRO are:

» Any paternity cases that have been computed in DNA·VIEW but not yet retrieved in PATER must be computed again (in **Paternity Case**).

» All old cases would need to be re-computed if it is desired to use the *Mutation History* (§VIII.K) command to analyze them for mutations.

Possibly a corruption problem with DNAAPPRO can be repaired with TUTILITY (§XI.G.2.a).

P. *Paradox* – Offer to open the Paradox user interface, starting in the DNAVIEW Paradox directory, in case of special maintenance work. (Note: Usually necessary to "Fix" first.) This options requires that you have a Paradox user interface. It is not provided as part of the DNA·VIEW package.

T. *Tutility* – verify the integrity of a Paradox table, and offer to try to rebuild it.

XI.G.2.a. *Tutility* options

F. *Fix –* same as above. Also include in the *Tutility* menu because it is a likely prerequisite to the other operations:

Verify any particular table, by typing in the table letter as indicated:

**R**dir, **P**dir, p**E**mb, **C**dir, c**A**sedir, **N**amedir, **D**naappro, **S**trdata.

The result of "verify" will be one of three possibilities:

XI.G.2.a.i.) Table does not exist. This may mean the table is not essential, or may mean that it has been erased or was never created. Starting DNA·VIEW will create any essential missing tables.

XI.G.2.a.ii.) Table verifies ok. Example:

```
Table Rdir verifies ok, but it is still possible it has a bad
index.

Rebuild it anyway?
```

As the prompt implies, the verify doesn't verify everything so if you are suspicious of the table, rebuild it.

XI.G.2.a.iii.) Table doesn't verify ok. Rebuild recommended.

The disadvantage of rebuilding is that rebuilding doesn't work as well as it should. It probably reconstructs the table ok (depends how bad the damage is), but it is likely not to reconstruct correctly some associated files called "index" files.

» Therefore, note any tables that you rebuild and subsequently, *Reindex* (§XI.F.1) them in DNA·VIEW.

Note that there are two tables that DNA·VIEW can re-construct completely, namely `RDIR` and `PEMB` (see above). Therefore there is no particular point in rebuilding these. You may as well erase them completely and let DNA·VIEW rebuild them.

# XII. EVALUATION OF EVIDENCE

## XII.A. Concepts

We are concerned with a question of evaluation of scientific evidence.

### XII.A.1. Scientific Decision Making; a Primer

The following point of view is about as general as you can get:

XII.A.1.a. There is a hypothesis to confirm or deny.

Example 1:

$H_1$ = this man is the father – as opposed to

$H_0$ = this man is not the father.

In practice though in order to be able to calculate we usually need a more explicit alternative hypothesis, such as $H_2$ = a random (unrelated) Caucasian man is the father

Example 2:

$H_1$ = this stain comes from the suspect

$H_2$ = this stain comes from a brother of the suspect

Example 3:

$H_1$ = the children are full siblings

$H_2$ = the children are half siblings

XII.A.1.b. There is scientific evidence

There is some scientific evidence, E. For our purposes, E always consists of the various DNA types from some blood and/or stain specimens. To say that evidence is scientific really just means that its significance is objectively quantifiable. The quantity is that mentioned below in §XII.A.1.c.

There will always be other evidence also, known as the anecdotal evidence, about which the laboratory presumes to know nothing.

XII.A.1.c. There are probabilities

It is possible to calculate the conditional probabilities

X = **Pr(**E | $H_1$**)** and

Y = **Pr(**E | $H_0$**)**.

**Pr(**E**)** means the probability (fraction of all occasions) — some number between 0 and 1 inclusive — that E occurs. **Pr(**E | H**)** means the "conditional probability of E given H." I.e. it is the fraction of the time that E occurs among those times that H occurs. These

Discussion::

XII.A.1.c.i.) This requirement explains what it means that the scenarios must be explicit. Thus "this man is not the father" is usually insufficiently explicit, because to calculate a probability may require making further assumptions about how likely it is that the father and "this man" are related, or about the race of the father. Nonetheless, we won't belabor this point, but will tacitly assume in the examples that $H_0$ is true if $H_1$ is false.

XII.A.1.c.ii.) Normally $H_0$ is formulated in such a way that Y is not 0. It may happen that X=0, which renders the alternative $H_0$ certain.

XII.A.1.d. Prior Probability

To understand what the scientist should calculate and report, it is necessary to consider the decision problem from the judge's point of view. Imagine that the judge considers the anecdotal evidence first, and intuitively or consciously ascribes some "prior probability" p=**Pr(**$H_1$**)**. I.e., in a case of example 1 above, the judge might think, "I don't believe this woman very much. If I heard this story 1,000,000 times, the man would probably be the father only about 100,000 times," so the judge is thinking p=0.1.

XII.A.1.e. Using X and Y

But now we explain to the judge the evidence E and the values of X and Y. Suppose X=0.25 and Y=0.00001. That means that of the 100,000 hypothetical cases in which the man is the father, E would be observed 100,000·0.25 = 25,000 times, whereas of the 900,000 times that the man is not the father, E occurs only 900,000·0.00001 = 90 times.

XII.A.1.f. Posterior Probability

The judge now reconsiders. "If I heard this case 1,000,000 times, I would see the evidence E a total of 25,090 times. Since we have evidence E, this case must be one of those 25,090. Those cases represent 25,000 cases where the man is the father, and only 90 cases where he is a coincidental victim. So it must be 25,000:90 or 99.64% (=25000/25090) that this man is the father."

XII.A.1.g. Bayes Theorem

It is important that the last paragraph be clearly understood. The reasoning that it embodies, albeit in concrete rather than symbolic form, is commonly known as Bayes' Theorem. Does it seem fishy to you that we can forget about the original 1,000,000 cases and imagine ourselves swimming randomly among the 25,090 "relevant" cases? Perhaps you should be suspicious at first; it certainly shows that you are paying attention, and in fact there is something clever about the point of view. **Pr(**$H_i$|E**)** (i=0, 1) is what the judge needs to evaluate. **Pr(**E|$H_i$**)** is what the scientist can compute, and is quite a different thing. How do you obtain one from the other? You should study the reasoning until it makes sense.

You may have heard that there exist "non-Bayesian" mathematicians. If you continue to feel uncomfortable with the logic, perhaps you are tempted to categorize yourself as a non-Bayesian and give up. I cannot condone this route. To the best of my understanding, the non-Bayesians' quarrel is not with the logic, but with the philosophy of assigning a prior probability. Since as a practical matter the judge only needs some absurdly conservative lower bound for p, personally I don't think there is any sensible ground to take the non-Bayesian tack here. However even a doctrinaire extreme non-Bayesian will understand the Bayesian logic and dispute only its applicability.

XII.A.1.h. Paternity index

Before leaving this section, we need to summarize the judge's reasoning in symbolic form. The simplest form will be obtained by using this tip, which will be a revelation to your statistician colleagues: formulate in terms of "odds" or "odds ratio" like the horse players do, rather than probabilities like a mathematician. I.e. the judge's initial judgement of probability p corresponds to odds of p:1−p. (The : means divided by. So you can say the odds are "p to 1−p" or you can say "p/(1−p) to 1" or you can just say "an odds ratio of p/(1−p)," all of which mean that it happens p times for every 1−p times it doesn't happen.) On hearing the scientific data, the judge's estimate of the odds change from p:(1−p) to pX:(1−p)Y. The judge's odds ratio is multiplied by X/Y.

### XII.A.2. The Likelihood Ratio

Therefore, *the scientific evidence is completely summarized by the ratio X/Y*. A ratio of this form is called a *likelihood ratio*. Precisely defined, a likelihood ratio is defined to be an expression of the form:

$$\mathbf{Pr(}E \mid H_1\mathbf{)} \,/\, \mathbf{Pr(}E \mid H_0\mathbf{)},$$

that is, the ratio of two probabilities representing the chance of the same event under mutually exclusive hypotheses.

So each of X and Y by itself is unimportant. Only the ratio matters. And the scientist need not be concerned with, nor make any assumptions about, the prior odds or the anecdotal evidence.

In a paternity case, we call this likelihood ratio PI (Paternity Index); in a forensic case, matching LR.

Formulated in terms of probabilities rather than odds ratios, the *probability of paternity W* is given by the expression:

$$W = pX / (pX + (1-p)Y), \qquad (*)$$

which can also be written $W = \dfrac{\frac{p}{1-p}\frac{X}{Y}}{\frac{p}{1-p}\frac{X}{Y}+1}$

exemplifies the relationship *probability = oddsratio* / (*oddsratio*+1) to convert between probability and odds ratio formulations.

Note that the probabilities in the numerator and denominator of a likelihood ratio need not sum to unity nor to any other particular value. Hence a likelihood ratio is quite different from an "odds ratio," which, at least as used here, is an expression of the form

$$Pr(E \mid H) \,/\, Pr(\sim E \mid H)$$

where the numerator and denominator represent probabilities of *complementary* events based on the *same* hypothesis, and which therefore do sum to unity.

Put otherwise, suppose you have a likelihood ratio of 9. It can of course be written in the form 0.9 / 0.1, numerator and denominator adding to 1, but it doesn't follow that there is any meaning to 0.9 or 0.1 as a probabilities. On the other hand, if the odds ratio for some event (e.g. paternity) is 9, then the probabilities 0.9 and 0.1 represent the probabilities of paternity and non-paternity respectively.

By contrast, multiplying an odds ratio (e.g. prior odds) by a likelihood ratio *does* result in an odds ratio. (e.g. posterior odds).

Likelihood ratio *vs.* Odds ratio

Here is a convenient fact about the odds ratio formulation. Suppose that after we finish explaining E to the judge and provide him or her with the corresponding likelihood ratio, some other scientist comes to the witness stand to discuss the ballistic evidence. The judge has only to multiply to account for each new piece of evidence:

odds ratio = (prior odds ratio)·(DNA likelihood ratio)·(ballistic likelihood ratio)·...

### XII.A.3. Essen-Möller's equation

Making the (unjustified) assumption in equation (*) above that p=1−p gives the classical formulation (Essen-Möller et al, 1939):

$$W = X / (X + Y).$$

### XII.A.4. Likelihood ratios in DNA·VIEW

DNA·VIEW computes a likelihood ratio result in several different circumstances. There are conventionally used names for some of them.

| Likelihood ratios (LR) in DNA·VIEW | | | |
|---|---|---|---|
| **Application** | | **programs** | **alternate name for LR** |
| paternity | | *PI* | Paternity Index (PI) |
| | | *Paternity Case Calculate paternity/maternity* | |
| kinship | | *Kinship* | |
| | | [*Automatic Kinship*] *Case immigration/kinship*, *Prototype Kinship* | |
| stain matching | simple stain | *DNA odds*, *DNA profiles* | matching LR |
| | mixture | *Automatic mixture*, *Mixture Calculator* | |
| | Y-haplotype | *Y-haplotype matching LR* | matching LR |
| racial estimate | | *Racial Estimate* | "relative support" is a likelihood; ratio of two of them a LR |

## XII.B. *PI*

**Use:** ① Find the paternity index when using DNA·VIEW Abridged (otherwise *Paternity case* takes care of it automatically). ② Check the PI in a consult case.

*PI* (in the **Casework** menu), requires that the user type in the (relevant) allele sizes, and perhaps the "matching pattern". It then automatically computes using a default or other selected database. Mutation is not considered.

There are several computation modalities. Under the simplest, the only allele(s) you have to type in are the (possibly) paternal allele(s). The other modalities are essentially useful for RFLP casework and will not be discussed here since the interest in them is low. The documentation is in old DNA·VIEW manuals, which are available on request.

### XII.B.1. Using PI

The PI program is a desk calculator for paternity indices and fragment probabilities. The major steps in using it are:

XII.B.1.a. Establish a fragment frequency database for calculations.

From the menu presented, which includes all available database, choose one. In this way you in effect specify at in one operation the locus, the race, and the particular database.

XII.B.1.b. Use the "control panel" (**Figure 136**) to make calculations for various fragment sizes and parameter values (e.g. window size).

```
Paternity Index Calculator    Esc to quit.   Then Print (F5)
   End current field with:     (A) to select a new database;
                              (B)    specify the matching pattern:   d
(C)  specify the (1 or 2) possible paternal alleles  17  bases Mo ■■
(D) delta, coelectrophoresis discrimination  0.0 % = ±0  bases Ch  ■■
(E)    sigma, standard error of measurement  0.0 % = ±0  bases Af   ■■
Calculations:                                Divisors for X, Y: (4,2)
(G) Floating bin        (H) Calculate per Gjertson
(K) FBI method  (L) count this allele 1  times
  P{random match} = 87/407
1/P{random match} = 4.68
PI = 2.34       X/Y=0.25/0.107
```

| | a | b | c | d | e | f | g | h | i | j | k | l | m |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| mother | ■■ | ■■ | ■■ | ■■ | ■■ | ■■ | ■■ | ■■ | ■ | ■ | ■ | ■ | ■ |
| child | ■■ | ■■ | ■■ | ■■ | ■■ | ■■ | ■ | ■ | ■■ | ■■ | ■■ | ■ | ■ |
| tested man | ■■ | ■■ | ■ | ■■ | ■■ | ■ | ■■ | ■ | ■■ | ■■ | ■ | ■■ | ■ |
| X, Y divisors | (4,2) | (2,2) | (2,2) | (4,2) | (4,2) | (2,2) | (4,2) | (2,2) | (2,1) | (2,1) | (1,1) | (2,1) | (1,1) |

```
*** D3S1358 STR probabilities from 406 observations
   JFS 44(6) Caucasians
```

**Figure 136** Paternity Index program control panel

Notes:

XII.B.1.b.i.) You don't need to ***Enter*** after entering each field in the control panel; just type the letter of the next option or action desired.

XII.B.1.b.ii.) Invoke action *A* to return to Step §XII.B.1.a.

XII.B.1.b.iii.) Option *C* allows 1 or 2 sizes. You might then be forced to select a consistent pattern at Option *B*.

XII.B.1.b.iv.) Parameter changes are temporary. Use *window size* (from *locus maintenance*, §IX.B.2) to change them permanently.

### XII.B.2. PI program examples

PI for paternal allele **17**, using default parameters and database:

At prompt *C*, type **`17g`**. (17 is the allele, **`g`** requests the computation.)

PI for same problem, but temporarily overriding the parameter to "count observed allele in database" (per §V.C.4.a):

At prompt *C*, type **`l0g`** (letter ***l***, number 0)

PI for the ambiguous paternal allele when Mother=Child=**17,18**; man is **18, 19**:

At prompt *C*, type **`17 18bag`**. (17 18 are the alleles, **`b`** refers to the matching pattern prompt *B*, **`a`** selects the pattern, **`g`** requests the computation.)

PI for the same problem, different database:

At prompt *C*, type **`a`** for the database menu, **`D3SABIBlack`** ***enter*** to select the D3S1358, ABI, Black database, **`g`** to compute.

### XII.B.3. PI control panel discussion

XII.B.3.a. Calculation options

Three methods are offered. to calculate versions of the allele probability.

XII.B.3.a.i.) *G* Window/floating bin method

Method *G* is essentially the floating bin approach. A bin size of 0 gives a discrete allele system which is usual for PCR data, although occasionally you may use e.g. a 1% bin to get the effect of lumping TH01 9.3 and 10 together. It takes into account the specified band pattern among the three tested individuals to compute a PI.

XII.B.3.a.ii.) *H* Gjertson method – RFLP only

XII.B.3.a.iii.) *K* FBI method – RFLP only ("fixed bin")

XII.B.3.b. Using the control panel

A special method of operation applies to the control panel — instead of ending an entry with ***Space*** or ***Enter*** as is typical, end by typing the letter (*A*–*H*) of the next desired action/choice. The meanings of the various letters are:

*(A)* action: go back to select another **database / locus / race**. (The currently active database is listed at the bottom of the screen.)

*(B)* choice: select a **matching pattern** from those displayed in the middle bottom of the screen. Each pattern represents a possible band pattern for a trio. For example, pattern **`f`** (see **Figure 136**) should be selected when a heterozygous child matches one allele with its heterozygous mother and the other with its homozygous father.

If there are two lengths at *(C)*, then the possible choices here will be **a**, **b**, **c**, corresponding to patterns with mother and child matching two alleles. If there is only one length at *(C)*, then the possible choices will be **d**–**m**, which are explained by schematics corresponding to the various possible matching patterns for the trio.

