Paternity Programs User's Manual 2020

PATER program version 17.28

May 28, 2020

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 of the form `setupPATER...exe`, which can be invoked using the Windows Explorer if necessary. See Chapter V.

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

Startup Normally installation creates desktop icons for starting the program. Alternatively, **Start**, **Programs**, etc.

If PATER is used stand-alone (i.e. not in conjunction with DNA·VIEW): to **initialize case numbering** the first time you use the program,

use the Initialize a case command (§II.A.6, page 32), then

save the case with the File command (§IV.B.9, page 41), specifying a case number. Subsequent cases created in PATER will by default be assigned consecutively increasing numbers.

Routine operation.

See **Preparing paternity reports** (§I.I, page 17).

Each time you use PATER, select the command Printer Setup (§II.G.7, page 79) before printing to a non-laser printer. (If you have a laser printer this is done for you automatically.)

Contact for information, inquiries, or problems

For information, inquiries, or problems:
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Overview of PATER — chapter I (page 11).

The 2020 PATER manual and program

New versions

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 `setupPATER...exe`.

See **Installation** above.

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

Charles Brenner

PATERNITY PROGRAMS

USER'S MANUAL

PARENTAGE TESTING REPORT

Case 16101-1 [Hawthorne]
August 4, 2011

		Race	Sample
Tested Man:	Reverend Mr. Dimmesdale	(C)	1000-00036
Mother:	Hester Prynne	(C)	1000-00034
Child:	Pearl		1000-00035

SUMMARY OF FINDINGS: Reverend Mr. Dimmesdale is not excluded.
Combined paternity index = 27000000
Probability of paternity = 99.999996% (50% prior probability)

T E S T	OBSERVED PHENOTYPES			CALCULATION OF PATERNITY INDEX		
loci PCR	Mother	Child	Tested Man	Paternity Assumed (X)	Nonpaternity Assumed (Y)	Ratio (X/Y)
D8S1179	12 14	12 15	14 15	0.25	0.101	2.47
D21S11	30	30 30.2	29 30.2	0.5	0.0154	32.5
D7S820	8 12	12	12	0.5	0.0545	9.17
CSF1PO	12	12 14	10 14	0.5	0.0112	44.7
D3S1358	15 17	15	15 17	0.25	0.153	1.63
TH01	8	8	8 9.3	0.5	0.208	2.4
D13S317	12	11 12	11	1	0.246	4.06
D16S539	9 11	9 11	9 11	0.5	0.254	1.97
TPOX	8	8	8	1	0.362	2.76
D18S51	14 17	16 17	12 16	0.25	0.0832	3
D5S818	11 13	11 12	12 13	0.25	0.183	1.37
FGA	25	22 25	22 24	0.5	0.217	2.31

cc: John J Fitzmorton
Patricia M. Cavenaugh

The observed combination of genetic markers of the involved parties is at least 27000000 times more characteristic of paternity by Reverend Mr. Dimmesdale than of paternity by an untested, unrelated Caucasian/ Black(US)/Hispanic man.

13:51:47

The above calculations are according to AABB guidelines.

_____ _____

Charles H. Brenner, Ph.D. Date

Shown here is the client or lawyer version of the report — inclusion format.

Tested Man: Reverend Mr. Dimmesdale Case 16101-1
Mother: Hester Prynne [Hawthorne]
Child: Pearl 2011 August 4

SUMMARY: Nonexclusion FILING CODES: rN m f - t12 [Hawthorne] (PATER 14.19) Combined paternity index = 27000000; Probability of paternity = 99.999996% (50% prior probability); Probability of exclusion (given mother and child) = 99.99999%