The pair of numbers listed as "divisors" are shown only to explain the role that the matching diagram plays in the PI calculation. The first divisor is 4, 2, or 1 according as X is ¼, ½, or 1. The second divisor is divided into the allele probability to obtain Y.

The answer to *(B)* is just one letter. Where you go next is either where you had asked to go before detouring to *(B)*, or, otherwise, *(C)*.

*(C)* parameter: the **size(s)** (allele number of base pairs) of the one or two possible paternal alleles. If you give an answer to *(C)* that isn't consistent with the current choice at *(B)*, then you'll be required to make a consistent specification for *(B)* before proceeding.

How does the program know if you mean allele number or base pair size? Any number less than 100 will be interpreted as a repeat number; greater, a length in base pairs. To specify an allele≥100, prefix it with **#**, such as **#123** means allele #123.

*(D)* parameter: **delta** (δ), the co-electrophoresis resolution threshold (half the size of the "matching window"), as a % of molecular weight. For discrete allele systems such as STR's, it is usually appropriate to use δ=0.

*(L)* parameter: **t**, the number of extra times to include this allele in the database. The reference value is set using the option *Count observed allele in database how many times?* (V.C.4.a) in *Case*, *options* (also accessible as *options from "Case"* under *Options*).

The remark about Temporary values above applies to this parameter also.

*(G)* action: **Calculate PI** using all of the above information.

Just below the control panel, the allele probability, and 1/allele probability are displayed in red letters. Below that the PI is shown in red, and any previous PI's in are scrolled down so that the results of the last few calculations persist on the screen.

The PI is labeled briefly as to the method of calculation (`floating bin` for computation *G*). A complete synopsis of the result and calculation parameters is appended to the log, which may later be printed using *Print*.

When you request computation *G* and the currently selected band pattern (field *B*) isn't consistent with the specified number of paternal alleles, you are obliged to select a new pattern before the calculation proceeds. See *(B)* above.

**Esc** action: *exit* from *PI*.

## XII.C. Paternity Index Discussion

The paternity index, PI, is a summary of the evidentiary value of the data. It is not an ad hoc statistic (as most statistical measures are) whose interpretation depends on tables and relative comparisons. Rather, formulae for PI are derived by an inevitable chain of mathematical reasoning starting from assumptions about the experimental process — i.e. starting from a (mathematical) model of the experimental process. As such, PI has an explicit and real meaning, which may be expressed by such sentences as:

(a) All other things being equal, this man is PI times as likely to be the father as not;

(b) Assuming a prior probability p of paternity, in light of the genetic evidence the probability of paternity is

$$W = p \cdot PI / (p \cdot PI + 1 - p);$$

(c) In light of the genetic evidence the prior odds should be multiplied by PI.

(c) and (b) are the same statement in terms of odds or in terms of probabilities. (a) is an informal statement of (c) for the specific case of prior odds of 1:1 ("all other things being equal"). In any case, think of PI as being an inference from the data.

## XII.D. Explaining the result

Classically people considered three statistics concerning paternity:

(1) A=exclusion probability

or its close cousin RMNE=1−A. The more quickly this historical but misguided statistic is forgotten, the better. That is particularly true in extended family analysis, where it often is the case that no combination of alleles at all can lead to exclusion. Nonetheless, the case may be eminently decidable.

(2) LR=L=likelihood ratio

This is the fundamental statistic, although in the case of paternity it is usually called the PI or "paternity index."

(3) W=probability

In the paternity case, W=probability of paternity. The present context may require different words of course.

Of the latter two statistics above, W is the apparently easier to explain and understand, especially if one is not inclined to concern oneself about the sticky question of "prior probabilities." Assuming for example that the issue is between paternity and unrelatedness, W answers the question

"What is the probability of paternity, in light of (=assuming) the genetic evidence (and also assuming a prior probability of 50%, or of $p$)."

The likelihood ratio has the merit of summarizing the scientific evidence without being polluted by non-scientific assumptions (prior probabilities), but it has the disadvantage of being unfamiliar and difficult to explain in English. As a starting point, it helps to realize that

the likelihoods (in the likelihood ratio) are probabilities *of the genetic evidence* assuming paternity or assuming non-paternity.

Thus, what is an assumption for W is a conclusion for the LR, and vice-versa. A likelihood ratio of 32 does *not* mean paternity is 32 times more likely than non-paternity. It means the evidence is 32 times more likely assuming paternity than assuming non-paternity.

To see the difference, consider this imaginary situation. A man is accused of paternity in Wisconsin and the LR turns out to be 32. Probably he is the father, I admit. But suppose that in the future we genetically screen the whole earth population and find a five-year-old child in Tibet who has the same DNA results in the tested loci. The LR is therefore 32 for him (or her) as well. But he or she is probably not the father. So the LR alone cannot properly be interpreted as implying paternity.

The question remains, how to explain a LR to a lay person. I suggest using the wording "characteristic of." Example phrasings for two situations are given below.

XII.D.1.a. Paternity question; moderate evidence

For the example case of the preceding few sections comparing paternity versus nonpaternity and with likelihood ratio computed in §IX.E.11, the result can be stated as follows:

LR explanation:

"The observed genetic results are 32 times more characteristic of paternity than of non-paternity (i.e. unrelatedness). That is, they are moderately strong evidence favoring the former vis-a-vis the latter hypothesis.".

W explanation:

"If, prior to genetic testing, the two hypotheses were assumed to be equally likely (50% prior probability of paternity), then taking the tests into account increases the probability of paternity to 97%. If some other prior probability assumption were made, then the result would be somewhat different."

XII.D.1.b. Missing body; powerful evidence

Scenario: A body in the woods is tested and compared with a family that has reported a missing child. The combined likelihood ratio is 1,234,000.

LR explanation:

"The observed genetic results are about a million times better explained by assuming that the body is indeed the missing child, than by imagining they are unrelated. If these are the possibilities, the evidence that the body is the child is overwhelming."

W explanation:

"If, prior to genetic testing, the two possibilities were assumed to be equally likely (50% prior probability), then taking the tests into account increases the probability to 99.9999% — virtual certainty — that the body is the missing child. If some other prior probability assumption were made, then the numerical result would be slightly different. However it is hard to imagine a scenario that would change the result materially."

## XII.E. Comparing three or more hypotheses

### XII.E.1. A 3-hypothesis case

XII.E.1.a. Likelihood ratios and likelihoods

Most of us learned that DNA testing compares two hypotheses, and the evidence by which the observed DNA types favor one hypothesis over the other is the *likelihood ratio*. That means there is a likelihood in the numerator and a likelihood in the denominator, and these two likelihoods correspond to the two hypotheses.

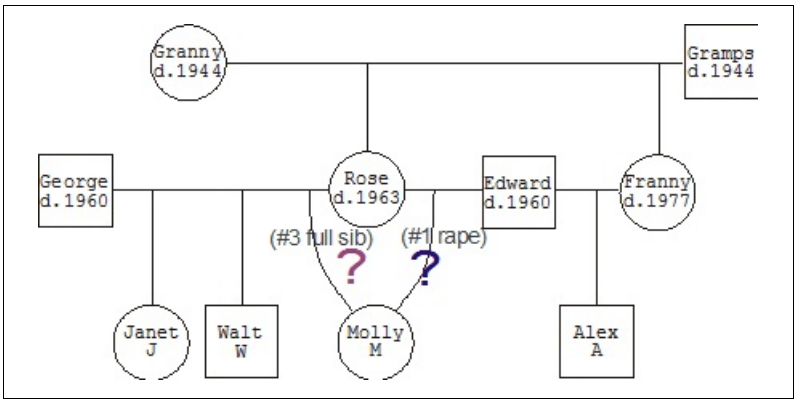



**Figure 137** The case of Molly

Suppose there are three hypotheses? Then there are three likelihoods. There is no likelihood ratio in the usual sense because division is limited to two things, but we can line up the three likelihoods and compare them, just as with two.

*Kinship* is willing and able to compare any number of scenarios at a time (§VI.D). However for the moment we stick to just two at a time.

XII.E.1.b. logic

To illustrate the logic, consider the case in **Figure 137**. It is desired to decide the parentage of Molly based on genetic types that are available for the four children in the youngest generation. These relationships are undisputed:

```
Rose, Franny          : Granny + Gramps
Janet, Walt           : Rose + George
Alex                  : Franny + Edward
```

and there will also be genetic information for some of the people.

In addition, we would like to consider these possibilities for Molly:

| | |
|---|---|
| Hypothesis $H_1$ (rape) | `Molly : Rose + Edward` |
| Hypothesis $H_2$ (stranger) | `Molly : ? + ?` |
| Hypothesis $H_3$ (full sib) | `Molly : Rose + George` |

XII.E.1.c. Three likelihood ratios

Given 3 hypotheses as above there are potentially 3 likelihood ratios to consider: the likelihood ratio for $H_1$ *vs* $H_2$, for $H_1$ *vs* $H_3$, and for $H_2$ *vs* $H_3$. However a moment's thought shows that any one of these can be obtained as either a ratio or product of the other two, so computing every hypothesis against every other is unnecessary.

Suppose for example that we compare $H_1$ vs $H_2$ using *Automatic Kinship*, writing the scenario as

```
M : Rose / ? + Edward / ?
```

in addition to the three undisputed lines above. After calculation, you will end up with a likelihood ratio, such as 50, which would mean that $H_1$ is 50 times better than $H_2$ as an explanation for the genetic results.

Then, compare $H_1$ vs. $H_3$ by using instead the line

```
M : Rose + Edward / George
```

plus the three undisputed lines. Suppose you get a likelihood ratio of 5 in this case, meaning that $H_1$ is only 5 times better an explanation than is$H_3$.

Now if you wish, you can also make a computation comparing $H_3$ and $H_2$ by writing instead the line

```
M : Rose / ? + George / ?
```

but it's hardly worth the trouble, because from the preceding comparisons we already know that $H_1$ compares to $H_2$ as 50:1, and $H_1$ compares to H3 as 50:10. Therefore likelihood ratio comparing $H_3$ to $H_2$ will inevitably be 10:1 = 10.

| | **rape** | **stranger** | **full sib** |
|---|---|---|---|
| **rape** | 1 | 50 | 5 |
| **stranger** | 1/50 | 1 | 1/10 |
| **full sib** | 1/5 | 10 | 1 |

XII.E.1.d. Likelihoods (without "ratio")

In summary, only two likelihood ratios need be computed in order to fill in the table of all pair-wise ratios.

The natural way to understand the situation is to think of the likelihood – without "ratio" – for[20] each individual hypothesis. Likelihoods are by definition relative (that's partly how the word differs in usage from "probability"), so the collection of three likelihoods make sense only relative to one another. Any of the hypotheses can arbitrarily be selected to be the reference hypothesis and its likelihood considered to be 1; the (relative) likelihood of any other hypothesis is its likelihood ratio compared to the reference hypothesis.[21]

In this case the likelihoods can be chosen as shown at right, or in any other way that is proportional to these numbers. The likelihood ratios comparing two hypotheses are obviously the ratios of the corresponding likelihoods.

| | **rape** | **stranger** | **full sib** |
|---|---|---|---|
| **likelihood** | 50 | 1 | 10 |

**Figure 138** Relative likelihoods of hypotheses

XII.E.1.e. Comparing many hypotheses

So only two computations are necessary, not three. Suppose there were five hypotheses, hence 10 pair-wise comparisons. But there are only five likelihoods, which can be determined from only four likelihood ratio computations.

XII.E.1.f. Posterior probabilities

If there is no information as to the prior plausibility of the various hypotheses – or, equivalently, if evaluating that information is not in the province of the scientist – then the likelihood table says all that can be said.

If one is willing to posit prior probabilities of the various hypotheses though, then that information and the likelihoods combined result in posterior probabilities just as when there are only two hypotheses.

In the present case, prior to DNA testing the three hypotheses might be regarded as plausible in the ratio 2 : 1 : 7 – "stranger" seems the least likely, "rape" while a more interesting family story than the innocuous "full-sibling" explanation is really not supported by evidence. These numbers could be scaled to add to 1 so that they become probabilities, but that is not necessary. **Figure 139** shows the method of

---

[20] I say likelihood *for* the hypothesis to distinguish clearly from the language "probability of", since what we are always considering the likelihood (=relative probability) *of* the event, the DNA data, *assuming* one hypothesis or another.

That said, the to a statistician "likelihood" is a technical term which Fisher introduced, and following Fisher they say, confusingly I think, "likelihood of".

[21] Exception: If the data is impossible under some hypothesis, then the likelihood for that hypothesis remains 0 no matter how you scale it, so it cannot be chosen as the reference hypothesis.

computation. The last line, posterior probability is obtained by dividing each "relative posterior probability" by their sum. The arithmetic is laid out in **Figure 139**, and also see http://dna-view.com/manyhyp.htm).

| Make a column for each hypothesis | | *primary* | *1st alternative* | *2nd alternative* |
|---|---|---|---|---|
| | | **rape** | **stranger** | **full sib** |
| Compute the relative likelihoods *L* | *L* | 50 | 1 | 10 |
| Estimate/assume prior probabilities *p*. Relative probabilities is good enough; they need not add to 1. | *p* | 2 | 1 | 7 |
| Product of the previous two rows is *w*, the *relative posterior probability* | $w=L\times p$ | 100 | 1 | 70 |
| The *posterior probability, W,* is obtained by normalizing the relative posteriors to sum to 1 | $W=w/\sum w$ | 58% | 1% | 41% |

**Figure 139** Posterior probability

## XII.F. Special situations

### XII.F.1. Motherless cases

Motherless cases are computed automatically by the command *Paternity case*, *calculate paternity/maternity*.

### XII.F.2. Homozygotes versus 1-banded patterns

A 1-banded pattern does not necessarily mean a homozygous person. Perhaps a second band ran off the gel or was too faint to be noticed.

The *Paternity case* program automatically makes appropriate computations in a variety of circumstances. The rules and formulas are discussed in §XIII.B.5.

Database compilation however, is (at present) *not* affected. A single banded pattern is always counted as two occurrences of the allele.

# XIII. ALGORITHMS

This chapter discusses in detail the way that DNA·VIEW performs some of its computations.

## XIII.A. Probability computations

### XIII.A.1. Match window probability

The purpose of this section is to explicitly document the details of the window computation.

Assume that a allele is given and a particular data base has been selected. There are a number of parameters and stages to the computation.

XIII.A.1.a. Band size in bases

Internally all alleles, even STR alleles, are expressed as some number of base pairs, possibly fractional. (However, whether the fraction is displayed depends on the setting of *Options* §IX.F.28.d).

The STR size is related to the allele number using the PCR parameters (§XIII.B.1) that have been defined by the user for that locus.

Let $s$ denote the size of the allele in question.

XIII.A.1.b. Determine δ (delta)

The value of δ is the "Match window" number that the user sets as discussed in §VII.B.2 such as 0 or 1%.

XIII.A.1.c. The match window

The basic meaning of δ is that two measurements more than δ apart are assumed to represent different sources.

δ determines the amount to be added and subtracted from $s$ to determine the match window. To be precise, the match window is the interval from

$s - s\delta$ to $s + s\delta$ inclusive.

For example, suppose δ=1% for TH01, and consider the match window around the TH01 allele 9.3. Suppose that the "size of allele #0" (§IX.B.3.a) for TH01 is defined as 134bp. Then

$s$ = 134+(9x4)+3 = 173bp.

So $s\delta$ = 1.7 bp.

Consequently the interval $[s - s\delta, s + s\delta]$ includes 9.2, 9.3, and 10, so

» alleles reported as 9.3 and as 10 would be regarded as matching

» database entries for these sizes would be added for purposes of counting the 9.3 probability.

XIII.A.1.d. The floating bin probability

Suppose the number of database alleles that are within the window as defined above is $k$, and that there are $N$ alleles in the database altogether. Then define

$$f_0(s) = k / N.$$

XIII.A.1.e. Adjustment

It remains to apply the effect of the options governing probability calculation. Let

***m***= *Minimum # of matching alleles to assume for frequency calculations* (V.C.4.b),

***t***= *Count observed allele in database how many times* (V.C.4.a),

XIII.A.1.e.i.) Adjust allele count

Using $m$=minimum #, $t$=count observed alleles times, $N$=alleles in database, we calculate the effective number of matching alleles $k$ as

$$k = \max(m, t+f(s)N).$$

Note that for discrete-allele systems such as STR that $f(s)N$ simply means the allele count that appears in the database.

One further exceptional refinement: If *Calculate crime allele frequencies simultaneously?* is **y**, the command **DNA odds** (only) "tosses in" *two* alleles when the type is homozygotes (regardless of whether the profile is typed in with one copy or two). Hence in this case $k = \max(m, 2t+f(s)N)$.c

To represent counting the allele as part of the database, and adjustment is also needed to the size of the database. Therefore we calculate

$$N' = \max(m, at+N)$$

where ordinarily $a$=1, but if *both*

`Calculate crime allele frequencies simultaneously?` (V.C.4.e) is **y** *and*

the command is a crime calculation (*Automatic mixture*, *Mixture ("LR") calculator*, *DNA odds*, *Y-haplotype matching LR* or *DNA exclusion*),

then $a$ means the number of alleles that are reported at the locus, meaning the number of alleles that are typed in. (However, again in the case of *DNA odds* only, always $a$=2).

XIII.A.1.e.ii.) Probability

The reported probability is

$$f = k / N'.$$

XIII.A.1.e.iii.) Examples illustrating "adjustments"

Suppose that 1=***m***= *Minimum # of matching alleles to assume for frequency calculations*

1=***t***= *Count observed allele in database how many times*,

and that alleles 14 15 16 occur in the database with counts 1/200, 5/200, 10/200.

If the alleles are 14 15, then for purposes of *Paternity Case* (including *immigration/kinship*) or *PI*, or even for crime cases if *Calculate crime allele frequencies simultaneously?* is **n** (which is recommended), the probabilities are 2/201 and 6/201.

If the *DNA odds* genotype is 14 15 and *Calculate crime allele frequencies simultaneously?* is **y**, the probabilities are 2/202 and 6/202.

If the *DNA odds* genotype is 16 and *Calculate crime allele frequencies simultaneously?* is **y**, the allele probability is 12/202.

## XIII.B. Computation of Paternity Index

### XIII.B.1. Basic approach (window style)

Assuming that match-binning (i.e. windows, floating bins, not the Gjertson method) is used for paternity calculation, the approach and formulas are basically as described in the American Association of Blood Banks "Parentage Testing Accreditations Requirements Manual."

By definition the paternity index $L$ is $L=X/Y$, where DNA·VIEW takes

X = Pr(child as observed | woman & tested man are the parents), and

Y = Pr(child as observed | woman & random man are the parents).

Note that these definitions mean that X will be ¼ in the most typical situation where everyone is heterozygous, and will also normally take on the value ½ or 1 depending on the pattern of apparent matching among the trio.

The Y value is $q/2$ in the common case where there is a clear-cut paternal allele and it has probability $q$. The factor ½ corresponds to the fact that the woman is only 50% to transmit the maternal allele.

To those who object that she must be not 50% but 100% to pass the maternal allele because she is certainly the mother, the answer is that this objection comes from ignoring the definition of Y and trying to answer some other question instead. Another way to answer the objection is to point out that the same factor of ½ will occur in X. The two ½ factors ultimately cancel one another when L is computed.

### XIII.B.2. Matching patterns

The mother-child-father types follow one of the 13 or patterns shown in the PI panel, **Figure 136** in §XII.B.1.b. At the bottom of each pattern are a pair of numbers labeled "X, Y divisors,"such as 4,2 for the pattern Mother=PR, Child=QR, Man=QS. The "4" means that X=¼. The "2" means the Y=$q/2$ where $q$ is the probability (in the sense of §XIII.A.1) of the paternal allele Q.

### XIII.B.3. Paternal alleles

When it is not clear which allele is the paternal one, $q$ is a weighted average of the probabilities of the possible paternal alleles.

Thus when Mother=Child=QR, either allele might be paternal and either allele is equally likely to be paternal. The appropriate weighting is ½, ½; i.e. $q$= ½Pr(Q) + ½Pr(R).

### XIII.B.4. Motherless case

Formulas to compute the Paternity Index in a motherless case are described in Brenner (1993), and may also be obtained from the **Kinship** (Chapter VI) program. For example, in the typical situation

| | | | |
|---|---|---|---|
| Child | P | Q | |
| Tested Man | | Q | R |

where P, Q, R are fragment sizes with respective probabilities $p$, $q$, $r$,

$$PI = 1/(4q).$$

Obviously X and Y can be chosen infinitely many ways so that PI=X/Y. DNA·VIEW reports X=(½)$p/(p+q)$ and Y=$2qp/(p+q)$. The common normalizing factor $1/(p+q)$ represents the idea that mythical "Mother" Nature contributes a P or Q for sure, but in the same ratio $p$:$q$ as does Nature. On average X=1/4 about.

It may seem obvious to cancel $p/(p+q)$ from both X and Y but this idea has no analogue when the man is excluded. On the other hand, $X=1/(2p)$ and $Y=2pq$ is perfectly defensible but is inconvenient because it gives such small numbers for X and for Y compared to a trio case.

### XIII.B.5. Homozygosity vs. single-allele

If the (pheno-)type determined at some locus is **12**, does it mean **12,12** or **12,null**?

There several situations in which the assessment of paternity may vary depending on whether a one-allele phenotype is assumed to represent a homozygous person, or whether the possibility is also considered that it represents a person with two different alleles one of which is does not produce an experimental result – normally because a primer binding site mutation (inherited, not a new mutation) causes failure of primer hybridization.

In order to account for such a possibility, DNA·VIEW allows including a number of *null alleles* as part of any database. I use the symbol ***o*** for this parameter. "Overlooked," "unobserved," "null," "silent," and "invisible" all mean the same thing in the present discussion.[22] The parameter ***o*** can also be specified on-the-fly during *Paternity Case* computation (§V.B.22).

The situations affected are

- » PI computations by the *Paternity case* program when there is a single-allele phenotype,
- » the *DNA exclusion* calculation for mixted stains.

Of the situations affected by the value (and non zero status) of ***o***, the most important is the so-called "indirect exclusion" — child and man are each single-allele, but different — but there are several other situations as well.

XIII.B.5.a. Null alleles and databases

XIII.B.5.a.i.) Database compilation

Null alleles are *not* considered when DNA·VIEW compiles a database; every 1-allele type is treated as 2 alleles.

XIII.B.5.a.ii.) Putting nulls in the database

You need to edit a database manually (§VIII.C.12) in order to put nulls into it. You may or may not choose to correspondingly remove some of the other alleles, depending on whether there are specific 1-allele types that you believe include a null among the sample population.

XIII.B.5.b. Null alleles and *Paternity Case calculate paternity/maternity* calculation

Whenever any of the three people of a trio – mother, child, or alleged father – has a single-allele phenotype, the calculation of paternity index is affected. Speaking generally the modification is that the program considers two possibilities. If for example the person is **12**, then the program considers both the **12,12** and the **12,null** possible genotypes, and in accordance with Bayes' theorem the relative probabilities of each of these in the adults are in the proportion $\boldsymbol{q}^2$ to $2\boldsymbol{qo}$ where $\boldsymbol{q}$=Pr(**12**).

---

[22] In RFLP practice an allele or "band" may be overlooked because it ran off the end of the gel. As long as experimental conditions are held constant – so that the allele will *always* be overlooked – such an allele is logically equivalent to any other null allele.

In particular, the effects of considering possible null alleles on the computation are

| situation | phenotype pattern | | | discussion | effect on PI of increasing ***o*** (null frequency) |
|---|---|---|---|---|---|
| | Mother | Child | Alleged father | | |
| M-C share null | **P** | **Q** | any | **Q** obviously paternal | none |
| AF-C may share null | **PQ** or **Q** or none | **Q** | **R** | null allele the main chance (also mutation) for paternity | increase greatly |
| possible AF null | **PR** or **P** or none | **PQ** | **Q** | man may pass null rather than **Q** | decrease slightly |
| possible Child null | **PQ** or **Q** or none | **Q** | **QR** or **Q** | paternal allele may be null | decrease slightly |

### XIII.B.6. Mutation calculation

The *Paternity Case*, *calculate paternity/maternity* (§V.B.7) options does a meticulous job of taking mutation into account, especially if with STR data if the STR mutation model (§XIII.C.3) is employed. As of this writing the Symbolic Kinship program (*immigration/kinship*, §VI.E) is not so meticulous (see §VI.F.7.l).

*Calculate paternity/maternity* always considers the possibility of a mutation in a trio (or duo) paternity calculation. See next section. However, if so many or so much mutation is required for paternity that paternity is far-fetched (as defined by user-set parameters, §XIII.C.1.c below), then paternity is excluded and mutation is not considered.

The effect is that if paternity is far-fetched, then the paternity index is reported as 0 for all the inconsistent loci (the loci that some people call "exclusions"). If mutation is deemed practically possible, then the PI is reported as some small number for each of those loci. The logic of this procedure is that in an ordinary non-paternity case, wherein the man is inconsistent with the child in perhaps five loci, no one wants to see a report that shows a small but non-zero PI for each of those loci and an overall minuscule but non-zero PI.

Note: The program correctly but not obviously considers a mutation possibility even in *consistent* loci. Such a calculation acknowledges correctly the slight difference between the following two situations:

| | |
|---|---|
| Mother 11 12<br>Child 12 13<br>Tested man 13 **14** | Mother 11 12<br>Child 12 13<br>Tested man 13 **18** |

The PI is slightly higher in the first situation because of the possibility of a *covert* mutation – the transmission 14 ➜13. Not all such improbable combinations are included in the calculation though; see §XIII.C.3.i.

## XIII.C. Mutation calculations

DNA·VIEW offers three models for dealing with possible mutations – *Mendelian* (no mutation, §XIII.C.2), *STR* (modified step-wise, §XIII.C.3), *Simple* (old, pre-2005, AABB recommendation, §XIII.C.4).

### XIII.C.1. Principles of mutation calculations

If at some locus in a paternity investigation, Mother=11,12, Child=12,13, Alleged father=14, then under Mendelian genetics the PI=0. A more sophisticated and realistic model however acknowledges that this pattern, though probably rare among true trios, is not impossible. That is, the right assessment is probably some very small PI, 0<PI<<1.

To assess the entirety of the DNA data, the PI's for the various loci must be multiplied together. If the negative evidence (from one or perhaps two loci that might be mutations) is not too overwhelming, then the combined PI, including those loci, is the PI to report.[23]

XIII.C.1.a. When the negative evidence overwhelms the positive

On the other hand, when there are too many inconsistent loci ("exclusion", though commonly used with various meanings and often in the same sentence ("One exclusion doesn't make the case an exclusion.") is especially confusing when applied to a nominally inconsistent pattern at one locus), there seems no alternative but to report "exclusion" – i.e. non-paternity.

XIII.C.1.b. DNA·VIEW policy for exclusion cases

In that case DNA·VIEW reports PI=0 overall and also PI=0 for each inconsistent system. ( Otherwise a typical report for a non-father would have a handful of loci with microscopic PI's, which would seem peculiar.)

XIII.C.1.c. Rejecting "paternity" or other primary hypothesis

In view of the mutation calculation, the cumulative or overall PI will never be 0. Therefore some different rule is necessary in order to decide to "exclude" – i.e. reject the primary hypothesis such as "paternity". DNA·VIEW offers two options.

XIII.C.1.c.i.) *Case Option* (§V.C.3) for minimum PI (other than 0)

`Minimum PI - report PI as 0 if less than  (0-1, reasonably 0.01)`

A value of e.g. 0.01 for this Case option parameter means that if the cumulative effect of mutation calculations drives the overall likelihood ratio below 0.01, then report that overall PI=0 and also consistently show PI=0 for each inconsistent locus. See §XIII.C.2.b.iv.

XIII.C.1.c.ii.) *Case Option* (§V.C.3) for the maximum allowed number of inconsistencies to explain by mutation

`Max # inconsistencies for a parent (rec: 999; prefer "Min PI")` **`999`**

Setting this parameter to a large number means rely on the previous, ("*Lower bound*") parameter.

A more traditional but less logical approach is to set this parameter to 1 (or 2), implementing the popular rule that "there must be two (or three) exclusions to exclude." This option is a concession to the traditional practice in handling the mutation?/exclusion? quandary in paternity testing..

[23] Some labs used to ignore the locus if there was only one inconsistency. That amounts to assigning a PI=1 meaning "no information," which is of course not true or fair.

### XIII.C.2. Mendelian model

XIII.C.2.a. Definition

In the Mendelian model there is no mutation.

XIII.C.2.b. Application

The Mendelian model applies to

XIII.C.2.b.i.) *Kinship* (the stand-alone version, §VI.H);

» example: The *Kinship* scenario **`C pq : M p + F rs /?`** gives LR=0.

XIII.C.2.b.ii.) *Case*, *immigration/kinship*) if *Do NOT consider mutation* (§VI.F.7.l) is selected;

XIII.C.2.b.iii.) any situation if the value of the mutation parameter μ=0 for a locus.

XIII.C.2.b.iv.) Any paternity calculation when preliminary computation results in a decision to reject paternity (see §XIII.C.1.c).

### XIII.C.3. STR mutation model

*Paternity case*, *calculate paternity/maternity*, incorporates a realistic step-wise mutation model.

XIII.C.3.a. Definition

The essence of the model, which applies (optionally) to STR loci, is that 1-step paternal mutations occur at the rate specified by the per-locus "mutation rate" parameter, 2-step etc. or maternal mutations less frequent by specified ratios.

XIII.C.3.b. Application

The STR mutation model applies

XIII.C.3.b.i.) only to STR loci (those with associated pseudo-enzyme #22, regardless of whether it is spelled "STR"),

XIII.C.3.b.ii.) only for calculations by ... *Case*, *calculate paternity/maternity*

XIII.C.3.b.iii.) if the *Case Option* of §XIII.C.3.c.i is set to **`yes`**.

XIII.C.3.b.iv.) The model is not yet implemented for *kinship* calculations.

XIII.C.3.c. Parameters of the model

The model is controlled by the parameters and switch discussed below, in addition to the few that are common to all mutation models, discussed above.

XIII.C.3.c.i.) Invoke the model?

```
Use the model below for mutation in STR systems?(recommend: yes)
```

Set this *Case option* parameter "yes" to apply the STR mutation model to STR loci. Set it to "no" to retain the old behavior (the "RFLP model", §XIII.C.4). In no case will the STR model apply to non-STR loci (such as those coded as SNP or as POLYmarkers. See §IX.B.6.a).

XIII.C.3.d. Rate of paternal 1-step mutations

Assuming the STR model is active, the per-locus *Locus Parameter* (§IX.B.4.a) "mutation rate" specifies the rate of 1-step mutations from father to child.

XIII.C.3.e. Rarity of 2-step (or larger) mutations

`Ratio of 1-step to 2-step mutations?` `(probably 10-50)`

Two-step mutations are rarer by some factor, perhaps 10 times rarer – or whatever value is specified in this *Case option* parameter. Three-step mutations are assumed to be rarer still by the same factor, etc.

XIII.C.3.f. `Ratio of paternal to maternal mutation rate` `(about 4)`

Set this *Case option* parameter to 3.5 or so to model the fact that maternal mutations are less common than paternal.

The model assumes the same relative proportion between paternal and maternal mutations for all STR loci.

XIII.C.3.g. `Fraction of mutations that increase allele size` `(0.5-0.6)`

Setting this *Case option* parameter to 0.5 is good enough. Future implementations may incorporate a more elaborate model to cater to Tim Clayton's claim (for which he cites Xu et al) that the ratio may be significantly unbalanced for large alleles.

XIII.C.3.h. `Lower bound, probability of any mutation` `(usu 0, or e.g. 0.0001)`

Leave this *Case option* parameter as 0 normally. It provides a way to cater to an oddball mutation if one is ever suspected.

XIII.C.3.h.i.) *Use motherless calculation for loci with maternal exclusion? (Recommend: no)* **n**

Using STR systems and the STR mutation model (§V.C.3.a), **no** results in an excellent computation properly taking into account the possibility of a maternal mutation. **Yes** means, if the mother is excluded, ignore her and make the motherless calculation as if she were not typed in this locus.