T E S T	OBSERVED PHENOTYPES			CALCULATION OF PATERNITY INDEX			
	Mother	Child	Tested Man	Paternity Assumed (X)	Nonpaternity Assumed (Y)	Ratio (X/Y)	Random Man Eligible
loci PCR							
D8S1179	12 14	12 15	14 15	0.25	0.101	2.47	0.364
D21S11	30	30 30.2	29 30.2	0.5	0.0154	32.5	0.0305
D7S820	8 12	12	12	0.5	0.0545	9.17	0.206
CSF1PO	12	12 14	10 14	0.5	0.0112	44.7	0.0223
D3S1358	15 17	15	15 17	0.25	0.153	1.63	0.519
TH01	8	8	8 9.3	0.5	0.208	2.4	0.373
D13S317	12	11 12	11	1	0.246	4.06	0.432
D16S539	9 11	9 11	9 11	0.5	0.254	1.97	0.758
TPOX	8	8	8	1	0.362	2.76	0.593
D18S51	14 17	16 17	12 16	0.25	0.0832	3	0.305
D5S818	11 13	11 12	12 13	0.25	0.183	1.37	0.597
FGA	25	22 25	22 24	0.5	0.217	2.31	0.387

cc:
John J Fitzmorton
Office of the District Attorney
Walden, Ohio

Patricia M. Cavenaugh
1422 N. Hampshire St.
Walden, Ohio

Effect of prior probability on the probability of paternity:

prior	10%	25%	50%	75%	90%
posterior	99.99%+	99.99%+	99.99%+	99.99%+	99.99%+

Avuncular and Incest Indices
Degree (0=alleged father or parthenogenesis, 1=uncle, etc.) of

relation	0	1	2	3	4
Paternal	27000000	209000	4720	278	36.1
Maternal	0	41	37.2	16.2	7.11

On the basis of the genetic observations listed above a child of Reverend Mr. Dimmesdale would be 27000000 times as likely to have the genetic types of Pearl as would the child of a random man of similar ethnic background.

chg: 2011 August 4 13:51:47

Shown here is the laboratory files report version.

PARENTAGE TESTING REPORT

Case 16101-2 [Hawthorne]
August 4, 2011

		Race	Sample
Tested Man:	Roger Chillingworth	(C)	1000-00037
Mother:	Hester Prynne	(C)	1000-00034
Child:	Pearl		1000-00035

SUMMARY OF FINDINGS: Roger Chillingworth is not the biological father of Pearl.

T E S T	OBSERVED PHENOTYPES			
	Mother	Child	Tested Man	
loci PCR				
D8S1179	12 14	12 15	13 15	
D21S11	30	30 30.2	29 31	Inconsistent
D7S820	8 12	12	8 10	Inconsistent
CSF1PO	12	12 14	11	Inconsistent
D3S1358	15 17	15	16 17	Inconsistent
TH01	8	8	7	Inconsistent
D13S317	12	11 12	9 11	
D16S539	9 11	9 11	9 11	
TPOX	8	8	8 11	
D18S51	14 17	16 17	12 16	
D5S818	11 13	11 12	10 11	Inconsistent
FGA	25	22 25	21 22	

Our opinion of **non-paternity** is based on the above noted inconsistencies. For each noted inconsistency the combination of phenotypes as observed occurs with negligible frequency when the tested man is the father, as compared to the frequency when he is not the father.

Or (listing the loci for AABB compliance, §II.G.4.l.iv):

Roger Chillingworth is **not** the biological father of Pearl. This conclusion is based on inconsistencies in loci D21S11, D7S820, CSF1PO, D3S1358, TH01, and D5S818.

cc: John J Fitzmorton
Patricia M. Cavenaugh

13:51:47

The above calculations are according to AABB guidelines.

_____ _____

Charles H. Brenner, Ph.D. Date

Shown here is the client or lawyer version of the report — exclusion format.

I. PATER Overview

I.A. Overview

PATER is a program that makes reports and calculations for paternity cases. It makes all serological calculations, saves work according to "cases", and prepares reports in various versions suitable for laboratory files and for submission to the court.

It also has numerous other facilities, such as

including DNA results in the case (which may either by typed in or imported automatically from DNA·VIEW

defining gene frequencies and races by the user

preparing summary reports, including phenotype counts across cases

showing calculation details for any serological system.

I.B. Starting PATER

Normally installation creates desktop icons (in a desktop folder called **DNAVIEW ICONS**) and/or Start Menu entries for starting the program.

See §V.B for fuller instructions.

I.C. Concepts of operation

I.C.1. PATER has a main menu of commands.

You can choose in any of several obvious ways. After each command executes, you normally are returned to the menu.

I.C.2. PATER is case oriented.