XIII.C.3.i. Various mutation possibilities

In principle, DNA·VIEW considers all mating patterns and even very unlikely mutations. As an example:

| | | | |
|---|---|---|---|
| Mother | 10 | 12 | |
| Child | | 12 | 13 |
| Man | | 12 | 14. |

There are numerous mutational combinations the permit the man to be the father –

» 14 ➜13 or 12➜13 paternal mutation

» 12➜13 maternal mutation

» various combinations of double mutations.

Mainly in order to permit manual verification of the computation, DNA·VIEW ignores the relatively unlikely combinations, where “relatively unlikely” is defined by the user-specified *Case option* parameter `Include mutation possibilities if effect > x%` (§V.C.3.f). See also §V.B.7.a.i for indication on the parentage report.

XIII.C.3.j. Mutation and Null alleles

Mating pattern including null alleles are also considered, and if appropriate null alleles and mutation will be considered simultaneously. For example:

| | | |
|---|---|---|
| Mother | 10 | 12 |
| Child | | 12 |
| Man | | 13. |

Paternity is consistent with either the man and child sharing an unobserved (null) allele – i.e. a mutation in the primer binding region that prevents hybridization – or a 13➜12 mutation.

### XIII.C.4. The simple or AABB mutation model (for RFLP)

XIII.C.4.a. Definition

DNA·VIEW assigns PI=μ, where μ is the locus-specific mutation rate established as described in §IX.B.4.a.

XIII.C.4.b. Application

This model applies to

XIII.C.4.b.i.) calculations by *... Case*, *calculate paternity/maternity* for non-STR loci

XIII.C.4.b.ii.) calculations by *... Case*, *calculate paternity/maternity* for all loci if the *Case Option* of §XIII.C.3.c.i is set to **no**

XIII.C.4.b.iii.) *Case*, *immigration/kinship*) if *DO consider mutation* (§VI.F.7.l) is selected.

XIII.C.4.c. Theory

The rule that PI=μ is based on this simple model of mutation, assuming for the sake of illustration an obligatory paternal gene of Q:

X = Pr(man without gene Q will contribute Q)

= Pr(contributed gene will mutate) · Pr(mutated gene will be a Q)

= μ · Pr(Q).

Y = Pr(random sperm is Q) = Pr(Q).

X/Y = μ.

That is, the model assumes that the chance a gene will mutate to Q is proportional to the prevalence of Q genes in general, without regard to such possible complications as different probabilities for a small vs. a large or positive vs. negative mutation size change.

XIII.C.4.d. AABB formula for mutations

The AABB recommends a slightly different formula. Their formula is derived by taking the view that the evidence consists of a man who does not have (either of) the (possible) paternal allele(s).

X = Pr(inconsistent pattern | true trio) = μ.

Y = Pr(inconsistent pattern | false trio) = $A$,

where $A$ is the so-called *probability of exclusion*; the probability that a random man would have a pattern inconsistent with paternity at this locus. The extra complication does not change the result very much (since normally $A \approx 1$ for the RFLP mini-satellites that were standard when the formula was suggested), and the relative improvement is anyway dwarfed by the gross inaccuracy in simplify the evidence from actual allele sizes to merely "inconsistent pattern".

The AABB however uses not the case-specific power of exclusion, A, but the mean power of exclusion ($\bar{A}$). I'm not sure why, but in either case the formula is correct on average (which mine is not), and in neither case is any of the three formulas anywhere near accurate.

The AABB recommended behavior can be obtained in DNA·VIEW by setting the parameter Pr(mutation) to $\mu/\bar{A}$ instead of to μ.

XIII.C.4.e. Mutation formulas not accurate

The general rule is that mutations change allele length by a small amount. Thus when the man mismatches the child allele by a small amount there is a lively chance of a mutation but the formula underestimates it, tending to help true fathers get off the hook. Conversely, when the alleles mismatch by a large amount, the same computation unfairly inflates the possibility of a mutation, perhaps unfairly overstating the evidence against a non-father.

Nonetheless, as regards RFLP, I don't have a better formula to suggest. Vigilance...

## XIII.D. Null Alleles in Kinship

*Kinship* (automatic or prototype) includes an option to consider null alleles. When the "null allele" toggle is set to **yes**, then for any one-allele genotype the possibility of a null allele is properly taken into account.

Example: Given a boy B and his putative father F – that is, the scenario is **B : ? + F/?**

Suppose the boy 13, 14 and the man 13. The probability for the man to pass a 13 is less than 100% because the possibility that he is 13,null is included as a possibility.

## XIII.E. Mutation and kinship

See §VI.F.7.l.

## XIII.F. Averaging sizes together

When DNA·VIEW has several measurements of the same allele, it determines a single average measurement by a combination of averaging and discarding outliers.

First, the average is computed of a set of numbers. Then a window is constructed around the average using a user-defined comparison threshold parameter which we call δ. The value of δ that the user has specified is interpreted as a half-window amount; any measurements that are more than δ/2 from the average are discarded as outliers, and the process is then iterated: again an average is taken, again outliers are discarded, etc.

Here is the reason it is more convenient to use δ /2 rather than δ: Often there are exactly two measurements. Then the effect of the algorithm is, that if they are within δ of one another both are acceptable and the average is used. If they are more than δ apart both are considered as outliers and both are discarded.

If there is a discrepancy about the number of bands, homozygous readings are ignored.

### XIII.F.1. Several reads of a lane

For example, when there are several readings of a lane — normal for RFLP data — the above procedure is employed with the "window size" parameter $\delta_r$ playing the role of the comparison threshold parameter δ in the above description.

### XIII.F.2. Several lanes

Sometimes there are multiple assays of the same person.

Normally[24] the program would then average them together as follows:

XIII.F.2.a. Per-lane average

Obtain a per-lane average following the procedure in §XIII.F.1 for each lane.

XIII.F.2.b. Average lane averages

Combine these averages using the parameter called $\delta_{\text{p}}$, which is also a "window size" parameter.

[24] Why only "normally"? There is also the possibility that the user, by using *restrict data* (§V.B.26.a), may select only one of the lanes.

# XIV. USE OF THE KEYBOARD AND MOUSE

This chapter details the rules for keyboard input.

## XIV.A. Answering prompts

There are several modes of interactive user input to the program: menu selection, type-in text (single or multi-line), choice via buttons, and fill-in form.

### XIV.A.1. Buttons

(new – 2010)

All modes make use of buttons (e.g. **Figure 140**) – labeled rectangles on which the user can ***single-left-click*** to create an action or for other purposes. The buttons may also be operated by means of keystrokes (hot-keys), and several consistent rules apply:

XIV.A.1.a. Hot button

If one of the several buttons is highlit yellow, the others being white (OK CANCEL), then

XIV.A.1.a.i.) ***enter*** operates the yellow (hot) button (except ***ctrl-enter*** during multiline input)

XIV.A.1.a.ii.) ***tab*** and ***shift-tab*** cycle the yellow highlight among the buttons

XIV.A.1.b. Standard buttons and Key customs

XIV.A.1.b.i.) ***Esc*** operates the **Cancel** or similar button, and backs up one operation.

XIV.A.1.b.ii.) ***Ctrl-z*** operates undo, restoring the initial contents of a type-in area.

XIV.A.1.b.iii.) ? is for help.

XIV.A.1.b.iv.) The button ____ erases the typed-in keystrokes so you can restart selection.

XIV.A.1.c. Purple buttons, which sometimes occur at the beginning of a row of buttons, are just labels for the row and don't themselves have any action.

XIV.A.1.d. Button help

The list of hot-keys is shown if you ***right-click*** on any button.

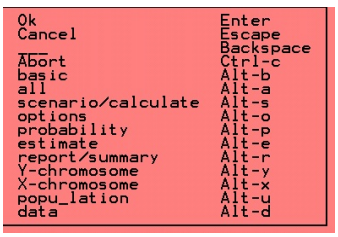



**Figure 140** Example button hot-key cheat sheet

### XIV.A.2. Selection from a list of choices

(enhanced – 2010)

It is often necessary to make a choice selection from a set of items, which are listed in a **green** box with buttons such as **Ok, Cancel, Properties** ....

Menu operation (for DNA·VIEW as for Windows) typically consists of two steps:

XIV.A.2.a. **Choose** one of the menu items by any convenient combination of ***arrow keys***, ***left-mouse-click*** on the item, or *typing search text*.

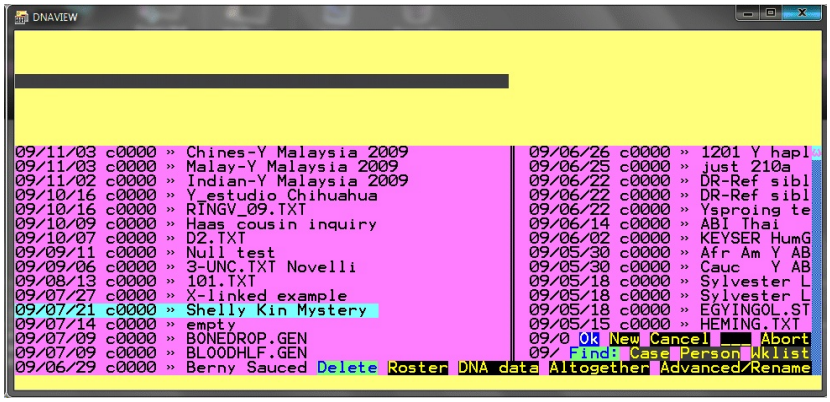
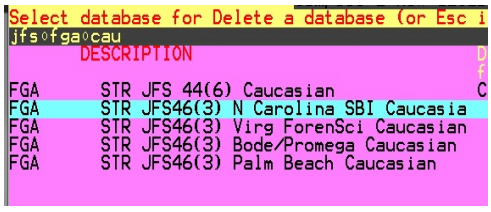



**Figure 141** menu selection and buttons

XIV.A.2.a.i.) ***arrow-keys*** operate as intuitively expected to move the red choice bar among the list of choices. The list is circular; last item precedes the first. A long list will scroll as necessary, and if narrow like **Figure 140** will be arranged in panels which are traversed with ***left-*** and ***right-arrow***.

XIV.A.2.a.ii.) ***mouse-left-click*** on an item also moves the red choice bar to that item. ***Double-left-click*** has the additional effect of confirming the selection at the same time.

XIV.A.2.a.iii.) ***typing search text*** utilizes the simple and novel method of *multiple-context-matching*. *Context* means to accept menu lines including a typed phrase, which improves on the traditional method of initial matching. *Multiple-* uses an intelligent heuristic to parse the user's stream-of-consciousness collection of key syllables or phrases to home in on the desired match containing all contexts. For example, typing `jfsfgacau` to browse the database (§VIII.C) list results in the parsing shown in at right. Searching is *case forgiving* (typing **`fga`** finds `FGA`) but *case aware* (the choice bar preferentially matches case).

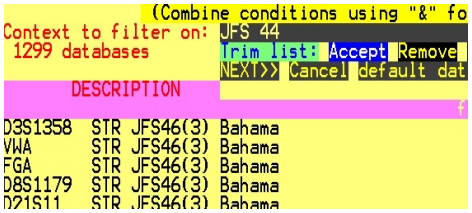



XIV.A.2.b. **Confirm** with one of the buttons (or it's hotkey equivalent such as ***esc***).

XIV.A.2.c. **Menu** button. Some menus that are lists of program options (i.e. commands). If there are a large number of possible options (as for example *immigration/kinship* §VI.E), to organize the potential confusion there is a special row of buttons purple-labeled **menu**. These buttons, with names like **all**, **basic**, **edit**, when ***left-clicked*** result in a corresponding subset of the options being shown.

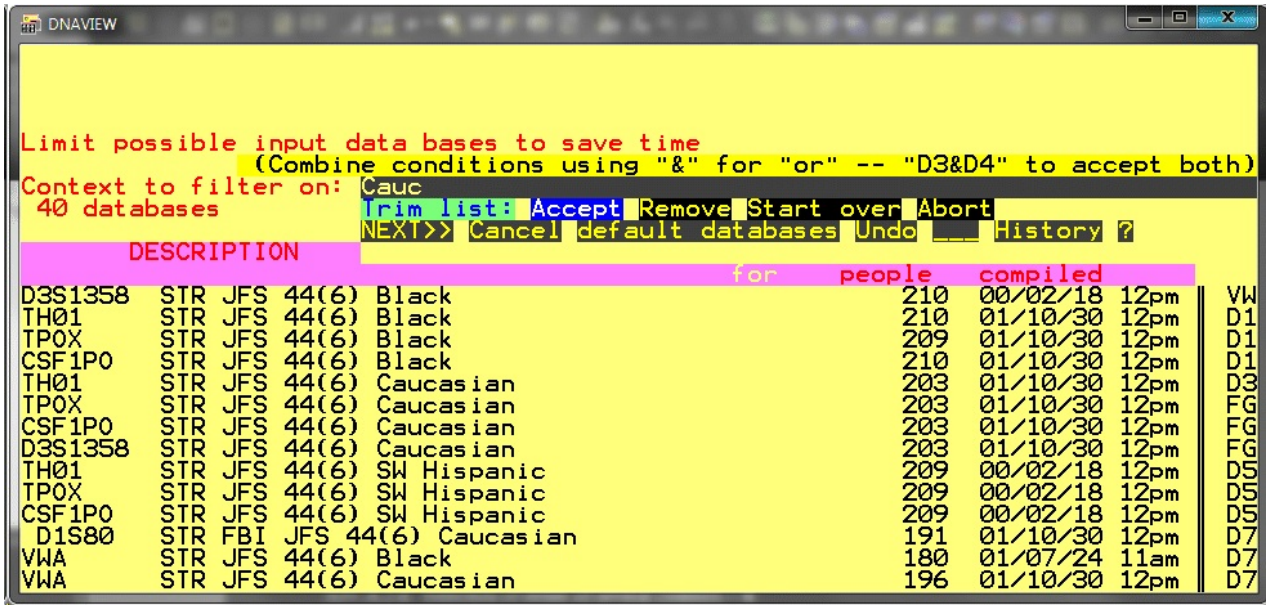
### XIV.A.3. Multiple selections

In some situations it is necessary to select multiple items from a list. For example, you need to select several databases to export them (§X.C.2) or to re-compile them (§VIII.C.7.b).




XIV.A.3.a. Selection by context

The method provided to do this is iterative selection by context. Example: Typing **`JFS 44`** ***enter*** reduces the list from 1299 (above) to 40 databases (below).




XIV.A.3.b. Iterative selection

At each iteration, you can reduce the list further to those lines that also satisfy an additional criterion. Example: Type **Caucas** ***enter*** to reduce the above list further, as at right.

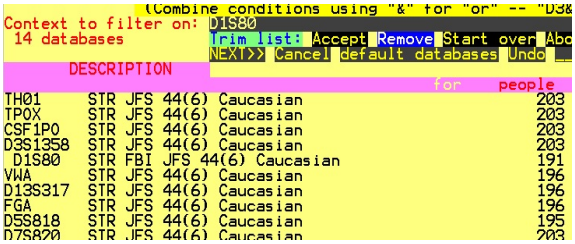



**Figure 145** ... and also including `Cauc`

XIV.A.3.c. Negative selection

The **Remove** button removes lines containing the typed context, such as the selection phrase **D1S80** from the picture at the right.

XIV.A.3.d. Selection of either of several contexts – **&**

In order to permit now selecting those databases for either **TH01** or **FGA**, use the selection phrase **TH01&FGA**. (**&** means "and" under the point of view that the resulting list is the **TH01** list *and* the **FGA** list.)

Of course you must be careful not to include any inadvertent spaces when specifying the context. Conversely, some ingenuity, including textual spaces, may sometimes help make the desired selections. For example, " **A& B& C& H**" might select all names with a default race of **A**sian, **B**lack, **C**aucas, or **H**ispan.

Tilde (**~**) combined with **&** is allowed but is probably not useful; **JFS&~Cauc** specifies all lines that include **JFS** as well as all those without **Cauc** – almost everything.

XIV.A.3.e. Selection of defaults

The **default databases** button reduces the list to default databases if any.

### XIV.A.4. Numeric and text responses

In many situations the user types an answer – text or numbers – into a box. Think of the process as two steps:

XIV.A.4.a. entering the information

Most often there is a suggested or *default* answer already in the box. If it is correct for you, skip to §XIV.A.4.b.

XIV.A.4.a.i.) **type to replace**. Per the normal Windows convention, if the first character you type is a visible character the default answer is erased and replaced by that character.

Aside from that, there are quite a few special buttons/keys to make the entry process efficient.

XIV.A.4.a.ii.) First among them is *help*:

The **?** button (key ***F1***) pops up a chart of useful editing keystrokes and options. This help is available whenever you are supplying a type-in response, whether one-line (e.g. when typing a comment or inventing a file name), or when entering a kinship scenario.

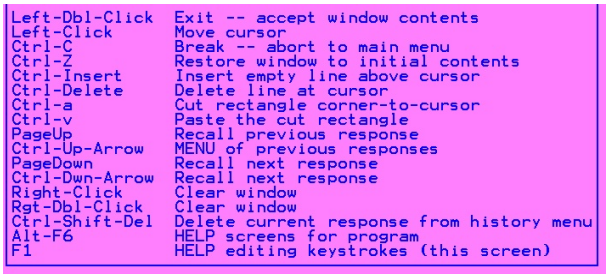



**Figure 146** useful keystrokes during text entry

XIV.A.4.a.iii.) Editing keystrokes

XIV.A.4.a.iv.) (Especially) for text input into a box of more than one line, **introduce or remove lines** with ***Ctrl-Ins*** (useful if editing a *Kinship* scenario) and ***Ctrl-Del*** respectively.

» ***Ctrl-z*** restores the text as it was before you began editing.

XIV.A.4.a.v.) **Navigational arrows** work as expected.

XIV.A.4.a.vi.) Memory

» ***PageUp*** and ***PageDown*** (sometimes also ***Up-arrow*** and ***Down-arrow***) scroll through previous type-in responses. ***Ctrl-Shift-Delete*** deletes an item from the memory list.

» ***Ctrl-Up-Arrow*** pops up a menu of previous responses.

» ***Del*** deletes an item from the list of previous responses.

XIV.A.4.a.vii.) Copy-and-paste

You can copy and paste a rectangular block of text (useful when entering kinship scenarios). A (part of a) single line counts as a "rectangular block". The rectangle is defined by opposite corners.

» ***Single left-click*** to position the “mark” (the small, character-width flashing rectangle which is by default at the beginning of the type-in area).

» Float (don't click yet!) the mouse arrow on the opposite corner of the desired rectangle.

» ***Single right-click*** to capture a rectangular block with “mark” and mouse arrow as opposite corners. (Also repositions the “mark”.)

» ***Single left-click*** again to position “mark” where you desire to paste.

» ***Ctrl-u*** to paste the captured rectangle with upper-left corner at “mark” (see **Figure 57**, page 126).

XIV.A.4.a.viii.) Copy-and-paste from Windows

» Capture some text from a Windows window in the usual way (e.g. ***ctrl-c***)

» Left click in the DNA·VIEW window into which you wish to paste and position the insertion mark.

» ***Right-click*** on the title bar (the strip at the top) of the DNA·VIEW window, put the cursor on the word **Edit**, slide out to **Paste** and ***click***.

XIV.A.4.a.ix.) Insert/replace mode: The ***insert*** toggles as would be expected between insert and replace modes. Also, the cursor height reflects the mode to give you visual feedback: Thin cursor=insert, thick cursor=replace.

XIV.A.4.b. termination character to confirm the input

Completion of entry into those fields requiring a number(s) or text as an answer is usually signaled by striking ***Enter***. If only one number is expected, then ***Space*** is usually allowed also. In some cases, ***Tab*** or a letter (a role letter signals exit from the "year" part of an accession number) may suffice; see the instructions. To conclude entering a kinship scenario (§VI.F.1), ***Esc***.

### XIV.A.5. Entering probabilities

A probability is a number in the range 0 to 1 inclusive. For convenience a few shortcuts are implemented:

XIV.A.5.a. Entering a number >1 is allowed, and it will be interpreted as either a percent, or a reciprocal. Example:

> Use what prior? (0–1; 0 means "none"): **999**
> Do you mean (a)99.9% (b)1/999 (c)neither? **a**

XIV.A.5.b. The NONE button indicates no probability. For example pick NONE for prior probability to avoid any posterior probability computation. Entering probability=0 has the same effect.

### XIV.A.6. Yes/No prompts

Answer **y** or **n**. Synonyms for **yes** include **j**, **s**, and **o**. Do NOT also strike ***Enter***. However, if a default answer such as **y** is already sitting in the answer box, then it is ok to ***Enter*** to accept the suggestion.

### XIV.A.7. Dates

Dates are input as month, day, year (for the US — other orders as conventional in other parts of the world) with today's date usually presented as the default. The month is accepted as a choice (§XIV.A.2, page 287) from a list of the months; day and year are numeric input (§XIV.A.4, page 290). Terminate selection for each field with ***Space*** to continue to the next field; terminate with ***Enter*** or ***Tab*** to accept the remaining default choices. To back up and correct mistakes, the keys ***left arrow*** (previous field) and ***home*** (start over) are supported.

Accession dates are used only for documentation. If they don't interest you, the option §IX.E.4 can save a little time.

### XIV.A.8. Mouse input methods

XIV.A.8.a. Menu selection

Double click on a menu choice to select it, or double click outside of the green choice area to select the currently highlighted item.

Single click on a menu choice to highlight it, or single click outside of the green choice area to clear the highlighting.

You can click on the scroll bar to scroll, or on *find* or *clear* or *help* for those tasks.

XIV.A.8.b. Yes/No questions:

Double left click for **yes**.

Double right click for **no**.

XIV.A.8.c. Questions requiring Numeric or Text response:

Double left click to accept the suggested default.

XIV.A.8.d. Within Help Screens:

Left click to select a help menu choice.

Right click to back up a help screen.

### XIV.A.9. People

People are described by an accession number, a race, and an accession date. Any place that a person entry is called for, a new person may be defined, or the race and accession date can be corrected.

XIV.A.9.a. Editing the Accession Number

The accession number is in two parts, separated by -. The first part is up to four digits, and may be used as a year code (e.g. **91** or **2000**). Type **-** or ***space*** to advance to the second part, which is up to five digits. Leading zeros are not significant. The "year part" is not allowed to be 0. Thus, 2000-0 is allowed but 0-1 is not allowed.

There is no requirement to use the leading part of the accession number to indicate the year — it is just a scheme that many labs do use.

Terminate the accession number entry with ***Enter*** (OK) skip editing the race and accession date, or with ***Space*** if you wish to edit either race or accession date. However, if the person is previously undefined you will not be allowed to skip.

XIV.A.9.a.i.) ***Home*** backs up from the middle to the beginning of entry for a person.

XIV.A.9.a.ii.) Two and four digit year numbers

Until the year 1999, DNA·VIEW only allowed two digits for the year number, eliding the century part. So you would enter 98-12345. The extra two digits are necessary in order to maintain the "year-sequence" scheme into the year 2000 and beyond.

The addition of the extra digits introduces possible ambiguity and confusion between pairs of numbers like 98-12345 and 1998-12345. DNA·VIEW handles this possible ambiguity as follows:

» 98-12345 and 1998-12345 are not the same — they are two separate people.

» If you type one of them — say 1998-12345 — and it does not exist, but the other one — 98-12345 in this case *does* exist, you will be warned, and given an opportunity to change your mind. A pop-up box prompts:

**There already is a 98-12345**

and the choice box offers the choices:

```
Then please use the old number: 98-12345
Create 1998-12345 anyway. We'll have both numbers.
```

with the first of those choices the default, already highlighted. Thus, hit ***enter*** if you made a mistake and want to use the number that already exists. Type **C*enter*** to insist on creating the alternate number.

XIV.A.9.a.iii.) Accession/sample number lookup by case

Very often when you want to enter an accession number the most convenient method is to copy it from a case in which the sample occurs.

At the accession number input dialogue, click COPY FROM A CASE. Or in the particular case of entering roster data in a worklist, put the red bounce bar on the lane to populate and click COPY FROM: CASE.

Either way, the `From case?` dialogue pops up. Enter the case number (the RECENT or SEARCH buttons may be helpful), then at `Copy whom?` choose the desired sample.

XIV.A.9.b. Editing the race code

The race is a single letter, or – (used for children) (§IX.A.4, page 197). If you type **?**, then you can see and edit the list of race names. You may select any race letter desired with OK, and the PROPERTIES button lets you enter or change any race name.

XIV.A.9.c. The accession date

The accession date is entered and edited as described in §XIV.A.7.

The accession date is printed along with the other accession fields on the case report, but has no importance at present in DNA·VIEW. Therefore it is reasonable to accept always the default, which is today's date.

### XIV.A.10. Case numbering

Parentage, kinship, and crime cases are referenced by number.

The case number must be a positive integer up to about 2,147,000,000. Sorry, nothing but digits are allowed – no spaces, letters, dashes, superscripts, dots, colors, italics, or Sumerian cuneiform marks although there is some flexibility in specifying a format for the number (§IX.F.2).

Several features make case number entry more convenient including popup memory of "popular cases" and keystroke shortcuts for automatically prefixing the year. See §IX.F.2 and §V.B.28.a.iii.

## XIV.B. Special Editing Screen

In a few rare situations – when some DNA·VIEW operation requires editing a large block of text – the editing takes place in a special editing screen with its own special rules.

You can tell that you are in the special editing screen if either

(i) There are bracketed numbers – e.g.

```
[0]
[1]
[2]
```

along the left margin of the screen, or

(ii) such numbers appear after you type ***ctrl-o***. (***Ctrl-o*** again to toggle the numbers off.)

The basic rules for editing are these:

XIV.B.1. Navigating

- You may use the cursor arrows or ***Enter*** to move about.
- You may use the ***Ctrl***-cursor arrows, and ***PgUp***, ***PgDn***, ***Home***, ***End***, with or without ***Ctrl***, to move about faster.

XIV.B.2. Input. There is a text full-screen editor, and a numeric one (used for editing probabilities).

XIV.B.2.a. Text input

- Type in the normal manner.
- The ***Ins*** key toggles between insert and replace modes.

XIV.B.2.b. Numeric input

- Navigate to the cell to change. Type in a number, which appears at the bottom of the screen.
  - Type ***Enter*** or any other navigation instruction to enter the number into the cell.

XIV.B.3. Deleting and Inserting

- ***Del*** and ***Backspace*** delete at and behind the cursor.
- ***Alt-F4*** deletes a line. ***Alt-F3*** inserts a line. Use these options with great restraint. There is no harm in reordering the list of readers, but it is important that each probe and each restriction enzyme retain its line number.
- Add a line at the bottom with ***Enter*** (from the last previous line).
- Add a new column (numeric input only, and it should never be necessary) with ***Alt-Enter***.

XIV.B.4. Exit

- ***Ctrl-F5*** or ***Ctrl-e*** to file the changes when done.
- ***Ctrl-q*** to quit and abort the changes.

XIV.B.5. Miscellaneous

- ***Ctrl-o*** toggles the line numbers on and off.
- ***Ctrl-h*** may get a help screen, but this is not guaranteed

# XV. APPENDIX — INSTALLATION AND UPDATE

## XV.A. Installation

The computer must be a PC, a Pentium for example. Mac with a Windows emulator doesn't work.

DNA·VIEW consists of one CD, or a file obtained from the Internet. The installation file has a name like `SetupDNAVIEW....EXE`. If using an installation CD, setup should autostart, but you can browse to the file (**My Computer** etc.) and click on it if not.

The installation procedure is

## XV.B. Preliminary for network installation

### XV.B.1. For installation to a network server,

note that there must be a mapped drive letter designating (part of) a shared folder. See the DNA·VIEW manual §XVI.D Also, **users must have permission to change and create files in the server folder**.

XV.B.1.a. Example: Install to **\\server-1**

» Create or choose a shared folder such as **\\server-1\DNA**, sharename=**sDNA** for example.

» Grant full access to **sDNA** to all DNA·VIEW users.

» Map network drive: Choose a drive letter e.g. X: and on every workstation that uses DNA·VIEW (including **\\server-1**), map X: ⇔ **\\server-1\sDNA**.

Use the same drive letter, e.g. X:, on every workstation.

## XV.C. Begin installation

Answer the prompts.

### XV.C.1. Password

There may or may not be a password.

### XV.C.2. Select Destination Location

XV.C.2.a. If updating, choose the directory where DNA·VIEW already is.

WARNING: Sometimes the installer suggests a location with an extra "dnaview", such as `C:\dnaview\dnaview`. Don't be fooled!

XV.C.2.b. For a **new installation**, anywhere you like. Examples:

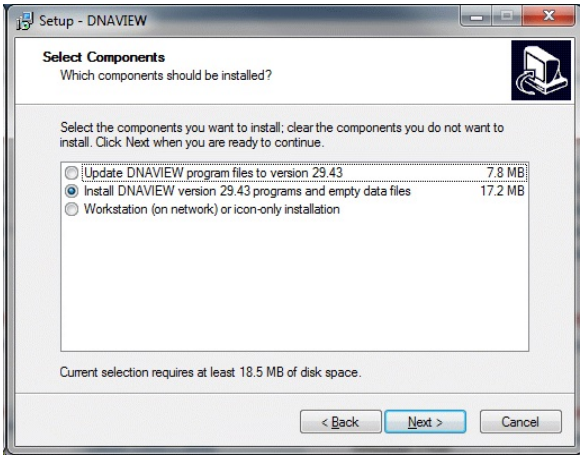
XV.C.2.b.i.) Traditionally DNA·VIEW was installed to `C:\dnaview`.

XV.C.2.b.ii.) Typical Windows style would be `C:\Programs and Files\dnaview`.

XV.C.2.b.iii.) Typical Network installation would be X:\dnaview.

» The same drive letter, e.g. X: should be chosen for every workstation.

» Installation ***must*** be to a drive-letter location. DNA·VIEW *will not run* if you type \\server-1\DNA\DNAVIEW as the install location.




### XV.C.3. Select components

XV.C.3.a. `Update DNAVIEW program files to version 29.43`

Use this option to leave your data unchanged, but to change the program version to a (probably newer) version.

XV.C.3.b. `Install DNAVIEW version 29.43 programs and empty data files`

For initial installation. In case of a network installation, recommended is to choose this option **once** for one install to the server. The installation onto the server can be performed either on the server itself or from some other workstation to the mapped network drive.

XV.C.3.c. `Workstation (on network) or icon-only installation`. In case of a network installation, perform an installation using this option on every additional workstation.

Click **Next**.

XV.C.3.d. **Start menu folder**

Use `DnaView`. This refers to where DNA·VIEW will be found when you click START, All Programs.

If you already have a DNA·VIEW desktop icon folder that you don't want to change, put a checkmark in the box "Don't create any icons."

Click **Next**.

### XV.C.4. Select additional tasks

XV.C.4.a. `Create DNAVIEW desktop icon`. Very handy – an icon on your desktop to click to start DNA·VIEW.

The installation creates a desktop folder called **DnaView Icons** with several icons, particularly the main program startup icon **DNAVIEW**. Drag or copy it directly to the desktop if you prefer. The icons are discussed below in §XV.D.2.

XV.C.4.b. `Create DNAVIEW start menu entry`. Less handy but some prefer it.

Click the obvious buttons to complete installation.

### XV.C.5. First execution of DNA·VIEW

☑ `Allow DNA·VIEW to update itself`

XV.C.5.a. Adjust window size

***Right-click*** on the title bar of the DNA·VIEW window. **Properties**. **Font** tab. **12×16**. ***Ok***.

On Windows XP or older you can also invoke full-screen mode via ***alt-enter***.

### XV.C.6. Install Pater

If installing PATER as well as DNA·VIEW, running the PATER installer – `SetupPATER....EXE` on the same CD – is a separate operation. Use My Computer to double-click on the installer to run it.

## XV.D. Networking

If you will operate DNA·VIEW from several workstations at the same time, you must establish the network protocol so that when two instances of the program running on two different machines need to write information to the same file they will cooperatively take turns.

### XV.D.1. Turn on networking

XV.D.1.a. Open DNA·VIEW on a first workstation (can be the server itself) and

**Housekeeping** ▸ *Maintenance* ▸ *network preferences*

`Do you want to make it networked?` YES

`Which protocol?` *LOCALSHARE* (try this one first).