Paternity cases can be numbered from 1 to 9999999 (any seven digit number), or, the case number can be 8 or 9 digits provided that the leading 4 digits represent a year, 2000≤year< 2100 (i.e. 200912345). You may allow the program to assign consecutive numbers, or you may assign a new arbitrary number to each case. A case consists of usually 3 people, but perhaps more, and maybe only two (missing mother). When there are many people in a case, each set of mother-child-man is a trio or "sub-case", and is considered separately for reporting. However, data (phenotypes) for the same person are shared among different trios. Sub-cases are numbered with a "dash number" following the main case number, such as 1234-2 for case 1234, 2nd man. Sub-cases are discussed fully in §I.D.

Each person is defined by an Accession number (which links to DNA·VIEW), a name, and usually a race. More on races below.

At any time PATER has one (sub)case active. You can see what it is with the command **Display**.

Thus any report printed or new data entered will pertain to that trio. However, if you fetch or create a new case, the program is careful to offer to save away any changed data.

I.C.3. PATER can either calculate or import calculations from DNA·VIEW

I.C.3.a. PATER calculation of PI for a system/locus

PATER has built-in tables – alleles and frequencies – for the traditional serological systems including ABO, HLA, etc.

You may define DNA systems – STR, SNP etc. – in the same way and use PATER to calculate them as well.

I.C.3.b. PATER can import DNA calculations from DNA·VIEW

More typically though users use DNA·VIEW to calculate DNA systems. There is communication between the two programs so that PATER can detect the DNA·VIEW calculations and merge them into the paternity/maternity report that it produces.

I.C.4. The most important commands for routine casework:

Initialize is used to begin a new case. It asks you for accession numbers, names, and races. The case will temporarily be numbered 0, which will be changed to the next sequential number when the case is saved routinely. (However, the command **File** will also let you assign your choice of unused case number and file the case.)

Retrieve is another way to select a case as the currently active case. It prompts you for a case number, which is usually a number that already exists (either in PATER or in DNA·VIEW). However, it need not be. **Retrieve** is willing to create a new case with the number you select.

So the fact is, you never need to use **Initialize** unless you like sequential case numbers. In particular if using DNA·VIEW as well, probably the cases are first defined in DNA·VIEW then are created in PATER via **Retrieve**.

In means "input genetic types for computation". All the usual systems are included, including HLA A-B haplotypes. User-defined STR systems may also be treated this way.

Report prints the several possible format of report.

I.C.4.a. Additional commands that are often useful for casework include:

Add person to add extra men or children (or women) to an existing case.

Delete to delete phenotype data for a system selected wrongly.

Printer setup specifies whether to send reports to printer, screen, or file, and the kind of printer (normally generic Laserjet).

In DNA to edit DNA data imported from DNA·VIEW. (DNA data entered this way is *not* computed by PATER, although you may edit/enter the X and Y values.)

I.C.4.b. Other necessary facilities:

Quit from PATER

Change frequencies Establish your own gene frequencies.

Create genetic system Create an STR system.

HLA splits

I.D. Extra people in a case

There can be multiple children (up to 9), men (up to 3) or even women (up to 3) in a case. All the data will be saved under the same case number (e.g. 1234). But each time you **Retrieve** (§II.A.10) the case number you will have to select one particular man, woman, and child by menu selection.

I.D.1. Sub-case numbering

When the program refers to a particular trio from a multiple-party case, it appends a subcase or dash number to the case number. The sub-case number is in principle three digits:

```
1234-wcm
```

where **w** is the woman number, **c** is the child number, and **m** is the man number. A number of **0** is used to mean "the one and only" whereas **1** means "number 1 of several". Thus, in practice the woman number is nearly always 0. Further, leading 0's are elided from the subcase number.

Based on the foregoing, here are some examples:

1234-2 2nd man, only woman and child

1299-12 2nd man, 1st of several children, only woman

2000-20 2nd child, only man and woman.

I.D.2. Ramifications with other features

I.D.2.a. Sub-cases and phenotype input

Phenotype data, created by **In — enter STR or serological phenotypes** (§II.A.4, page 28) is properly shared between sub-cases. That is, if you enter data for woman, man, and child 1 (sub-case –10) then **retrieve** sub-case –20 (woman, man, child 2), you will see that the phenotypes for the adults are already there and you only have to ***Space*** through them.