`Name for this workstation?` **`Octopus`** ***Save*** (a name representing the DNAVIEW installation)

NETWORK LOCK TEST. Now leave the first workstation and enroll a second one to continue the test.

XV.D.1.b. On a second workstation (and additional workstations can be added as needed)

XV.D.1.b.i.) (optional, and see §XI.D.1.a) Copy X:\dnaview\DNACFG.SF to *exactly* **C:\dnaview\DNACFG.SF**.[25]

This allows the workstation to have its individual name and a few other parameters of its own.

» **Housekeeping** ▸ *Maintenance* ▸ *network preferences*

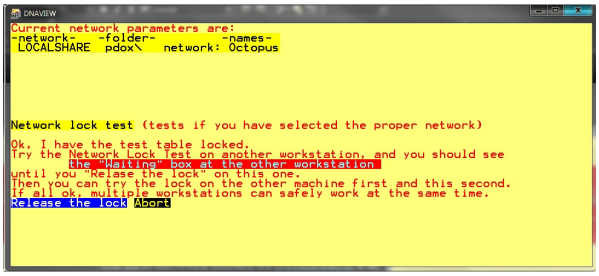
CHANGE: NAME. `Name for this workstation:` **`Anthill`** OK

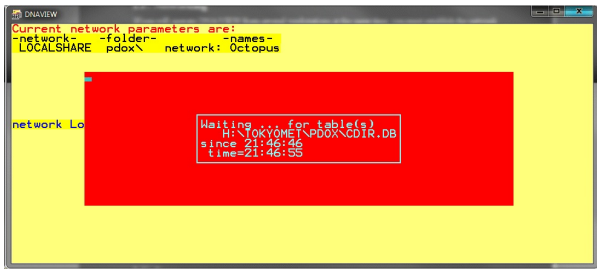
**Note**: The network and any workstation *without* its own **C:\dnaview\DNACFG.SF** will have a workstation name **`Octopus`**, or **`Octopus1`**, etc. CHANGE NAME from such a workstation would change the network name (from **`Octopus`**).

XV.D.1.b.ii.) Continue the network lock test, with the first workstation still waiting:

**Housekeeping** ▸ *Maintenance* ▸ *network preferences*. NETWORK LOCK TEST

[25] That's the default. If you really want it somewhere else, specify the folder in X:\dnaview**dnaview.ini**, **[config]** section, **LCFGFolder=** parameter.




Locking workstation | Waiting workstation

The test is successful if the screen shows a large "waiting" box.

Continue the test by clicking RELEASE THE LOCK, then NETWORK LOCK TEST on the locking (1st) workstation and notice that the waiting box closes on the 2nd workstation and a waiting box opens on the 1st one. Etc.

XV.D.1.b.iii.) If the test *fails*, then on one workstation click CHANGE: NETWORK, choose the next network choice of *OTHERNET*, OK, SAVE. Notice that the `Current network parameters` update. Try the test again.

### XV.D.2. Icons

The installation produces a folder called **Dnaview Icons** on the desktop with a collection of icons.

XV.D.2.a. Icon functions

Their functions are as follows –

XV.D.2.a.i.) **DNAVIEW** – starts DNA·VIEW

XV.D.2.a.ii.) **PATER** – starts PATER

XV.D.2.a.iii.) **DNAVIEW Plot** – starts DNA·VIEW and allows the few graphics commands, i.e. *Plot* §VIII.E for drawing of allele frequency histograms.

XV.D.2.a.iv.) **DNAVIEW Report Viewer** – Utility program to open DNA·VIEW Paternity (§V.C.5.d) or Kinship reports (§VI.F.2.a, the "formatted" version) cleanly in Excel.

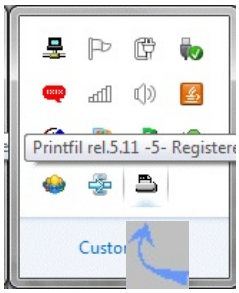
XV.D.2.a.v.) **Connect USB printer** – see How to print, §XV.D.3.

### XV.D.3. How to print to a USB printer

Two possibilities.

XV.D.3.a. The new way is to use the program PrintFil (http://printfil.com).

It might be included with the DNA·VIEW installer. If not you may buy it from the Internet. It works easily and offers some special advantages.




XV.D.3.a.i.) Ensure PrintFil is running when DNA·VIEW is. (If PrintFil was installed with DNA·VIEW – in a folder DNAVIEW\PrintFil – or if you install it to that location this should be automatic because starting DNA·VIEW will also start PrintFil.)

XV.D.3.a.ii.) Set the DNA·VIEW printer port (§IX.F.10) as *print via PrintFil*, which will send DNA·VIEW print jobs to be processed by PrintFil.

XV.D.3.a.iii.) Normally specify the DNA·VIEW printer (§IX.F.9) as *PrintFil*, which

means to use a printer driver compatible with default settings of the PrintFil program. However, a different printer such as Laserjet is also possible provided that you configure PrintFil consistently. In particular the PrintFil's **Raw** checkbox may be useful.

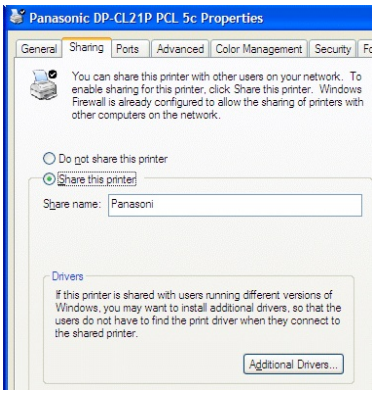
» Right click on the **Printfil** icon in the task bar tray (see figure) and choose **configuration** to make various customizations.

XV.D.3.a.iv.) To include a laboratory logo on every page printed, replace the file **C:\Windows\PrintFil**\yourlogo.bmp.

XV.D.3.b. Traditional way – use one of my customizing icons "LPT1" or "LPT2"

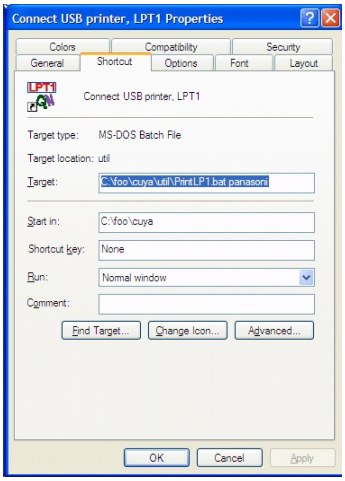
These several icons create the connection that is necessary for DNA·VIEW or PATER, which nominally print to an old-fashioned "parallel" port – LPT1 or LPT2 – to print to a USB or network printer. You will probably only need one of them; which one is a matter of convenience.

Customization of the icon is necessary. If you double click on one of them and it is not yet customized, it will give instructions for customization.




XV.D.3.b.i.) Print icon customization

» Choose the network or USB printer you wish to print to.




» Determine its "Share name". The easiest situation is if the printer can be shared through your own computer. Open "Printers and Faxes" from or near the Control Panel, right click on the printer you want to use, and choose Properties. Choose the Sharing tab. Choose the option "Share this printer" if possible. Note the Share name. If there is none, add one of your choosing, preferably without spaces.

Sometimes the share option for the printer is ***greyed out***. In that case the solution may be to install the *Microsoft Loopback Adaptor* ( per http://support.microsoft.com/kb/839013) to create the illusion that you are on a network.

Be sure that **enable bidirectional printing** (if a local printer) is *not* checked.

Click ok to close the printer properties window.

» Right click on the icon **Connect USB printer, LPT1**. Choose **Properties**. Select the **Shortcut** tab. Click in the **Target** window, hit ***End***, and type ***space*** followed by the printer's share name. Click **Apply**.

» While the **Properties** are open is a good time to complete customization of the print icon per §XV.D.4.

XV.D.3.b.ii.) Establish printer connection

Now you can double-click on the icon itself to invoke it in order to establish a connection with the printer. The connection is supposed to be permanent but if it happens to disappear you can invoke it again.

XV.D.3.c. The **Repair** icon starts some maintenance processes discussed in §XI.G.

XV.D.3.d. **Reset** – When clicked *tells how* to reset the DNA·VIEW data files – deleting all cases and *DNA profiles*. Then you need to follow those instructions to actually delete data.

### XV.D.4. Customizing the icons

There are a few detailed icon option settings that it was not possible to incorporate into the installer. Therefore for best performance and appearance I suggest the following steps, for each icon that you will use:

XV.D.4.a. Right click an icon and choose **properties**

XV.D.4.a.i.) font/window size

» Click the **font** tab and choose a font of comfortable size, such as **font: raster font** and **font size: 12 x 16**.

» Click the **options** tab. Under **Display options** click on the **Windows** radial button.

» Click the **layout** tab and make sure that **Window size** is **80** and **25**.

XV.D.4.a.ii.) enable mouse for menu. Click the **options** tab and make sure that **Quick edit mode** is *not* checked.

### XV.D.5. Start DNA·VIEW

Execute (or allow to execute) the command *Update* (§XI.B) from the **Housekeeping** menu.

### XV.D.6. Graphics checkout

Start DNA·VIEW using the special **DNAVIEW PLOT** icon.

Select *Plot*, choose any database, and hit ***Enter*** to select all the default choices until *DISPLAY*. You should now see a graph on the screen.

If not, exit from DNA·VIEW and examine the file `\DNAVIEW\GSS\CGI.CFG` with a text editor (WordPad will do). Look at the file `CGI.CFG` and compare what you see with the contents of the directory. Are there perhaps incorrect drive letters mentioned in `CGI.CFG`, out-of-date paths, or missing files? Are there any error messages when you start up DNA·VIEW? Call for help.

If *DISPLAY* works, hit ***Enter*** to end the display, and choose *PRINTER*. A similar picture should appear on the printer.

### XV.D.7. Mouse checkout.

The (little-used) commands *PCR Read*, and *Import/Export gel image* require a special mouse driver. For documentation, obtain an old DNA·VIEW manual from the Internet.

### XV.D.8. De-installing a DNA·VIEW System Update

XV.D.8.a. Backup subdirectories

Before installing a new program version it is advisable to save the old one. The essential files to save are the program files `DNATREE*.SF`, `DNAVIEW.WS` and the configuration data files `DNACFG.SF`, `DNAMAINT.SF`, all from the installation folder of DNA·VIEW such as `C:\DNAVIEW`. (Don't worry about anything in the `UPDATES` folder.)

Earlier versions of the DNA·VIEW update routine automatically sequestered these files in folders named e.g. `C:\DNAVIEW\PRE27.12` (if the new version is 27.12). You can delete these subdirectories once you feel you will not need them.

XV.D.8.b. Retreating to a previous version

So long as the old and new version numbers have the same integer part (e.g. 27.04 and 27.12), you can retreat just by copying back the files `DNATREE*.SF` and `DNAVIEW.WS`.

If the old and new version numbers are farther apart, then it depends, so please call DNA·VIEW for special instructions. If you wish to retreat and have NOT already run the DNA·VIEW *Update* command (§XVI.B.1.i.iii), then don't run it.

### XV.D.9. Installing Frequency databases

Allele population frequencies ("databases") are included when DNA·VIEW is initially installed. Additional databases can be added in a variety of ways (the *Import/Export* command options described in §X.C and §X.B). Use the method of §X.C.1 to merge databases into the active DNA·VIEW library from a source in proprietary DNA·VIEW format:

XV.D.9.a. Distributed of DNA·VIEW frequency databases

Large files of frequency databases are included on distribution CD's including the update CD's that are distributed yearly. They are in the **\FREQ** folder.

XV.D.9.b. Importing DNA·VIEW frequency databases from another user

DNA·VIEW users can trade frequency databases by using the *Import/Export* command options described in §X.C.

### XV.D.10. Installation — Check

XV.D.10.a. Graphic printing

Check that \dnaview\CGI.CFG mentions the correct printer driver. If not, you won't be able to print graphs. Other reports aren't affected.

XV.D.10.b. Numlock status

See §IX.F.24 for the *Option* to toggle the numlock status.

XV.D.10.c. Printing

See §IX.F.9 and §IX.F.10 for *Option*s to specify the printer and the printer port, and §XV.D.2.a.v for printing under Windows.

XV.D.10.d. **Extended or expanded memory** for DNA·VIEW under Windows

Especially if you experience a WS FULL error message from DNA·VIEW, edit the file (Notepad, Microsoft Word, or any text editor will do, but be sure to re-save as "Text"!) `\DNAVIEW\APLII\CONFIG.APL` and check the `wssize=` line. The number is the number of kilobytes of RAM that DNA·VIEW will be allocated.

```
wssize=100000
```

is a modest allocation (representing 100 megabytes) considering the amount of memory most computers have available. The minimum is about `5000`; the maximum is about `500000` (half a gigabyte).

XV.D.10.e. **File handles**

DNA·VIEW requires at least 70 of a system resource called "file handles." Otherwise the program may produce an error message `Not enough file handles`.

XV.D.10.e.i.) Windows XP etc.

Ensure the `FILES=` line is at least `FILES=70` in a file called `Config.NT`. There is a sample version of `Config.NT`, with sufficient file handles specified, in the directory where DNAVIEW is installed (possibly named `C:\DNAVIEW`.)

The normal copy of `Config.NT` is located in the system directory — usually at `\Windpws\System32\Config.NT`, but more generally at the symbolic location `\%systemroot%\System32\Config.NT`. It may be invisible. You need administrative rights to change it. Even some administrators are unaware of it, but it is there!

XV.D.10.f. Windows XP/NT/2000 startup glitch

Starting DNA·VIEW or PATER under Windows XP may produce a pop-up window with the message:

> 16 bit MS-DOS Error
>
> The system cannot open COM1 port requested by the application. Click 'Close' to terminate the application

Click 'Ignore' and the program will start ok. To avoid the message altogether, examine two files:

XV.D.10.f.i.) In `dnaview\aplii\config.apl` there is probably an `!----- Environment Information` section (`!` indicates a comment line) followed by an `Env=` line.

Change `Env=2` or `Env=258` to `Env=18`.

Change `Env=0` or `Env=256` to `Env=16`.

XV.D.10.f.ii.) In `dnaview\dnaview.bat`, change any `E2` or `E256` or similar phrase at the end of the line (near the end of the file) that includes `APLPDX` according to the same pattern. (This will apply only to an old installation of DNA·VIEW.)

**XV.D.11. Printing from DNA·VIEW (in Windows)**

Printing from DNA·VIEW or PATER when running under Windows may require a trick.

XV.D.11.a. Printing to an LPT port

DNA·VIEW only knows how to print to LPT1 or LPT2 (§IX.F.10), which are traditional names for plugs on the back of the computer where printers used to be plugged in. DNA·VIEW sends messages to one of these old-fashioned port names in order to print. Windows needs to be told somehow to intercept and re-route all such messages. If the printer is physically plugged into LPT1 (or LPT2) on the local machine, there is no problem. Just tell DNA·VIEW or PATER to print to that port.

A small complication arises when the printer is a network printer and/or a USB printer, which is the usual arrangement these days. Therefore, to print it may be necessary to ask Windows to intercept LPT messages and re-route them to the actual printer. How to do this depends on the version of Windows.

XV.D.11.b. Printing to a network or USB printer from Windows XP (2000, NT, Vista)

See **Connect USB printer**, §XV.D.2.a.v. (For the technically inclined, it invokes a `NET USE` command.)

XV.D.11.c. Printing to a network or USB printer from Windows 98

In Windows 98 there is a “capture printer port” option on the printer properties window. Use it to ask Windows to re-route any requests for the port to which you elect (§IX.F.10 – e.g. LPT1) to print. to the specified printer:

Double click on **My Computer**

Double click on the Printers Folder

Click on Printer

Click on **Properties**

Click on the **Details** Tab (the tabs are at the top of the current window)

Click on Capture Printer Port Button

Set the device as LPT1, or whatever you selected as §IX.F.10.

Choose the path for the desired printer – e.g. \\YourServerName\PrinterName

Check "Reconnect at logon"

Click on OK

## XV.E. Running DNA·VIEW under Windows

DNA·VIEW will run under 32 bit Windows 7 or earlier in an ordinary DOS box. If you are stuck with 64-bit operating systems, documentation for running in Virtual PC or under DOSbox can be supplied on request.

### XV.E.1. Startup

Usually click the **DNAVIEW** icon or choose DNA·VIEW from the start menu.

### XV.E.2. Hardware security key

If you have a hardware security key, it must be plugged into a USB port. In case of a network installation, the port must be on some accessible computer such as the server on which DNA·VIEW is installed.

### XV.E.3. Startup with graphics

To start DNA·VIEW in an enhanced mode that enables a few special DNA·VIEW commands such as *Plot* (§171), use the special icon **DNAVIEW PLOT**. It will only run on Windows XP or below.

Unfortunately this mode comes at a small price – a Windows XP / DOS *process priority* conflict that causes DNA·VIEW to play badly with other Windows processes. Even printing from DNA-VIEW takes15 seconds.

XV.E.3.a. A basic fix – **Lowering priority of DNA·VIEW in Windows**

XV.E.3.a.i.) Start Windows' Task Manager (While holding ***Ctrl-Alt***, hit ***Del***). Click the "Processes" tab.

XV.E.3.a.ii.) Start **DNAVIEW PLOT**.

XV.E.3.a.iii.) Click on the Task Manager.

» Find the DOS process, e.g. by hitting **n** repeatedly. It will be called `ntvdm.exe`. Right click on it.

» From the fly-out, choose `Set Priority`

» From the next fly-out, click on `Below Normal`

» Close the Task Manager with the X.

This change is temporary, lasting only until you close DNA·VIEW.

### XV.E.4. Annoying message

If, when trying to start DNA·VIEW/Toolbox, Windows complains about COM1 and asks "**Close or Ignore?**", click "Ignore" and everything will work fine. This probably only happens on older DNA·VIEW installations. See §XV.D.10.f for the fix.

XV.E.4.a. **Windows Lore**. Did you know?

When DNA·VIEW is running, you can use ***alt-enter*** to toggle between full- and windowed- screen.

***Alt-tab*** cycles among running Windows processes, including DNA·VIEW.

XV.E.4.a.i.) What is that funny Flying-Windows key good for?

The flying-windows key brings up the desktop and the Start menu.
Flying-window+D clears away all windows. Do it again to bring them back.
Flying-window+M minimizes all windows. Flying-window+shift-M to restore them.
Flying-window+E opens the Windows Explorer (i.e. "My Computer")
Flying-window+R opens the **Run** dialogue.

XV.E.4.a.ii.) Host Access Error

In the process of transmitting files from one computer to another there are various ways that the "Read only" attribute can get turned on. For example, copying a file from a CD usually has this result. If DNA·VIEW or PATER then tries to write to that file, the result is the error message `HOST ACCESS ERROR`. You can remove the read-only status and fix the problem by right-clicking on the file name in Windows and changing the "properties".

## XV.F. DNA·VIEW and Networks

DNA·VIEW can be installed on the server and thereby accessed from every workstation.

### XV.F.1. Installation of DNA·VIEW to the server

However, DNA·VIEW can only access files that appear to be located on a traditional drive letter. For example, suppose the server is called `\\Servoir` and you wish to install DNA·VIEW in the folder `\\Servoir\programs\DnaView`. This means that the main data files such as `DNAREADS.SF` will be in that folder, and there will be subfolders such as `\\Servoir\DNAprogs\DnaView\import`.

XV.F.1.a. Establish a place on the server for installation

For example suppose there is a folder `\\Servoir\DNAprogs\` on the server `\\Servoir` within which DNA·VIEW (and perhaps other programs) will be installed. Then an IT administrator must grant universal access – read, write, create, delete – to this folder for the DNA analysts who will use DNA·VIEW from their workstations.

(If access to the entire folder is undesirable, then at a minimum the administrator may create the subfolder `\\Servoir\DNAprogs\DnaView\` and enable universal access to just that folder. This is less preferred because normally the `DnaView\` subfolder is created by the DNA·VIEW installer.)

XV.F.1.b. Establish a drive letter

In order to install DNA·VIEW and also in order to operate it from any workstation, you will need to define a drive letter – using the Windows facility called **Map network drive** – that equates to a folder on the server.

**Map network drive** is something that you do on each client (workstation) machine, not on the server.

☞ *The drive letter mapped must be the same letter for every workstation!*

Suppose the drive letter `P:` is available. For the example above, there are three possibilities:

| map `P:` to | | **Destination location** for DNA·VIEW installation | comments |
|---|---|---|---|
| entire server drive | `\\servoir\` | `P:\DNAprogs\DnaView` | Entire server drive is accessible to workstations |
| just DNA·VIEW | `\\servoir\DNAprogs\DnaView` | `P:\` | Consumes a drive letter for just one application |
| in between ➜ | `\\servoir\DNAprogs\` | `P:\DnaView` ➜ | Normal compromise |

Choose one of the above patterns – probably not the first one – and (from a workstation)

XV.F.1.b.i.) map network drive

» open My Computer, click **tools**, and choose **Map Network drive**.

» Select `P:` as the drive and type in the desired mapped path as **Folder**.

» Check **Reconnect at login**

XV.F.1.c. Install the program

Run the DNA·VIEW installer per §XV.A from any workstation after having mapped the network drive as above.

XV.F.1.c.i.) Select the **destination location** (§XV.C.1) following an example from the table above.

That completes installation to the first workstation

### XV.F.2. Install to other workstations

XV.F.2.a. Map network drive as above for the next workstation

XV.F.2.b. Create a startup icon on the desktop

This can be done by simply copying the icon folder from another workstation, or can be done with the DNA·VIEW installation file (§XV.C.4).

### XV.F.3. File sharing among workstations

When several users run DNA·VIEW at the same time, since they will all be sharing the same files containing cases, DNA profiles, etc. it is important to have some kind of discipline to ensure that simultaneous writing of information to the same file by different users doesn't cause the data files to become jumbled. Ideally the software enforces the necessary discipline by some sort of **file-locking mechanism**.

Some of the DNA·VIEW data is stored in "APL" files (principally those with names of the `DNA....SF`) and these files are automatically robust.

Other DNA·VIEW data is stored in "Paradox tables" and to ensure that they don't become inconsistent or even corrupt some installation care is required.

XV.F.3.a. Network protocol

Use the *Network preferences tool* per §IX.A.8 to establish and check network operation.

XV.F.3.a.i.) Network details and comments

The most popular network is of course Novell (NE 2000 compliant cards), and DNA·VIEW seems to work well with it. However, there are some oddities. Using Windows 95 I suggest choosing a "client network component" of **Microsoft client for NetWare networks**, rather than **Novell Client** due to early rumors of software problems (pre Feb 96, may be ok later) in the latter.

Similarly, in the *Network preferences* (§IX.A.8) includes a network option of *NOVELLNET* but in practice *LOCALSHARE* sometimes works better.

The acid test is to try the *Network Lock Test* which the Network tool offers.

» Miscellaneous notes

Write-back caching can only mean trouble. Disable it.

One successful combination: "Windows Login loaded; IPS, NetBEUI (certainly used), TCP/IP (presumably idle), NE 2000 compliant."

Windows NT is not validated, but should work, since "APL environment switch env=18" — jargon for a parameter setting in DNAVIEW.BAT.

OS/2 installations have worked.

MS-DOS protected mode DPMI. Auto. (I don't know what this means, but wish to retain the note!)

### XV.F.4. Cloned DNA·VIEW on a network

When DNA·VIEW is added to various workstations as described above, all users on the network will access the printer in the same way, will have the same report title on printed reports, will share memory for things like the most recent *DNA odds* worksheet, and so on. Workstation names will vary slightly – e.g. **Snoopy**, **Snoopy1**, etc. if several users are signed on simultaneously. The name variety is generated automatically based on the user-specified name (such as **Snoopy**) and is necessary to ensure file-writing discipline and synchronicity.

### XV.F.5. Personalized network configuration

The simplified – "cloned" – installation can be elaborated to have a custom configuration file (and hence custom report title, etc.) on each client machine. In that case, the structure of a networked DNA·VIEW system is like so:

XV.F.5.a. The programs and data files all reside on the server, such as in `P:\DnaView`.

XV.F.5.b. For any workstation that you wish to "personalize",

XV.F.5.b.i.) Create a folder at the root of the local `C:` drive called `C:\DnaView`.

XV.F.5.b.ii.) Copy a file `DNACFG.SF` into `C:\DnaView\`.

`DNACFG.SF` may be initially be copied from the server – copy `P:\DnaView\DNACFG.SF`.

Consequently from some workstations there are two files `DNACFG.SF` – a local one and a global one. `C:\DNAVIEW\DNACFG.SF` is a control file with information specific to the local workstation, including the workstation name (which appears in the corner of the computer screen above the main menu).

XV.F.5.b.iii.) Customize the local `DNACFG.SF`

You may then customize it in various ways from within DNA·VIEW such as

» Use *Options* to choose a printer, report title, etc.

» Use *Network* to give this workstation its own name, something with more pizzaz than **Snoopy1**.

» Various memory features such as the *DNA odds* worksheet will be local to this station.

## XV.G. Moving DNA·VIEW

(updated – 2011)

To move DNA·VIEW to a new location, such as a new computer, simply copy installation folder (e.g. `C:\dnaview\`) to the new location. Create startup icons (e.g. the desktop folder **DNA·VIEW Icons**) that point to the new location preferably using a DNA·VIEW installer (an icon-only version is available at `http://dna-view.com/downloads/setupDNAVIEW28.78Icons.exe` but any installer will do) or alternatively my manually modifying the old startup icons.

If the DNA·VIEW installation is older than about 2004 there may be some location dependencies, especially drive-letter dependencies, in the installation. Check `DNAVIEW.BAT`, the graphics control file **CGI.CFG** (**Figure 147**, page 313), *Network preferences* (§IX.A.8), and the paradox file location (*Options*, *Paradox directory*).

## XV.H. Create a test platform for a new DNA·VIEW version

(new – 2014)

Assume you have been using DNA·VIEW version 30 for production, and would like to evaluate a newer version, 33.04. Both versions can run in parallel, even on the same computer, for a limited testing period. For the discussion, assume the production system is installed at **Z:\dnaview** on the network drive **Z**. The test platform will be installed on one workstation.

### XV.H.1. Installation

XV.H.1.a. Copy the production system

On the workstation create a folder **C:\DVtest33.04**.
Copy the *contents* of **Z:\dnaview** into **C:\DVtest33.04**.

XV.H.1.b. Careful not to over-write the production startup icon.

Rename the desktop folder **DNAVIEW icons** to e.g. **DV prod icons**. Or, if you start DNA·VIEW with a desktop or start menu item called **DNAVIEW**, rename it to e.g. **DNAVIEW prod**.

XV.H.1.c. Install the new version

XV.H.1.c.i.) Run the update installer for the new version. The installation target is **C:\DVtest33.04**.

XV.H.1.c.ii.) Check the installation box to create a desktop startup icon folder.

XV.H.1.c.iii.) Rename the test startup icon folder **DV 33.04 (test) icons**

XV.H.1.d. Copy any local custom options from the production system

XV.H.1.d.i.) If the production system is networked the workstation may have a local file **C:\dnaview\DNACFG.SF**.

If so, copy that file into the test installation folder replacing **C:\DVtest33.04\DNACFG.SF**.

XV.H.1.e. Ensure no interaction between the two platforms

Edit the file **C:\DVtest33.04\dnaview.ini** (with Word or Notebook). Find the line like

```
LCFGFolder=  ; location for local DNACFG.SF.
```

Change it to

```
LCFGFolder=. ; (dot keeps test platform to itself).
```

XV.H.1.f. Initialize test platform

XV.H.1.f.i.) Open the test platform (using the **DNAVIEW** icon in the **DV 33.04 (test) icons** folder).

XV.H.1.f.ii.) Allow *Update* to run.

XV.H.1.f.iii.) **Housekeeping** ▸ *Network* ▸ DE-NETWORK

### XV.H.2. Using and testing

The systems can now be run in parallel for testing. They can even run simultaneously, though a single workstation running two copies of DNA·VIEW at once will become sluggish.

### XV.H.3. Updating to the new version

When you are ready to upgrade production to the new version,

XV.H.3.a. Use the update installer to update the production version.

XV.H.3.b. Delete the test platform by trashing the folder **C:\DVtest33.04**\.

## XV.I. Web conference

### XV.I.1. What is a web conference?

It's a way of viewing another computer's display remotely, and even of taking control of a remote computer. We use the computer screen for video, and have a phone connection on the side.

A reasonably fast Internet connection – DSL or cable – is necessary. And of course many labs don't allow their data-critical computers such as those running DNA·VIEW to connect to the Internet, so that's a limitation too.

It is therefore useful for

- » demos – Through a web conference link, I can show DNA•VIEW or PATER in operation
- » training – Not as good as being there, but a lot better than just a telephone
- » solving operational problems (debugging etc.)

Normally this means running the software on your computer and letting me watch or even control.

I use Unified Meeting for web conferencing.

### XV.I.2. How to join

XV.I.2.a. If we will want to run DNA·VIEW on your computer, and you use Windows XP or similar operating system, then first be sure that it will run with "below normal" priority as explained above.

- » If you start DNA·VIEW with the regular icon per §XV.D.2.a.i, there will be no problem.
- » Otherwise, please be aware of §XV.E.3.a.

XV.I.2.b. Install meeting control program on your computer (necessary only for 1st meeting on a computer)

So that I can see and possibly operate your desktop, you need to install a meeting control program:

http://www.meetingconnect.net/umgo UnifiedMeeting.msi (US version)

XV.I.2.c. Join a meeting per our mutual arrangement.

XV.I.2.c.i.) Open Internet Explorer (necessary because we need Active-X)

- » Ensure Active-X enabled. **Tools > Security tab > Internet zone > Custom level > Active X controls**

  Binary & script behaviors – enable. Download signed ActiveX controls – prompt. Only allow approved domains to use ActiveX without prompt – enable. Run ActiveX controls and plug-ins – enable. Script ActiveX controls marked safe for scripting – enable.

- » Turn off pop-up blocker.

XV.I.2.c.ii.) Join the meeting

- » http://www.meetingconnect.net . Under **Web Conferencing** click **United Meeting**.
- » Under **Start / Join a meeting – United States** click **Join a meeting**.
- » Allow add-ons to run if asked.
- » Enter the 7-digit Moderator User Login number which I will have supplied: **24**...
- » **Join Meeting as Participant**
- » Enter your name

» **How are you joining the meeting?** `Already dialed in` (save money with direct phone or Skype connection).

# XVI. APPENDIX — REFERENCE TABLES

## XVI.A. Role letter/name assignments

| Paternity roles | Maternity roles | Crime roles | Kinship roles |
|---|---|---|---|
| (required) | | (suggested) | |
| M Mother | F Tstd Woman | M Victim | J PersItem | U Sibling |
| N Mother #2 | G Woman #2 | N Victim #2 | K PersItem#2 | A Sibling #2 |
| O Mother #3 | H Woman #3 | O Victim #3 | L PersItem#3 | B Sibling #3 |
| C Child | C Child | U Unknown | V Victim | C Sibling #4 |
| D Child #2 | D Child #2 | V Unknown #2 | W Victim #2 | H Half-Sib |
| E Child #3 | E Child #3 | W Unknown #3 | M Mother | I Half-Sib#2 |
| A Child #4 | A Child #4 | X Unknown #4 | N Mother #2 | X Other |
| B Child #5 | B Child #5 | Y Unknown #5 | F Father | Y Other #1 |
| Q Child #6 | Q Child #6 | C Unknown #6 | G Father #2 | Z Other #2 |
| V Child #7 | V Child #7 | D Unknown #7 | D Daughter | R Other #2 |
| W Child #8 | W Child #8 | E Unknown #8 | E Dghter #2 | O Other #2 |
| X Child #9 | X Child #9 | A Unknown #9 | S Son | P Spouse |
| Y Child#10 | Y Child#10 | B Unknown#10 | T Son #2 | Q Spouse #2 |
| F Tested Man | M Father | S Suspect | | |
| G Man #2 | N Father #2 | T Suspect #2 | | |
| H Man #3 | O Father #3 | K Suspect #3 | | |

**Figure 154** Role letters and role names

Role letters designate the various individuals or DNA samples that may constitute a case (§V.B.1). The above chart shows the correspondence between role letters and names for the four different *genre*s (§I.E.1.a.iii) of cases.