I.D.2.b. Sub-cases and DNA data

DNA types imported from DNA·VIEW, however, are *not* shared among sub-cases. That normally doesn't raise an issue because DNA·VIEW generates the data for each relevant sub-case. But it is necessary to keep in mind if you edit using the In DNA command; be sure to edit each relevant sub-case separately.

I.D.2.c. Sub-cases and **Maternity**

Suppose you have a multiple person case with sub-cases 1234–1 and 1234–2, and you invoke **Maternity** (§II.B.7), then save the corresponding maternity case at number 2000. The corresponding maternity sub-cases will then be 2000–1 and 2000–2, as seems logical and obvious.

The sub-case numbering scheme was carefully devised with the maternity facility partly in mind. That's why the last digit refers specifically to **man** (rather than to "tested parent", which would be awkward).

I.E. Remarks & Caveats

I.E.1. Gene frequencies.

An attempt has been made to supply frequency data for several populations and all serological systems. However, the correctness of the numbers is the responsibility of the user.

I.E.2. Mysterious exclusions.

Occasionally you might see an exclusion even though the phenotypes look consistent. A frequency of 0 for some gene could account for this. Check the frequency tables.

I.E.3. Races

There are up to THREE versions of race for each person:

I.E.3.a. The DNA·VIEW race

which is next to the accession number input block. This race is used mainly for DNA computations and is a small letter (c, g, b, etc.)

I.E.3.b. The PATER calculation races

entered by the user in response to the prompt

```
0=Cauca 1=Hispa 2=Black 3=Asian 4=German
Race for alleged father, [mother], [random man]:
```

The race here is a number (perhaps different numbers for mother & random man, perhaps not).

The number may be several digits to indicate mixed race. For example, **4442** would mean 3/4 German, 1/4 Black and so **30** of course means half Cauc, half Asian. But don't say **03** — leading 0's are ignored.

The Caucasian tables are US Caucasians, including Terasaki tables for HLA. Some of the serological systems are not common in the US, and frequencies if any are unreal.

I.E.3.c. The Filing Code race

Associated with each (sub)case, PATER keeps a string that it calls "filing codes." These are unimportant for casework, but are used for compiling summaries. Included among the "filing codes" are 1-(capital) letter codes for races, which are used when compiling phenotype counts. The race part of the code might look like **fC mB** meaning that the alleged father is "C" and the mother is "B".

I.F. PATERNITY programs and DNA·VIEW — typical routine

PATER does serological computations. DNA computations, especially for RFLP minisatellite systems, are typically done by DNA·VIEW. PATER just imports them, and includes them on the report. STR systems may be done either way. They can be treated by PATER in the same way as serological systems, but if DNA·VIEW is available it is better to let DNA·VIEW handle them instead.

When using DNA·VIEW together with the PATERNITY programs described herein, the following scheme would be typical in order to assign code numbers logically:

I.F.1. Accession numbers (or patient numbers — the system does not make a distinction) are chosen by laboratory people and assigned to clients. Possibly this is done by removing the next label sticker from a pile of preprinted labels.

Accession numbers are of the form `yyyy-xxxxx`, where `yyyy` is a number of one to four digits, typically representing the year (although it doesn't have to), and `xxxxx` is a number of from one to five digits which may be a sequence number. When these numbers are printed by PATER reports, each part of the code may be padded with leading 0's: `90-00123`, or `01-00001`, or `2000-00123` (rather than `90-123` or `1-1` or `2000-123`). However, the leading 0's don't need to be typed in — they are not significant.

The "year" part `yyyy` may not be 0. Four digits are allowed for the year part of course to accommodate the year 2000 and beyond (especially since a year code of `00` is not allowed!). However, this means there might occasionally be confusion as to whether to enter 2-digit year codes, or 4-digit. In order to avoid the user accidentally and confusingly creating two different versions e.g. 98-12345 and 1998-12345 of the same person, the program has the following provision—

Suppose that 98-12345 is already defined and you type in the number 1998-12345 which is a new, undefined number. In that case, the program will pop up a dialogue box warning you of the situation and asking whether you really mean

A. to have just one number, 98-12345, the one that already exists, or

B. to have both numbers, which will represent two different unrelated people.

Hitting ***enter*** will select option A (because I think it is the more likely). To select option B you have to move the bounce bar first.