### XVI.A.1. Role name semantics

For most kinds of analysis the role names are only suggestions and the various roles have no functional distinction.

XVI.A.1.a. *Paternity Case* roles

An exception is the *Paternity Case calculate paternity/maternity* option, which treats M as stipulated parent (! not necessarily "mother"), C, D, etc as children to be tested in trios, and F (and G, H) as putative parent.

XVI.A.1.b. *Screen disaster matches* and personal references

For purposes of mass victim identification – *Screen disaster matches*, it can be advantageous (see §VII.B.1.a.ii) to designate personal items with the role letters J, K, and L, per **Figure 154**.

But otherwise – in a *Kinship* or *Crime* (mixed-stain) context, a person is a Sibling or a Suspect not because of the role letter, but because of the way the letter is used. However –

XVI.A.1.c. Automatically generated kinship scenario

DNA·VIEW can automatically generate a kinship scenario if you use role letters according the the suggestion for Kinship in the role letter table in §XVI.A. This applies to

XVI.A.1.c.i.) The scenario-entry facility described in §VI.F.1.b.i;

XVI.A.1.c.ii.) and optionally can be accessed in *Disaster screening* if you use pedigree searching (experimental facility – documentation pending).

XVI.A.1.d. Custom names

There is no user-friendly interface to re-define the role names, but a new set of names can be provided for free on request.

### XVI.A.2. Official locus list

The tables below give the official DNA·VIEW numbering scheme for all the human PCR loci defined so far.

XVI.A.2.a. To check for a locus not on the list,

XVI.A.2.a.i.) Use the *Maintenance* option *Locus list by chromosome*, §IX.A.3, to see the latest version of the chart, or the corresponding information for RFLP probes etc.

XVI.A.2.a.ii.) Check the web – http://dna-view.com/pcrtable.htm – for an updated version of the table.

XVI.A.2.a.iii.) Check with DNA·VIEW international headquarters.

| | |
|---|---|
| [100-119] | reserved for **Site Specific Use** |
| [200-229] | reserved for **non-human animals** |
| [237-etc] | for additional **Y**'s |
| [258-etc] | for additional **X**'s |
| [300-399] | reserved for **SNP**'s |
| [400-etc] | for additional **Autosomal** |
| [441-491] | European **SNP** panel |
| [500-599] | more **non-humans** |
| [600-] | additional **autosomal** |
| [651-] | **insertion/deletion** loci |
| [700-] | **mtDNA** |

**Figure 155** locus number plan

| | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| D1 | [32] | D1S80 | MCT118 | | [409] | D5S2500 | | D11 | [39] | HRAS1 | 11p15.5 |
| | [86] | F13B | 1q31-q32 | D6 | [68] | SE33 | D6S | | [69] | TH01 | 11p15.5 |
| | [125] | D1S111 | 1q21-31 | | [71] | F13A1 | 6p25-24 | | [136] | D11S554 | |
| | [145] | D1S533 | | | [134] | DHFRP2 | | | [164] | D11S488 | |
| | [162] | D1S518 | | | [149] | DRB1 | 6p21.3 | | [602] | D11S4463 | |
| | [196] | D1S1656 | | | [155] | Penta F | 6q | D12 | [10] | PLA2A1 | 12q23-qtr |
| | [419] | D1S1677 | | | [403] | D6S1043 | | | [16] | D12S391 | |
| | [607] | D1S1627 | | | [408] | D6S474 | | | [70] | VWA | 12p13.3 |
| | [614] | D1GATA113 | | | [414] | D6S2439 | | | [83] | COL2A1 | 12q12-13 |
| D2 | [30] | ApoB | 2p24 | | [415] | D6S1056 | | | [96] | CD4 | 12p12 |
| | [85] | TPOX | 2p25-p24 | | [610] | D6S1017 | | | [142] | D12S1090 | |
| | [147] | D2S1338 | 2q35-37.1 | D7 | [130] | D7S820 | | | [601] | D12ATA63 | |
| | [401] | D2S1371 | | | [131] | IL-6 | 7p15-p21 | D13 | [129] | D13S317 | |
| | [407] | D2S1360 | | | [151] | Penta B | 7q | D14 | [161] | D14S306 | |
| | [420] | D2S441 | | | [197] | D7S1517 | | | [165] | D14S118 | |
| | [604] | D2S1776 | | | [405] | D7S821 | | | [166] | D14S543 | |
| D3 | [64] | L892 | D3S17 | D8 | [88] | LPL | 8p22 | | [423] | D14S1434 | |
| | [127] | D3S1358 | 3p | | [132] | D8S306 | | D15 | [72] | FES/FPS | 15q26.1 |
| | [137] | D3S1744 | 3q24 | | [133] | D8S639 | | | [94] | CYAR04 | 15q21 |
| | [198] | D3S1545 | | | [139] | D8S1179 | | | [154] | Penta E | 15q |
| | [222] | FCA441 | | | [150] | Penta A | 8p | | [194] | CYP19 | 15q21 |
| | [400] | D3S2406 | | | [163] | D8S320 | | D16 | [12] | D16S83 | 16p13 |
| | [606] | D3S3053 | | | [193] | D8S1132 | | | [138] | D16S539 | 16q24 |
| | [608] | D3S4529 | | | [402] | D8S1477 | | D17 | [9] | D17S5 | 17p13.3 |
| D4 | [14] | FGA | 4q | D9 | [144] | D9S304 | | | [146] | D17S766 | 17q26 |
| | [97] | GABRB1 | 4p13-12 | | [152] | Penta C | 9p | | [418] | D17S1290 | |
| | [225] | FCA742 | | | [404] | D9S925 | | | [609] | D17S974 | |
| | [406] | D4S2368 | | | [416] | D9S1118 | | | [613] | D17S1301 | |
| | [412] | D4S2366 | | | [603] | D9S1122 | | D18 | [42] | D18S51 | 18q21.33 |
| | [417] | D4S2639 | | | [612] | D9S2157 | | | [143] | D18S849 | 18q12-21 |
| | [421] | D4S2364 | | D10 | [160] | D10S516 | | | [425] | D18S535 | |
| | [611] | D4S2408 | | | [411] | D10S2325 | | | [615] | D18S853 | |
| D5 | [84] | CSF1PO | 5q33-34 | | [422] | D10S1248 | | | | | |
| | [128] | D5S818 | | | [605] | D10S1435 | | | | | |

**Figure 156** Human autosomal STR loci assigned so far

| | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| D19 | [76] | D19S253 | 19p13.1 | | [199] | D20S156 | | | [153] | Penta D | 21q |
| | [140] | MYD | 19q13.3 | | [413] | D20S480 | | | [410] | D21S2055 | |
| | [148] | D19S433 | 19q12-13. | | [600] | D20S1082 | | D22 | [156] | Penta G | 22q |
| D20 | [92] | D20S161 | | | [616] | D20S482 | | | [159] | D22S684 | |
| | [135] | D20S470 | | D21 | [99] | D21S11 | | | [424] | D22S1045 | |

**Figure 157** more human autosomal STR loci assigned so far

**STR loci – sex-linked**

| | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| **DX** | [18] | DXS52 | | | [269] | GATA165B12 | | | [181] | DYS435 | |
| | [89] | HPRTB | DXq26.1 | | [270] | DXS6809 | | | [182] | DYS436 | |
| | [90] | Amelogeni | | | [271] | DXS10075 | | | [183] | DY-A4 | |
| | [250] | DXS101 | | | [272] | DXS10079 | Xp11 | | [184] | DYS460 | |
| | [251] | DXS10011 | | | [273] | DXS6807 | | | [185] | DYS461 | |
| | [252] | DXS8378 | Xp22 | | [274] | DXS10148 | Xp22 | | [186] | DY-A8 | |
| | [253] | DXS9895 | | | [275] | DXS10103 | Xq26.2 | | [187] | DY-A10 | |
| | [254] | DXS9898 | | | [276] | DXS10146 | Xq28 | | [188] | DYS635 | |
| | [255] | DXS8377 | | **DY** | [167] | DYS19 | | | [189] | YGATAH4 | |
| | [256] | DXS7424 | | | [168] | DYS389I | | | [190] | DYS426 | |
| | [257] | GATA172D5 | | | [169] | DYS389II | | | [191] | DYS447 | |
| | [258] | DXS10135 | Xp22.31 | | [170] | DYS390 | | | [192] | DYS448 | |
| | [259] | DXS10101 | Xq26.3 | | [171] | DYS391 | | | [230] | DYS464 | |
| | [260] | DXS7132 | Xq11.2 | | [172] | DYS392 | | | [231] | DYS446 | |
| | [261] | DXS10074 | Xq28 | | [173] | DYS393 | | | [232] | DYS463 | |
| | [262] | DXS10134 | Xq11 | | [174] | DYS385a | | | [233] | DYS458 | |
| | [263] | DXS7423 | Xq28 | | [175] | DYCAIIa | | | [234] | DYS456 | |
| | [264] | DXS6800 | | | [176] | DYS437 | | | [235] | DYS452 | |
| | [265] | HumARA | | | [177] | DYS438 | | | [236] | DYS449 | |
| | [266] | DXS6789 | | | [178] | DYS439 | | | [237] | DYS385b | |
| | [267] | GATA31E08 | | | [179] | DYS388 | | | [238] | DYCAIIb | |
| | [268] | DXS7133 | | | [180] | DYS434 | | | | | |

**Figure 158** Human sex-linked PCR loci assigned so far

| | | | |
|---|---|---|---|
| D4 | [123] | GYPA | |
| | [124] | Gc | 4q12-13 |
| D6 | [41] | DQAI | 6p21.3 |
| | [195] | HLADRB | |
| D7 | [122] | D7S8 | |
| D11 | [120] | HBGG | 11p15.5 |
| D19 | [121] | LDLR | 19p13 |

**Figure 159** Polymarker locus assignments

### XVI.A.3. Official enzyme and pseudo-enzyme list

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| [1] | Pst I | [7] | Bgl II | [13] | EcoRI+BSSHII | [19] | free |
| **[2]** | **Hae III** | [8] | Pvu II | [14] | EcoRI+PstI | [20] | free |
| **[3]** | **Hinf I** | [9] | BamHI | [15] | EcoRI+SacII | **[22]** | **PCR** |
| [4] | Alu I | [10] | EcoRI | [16] | Eag I | [23] | BCL I |
| [5] | Taq I | [11] | SacII | [17] | **POLY** | **[24]** | **SNP** |
| [6] | Msp I | [12] | BSSHII | [18] | free | | |

**Figure 160** (Pseudo-)enzyme Official List

## XVI.B. Paradox tables

Some of the DNA·VIEW data is maintained in Paradox tables. The tables are described here so that you can build a front or back end to DNA·VIEW if you wish. Documentation including examples can be obtained on-line using a DNA·VIEW **tool**: Select **tools** from the command menu, ***F1*** (**`Push_Me`**) and ***F7*** (**`Paradoc`**).

### XVI.B.1. PDIR

The PDIR table is a directory of people. **Figure 160** shows the table structure and some example records of the version of it that DNA·VIEW will create and maintain. If you wish to create and post to PDIR through some external mechanism, you may add additional fields (such as names).

### XVI.B.2. CDIR

The CDIR table is a directory of cases. **Figure 160** shows the table structure and some example CDIR records. Notice that a case is defined not by a single record, but by a collection of several records having the same case number but different "role" letters. Therefore the primary key has to be on both of these fields. There should also be a secondary key for speed on the "person" field.

The allowable role letters are shown in **Figure 154**, on page 311.

### XVI.B.3. DNAAPPRO and *Post to a text file*

The *Paternity Case* report will optionally (see §V.C.5.b), as a side effect, post the essential data to a Paradox table called DNAAPPRO. The structure is shown in **Figure 160**. The *Case* option *Export paternity calculations to a text file?* (§V.C.5.c, assuming **n** for *If so, use tab-delimited text ("standard"/LIMS) format?*,

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| [1] | CASE | N* | [12] | Af2 | N | [23] | Oktime | A8 |
| [2] | Subbase | N* | [13] | x | N | [24] | Colldate | D |
| [3] | Probe | N* | [14] | y | N | [25] | Colltime | A8 |
| [4] | Enzyme | N* | [15] | b | N | [26] | Compute race | A1 |
| [5] | Ma1 | N | [16] | ProbeName | A10 | | | |
| [6] | Ma2 | N | [17] | EnzymeName | A10 | | | |
| [7] | Ch1 | N | [18] | PI | N | | | |
| [8] | Ch2 | N | [19] | excl | N | | | |
| [9] | Oblig1 | N | [20] | last rec? | A1 | | | |
| [10] | Oblig2 | N | [21] | LabOk | A10 | | | |
| [11] | Af1 | N | [22] | Okdate | D | | | |

**Figure 161** Paradox DNAAPPRO structure

§V.C.5.d) creates records of a similar format, except with commas. There will be one record per case-probe-man-child-mother combination. Here is some sample data and some explanations:

```
 case sub pb/ez mother    child          man
16079   0   8 2 1940 1120    0 1120 0 0 1120 1540
16079   0 15 2 4280 7000 5210 7000 0 0 5210 2550
16079 10   8 2 1940 1110 1110 1940 0 0 1110 1540
16084   0   6 2 1490 2930 2560 2930 0 0 2560 1020

      x       y        b    probe  enzyme  PI   excl
     0.25 0.01866 0.07326 TBQ7   Hae III 116.2 0   0  0
     0.25 0.02882 0.11190 EFD52  Hae III 116.2 0   0  0
     0.25 0.03933 0.15110 TBQ7   Hae III   6.3 0   0  0
     0.25 0.01423 0.05611 pYNH24 Hae III 791.0 0   0  0
```

[2] subcase — which (of several) men, children, or women? The last digit indexes the man — 0 for the first man, 1 for the second man, etc. The 10's digit indexes the child, and the 100's digit the woman. Hence the third record above pertains to the second child of case 16079.

[3 4 16 17] probe/enzyme number/name. The names are included for the occasions that a person might want to read the record.

[5 6, 7 8, 11 12] fragment sizes for mother, child, man in base pairs.

[13 14] the x and y values. x=0 for an exclusion.

[15] b=1−A; i.e. b is the fraction of random men who would be eligible.

[18, 19] PI (omitting exclusions) and total number of exclusions not just for this record, but for this subcase. These fields are included just for convenience in browsing the table.

[20−25] are all created as blank or zero by DNA·VIEW.

[20 21 22] for initials and time stamp if the record is approved by a supervisor. These fields are inspected by the Paternity Reporting Program. If they are filled in, then the record will be collected from DNAAPPRO ("approved DNA data") by the Reporting Program without comment; if they are not filled in, then the Reporting Program will ask for approval before retrieving the record.

[24, 25] These time stamp fields are filled in by the Reporting Program when it collects the record.

[26] the one letter code for the race used to compute the PI for this locus

### XVI.B.4. STRDATA

The *Compare* command will optionally (depending on options setting in *Options* §IX.F.4), as a side effect, post records to the Paradox table STRDATA. Each record of STRDATA is the average of the (typically) several reads (as user-selected when using *Compare*), of a single lane of a particular worklist. The field "Investigation" is 1 or 2 according as the person was listed in the first or second roster column.

```
[1]   Accession          A7*
[2]   Probe              A3*
[3]   Investigation      A1*
[4]   Small size         A6
[5]   Large size         A6
[6]   Worklist           A6
[7]   Probe Name         A10
9105307     32    1      18·4   23·15  1123  MCT118
9105307     32    2      18·2   23·14  1126  MCT118
9105308     32    1      23·14  27·1   1126  MCT118
9105308     32    2      24·0   27·1   1123  MCT118
```

**Figure 162** STRDATA

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To obtain this manual: AABB, 8101 Glenbrook Road, Bethesda, MD 20814-2749, phone (301)215 6556, fax(301)907-6895. $41 or so.

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introducing the likelihood ratio (in the form Y/X) for paternity

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famous early paper which introduced the idea of using Bayes' Theorem for understanding paternity evidence, but failed to understand the role of prior probabilities. They seemed to think that the scientist could be neutral by choosing a 50% prior (as opposed to no prior at all as we understand nowadays).

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# Index