I.F.2. In **PATER**, use program (**Initialize a case** — §II.A.6, page 32) to create a paternity case, and to define the accession numbers. The program will assign a case number.

I.F.3. In **DNA·VIEW**: Perform a DNA analysis in DNA·VIEW as detailed in the DNA·VIEW manual. Be sure that, when you run the **Paternity Case** report calculation, the options for posting DNA calculations to Paradox files are set correctly for transfer of data to PATER.

I.F.4. In **PATER**:

I.F.4.a. Use **Retrieve** (§II.A.10, page 35) to recall the case.

I.F.4.b. You can modify or add to the DNA data using **In DNA** (§II.A.5, page 30).

I.F.4.c. If there are serological results (or STR types not processed through DNA·VIEW), enter them using **In — enter STR or serological phenotypes** (§II.A.4, page 28).

I.F.4.d. Use **Report** (§II.A.7, page 33) to print the report.

I.F.5. If you want to change the spellings or other *properties* of any loci that appear on the paternity reports, use **Housekeeping**→Maintenance→locus parameters and preferences in DNA·VIEW, or the tool **Serology** (§II.H.1.b, page 83) in PATER.

I.G. COMMAND: Quit from PATER

Close the PATER program.

I.G.1.a. Errors

In case of a prompt similar to `Have you sent off the error report from May 6 yet?` please see §I.H.2.c.

I.H. Exceptional Circumstances

I.H.1. Aborting

Ctrl-Break

Hold the ***Control*** key and press ***Pause***. This will immediately interrupt any operation in progress and return to the main menu.

See also:

Quit from PATER §I.F.5, page 16

I.H.2. Program Errors

Despite the greatest care in preparation and testing, programs inevitably have errors. I need your help with both kinds:

I.H.2.a. **Errors in answers** 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 reports, with a frequency depending on your degree of confidence. When you get a new version of the programs, be especially watchful.

I.H.2.b. **Fatal errors** 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`. At the point you may call for help (please do if possible), and/or press ***F3*** to resume from the Command menu.

I.H.2.c. Please **report errors**, both to help yourself and because this is one way to keep the program in a constant state of improvement. The easiest and best way is to email the `DNAERROR.SF` file (from the `DNAVIEW\` main folder).

I.H.2.d. **Error recovery**

Aside from that, here are some procedures that will sometimes get you past such a problem:

» Turn the computer off and back on again. Sometimes a computer gets into a strange state.

» Waiting for Laserjet! If PATER expects a laserjet printer (II.G.4.g) and the printer isn't plugged into the computer or isn't turned on, PATER will hang on startup. Hit ctrl-pause (break) and wait 60 seconds.

» Waiting for DNA·VIEW. Sometimes a red message appears on the screen "WAITING FOR TABLE ...". It should normally only last for a second. If it persists for longer, it may be necessary to sign of from DNA·VIEW or PATER or both, and to delete any stray Paradox locking files using the **`fix`** utility described in the DNA·VIEW manual.

» Try looking at the data in a different way. If you can determine that some part of it is peculiar, try deleting that part.

I.I. Paternity Reports

I.I.1. Preparing a Report

I.I.1.a. The typical routine, when PATER is a stand-alone program (without DNA·VIEW) consists of the sequence:

» COMMAND: **Initialize a case** (§II.A.6, page 32)

or COMMAND: **Retrieve** (§II.A.10, page 35)

(**Retrieve** is better if you want to assign case numbers; **Initialize** if you like consecutive numbers.)

either of which creates an empty case, then

I.I.1.b. to enter data, print, and file the case

» COMMAND: **In — enter STR or serological phenotypes** (§II.A.4, page 28)

» COMMAND: **In DNA — enter DNA data** (§II.A.5, page 30)

» COMMAND: **Report** (§II.A.7, page 33)

(Print the inclusion or exclusion (§II.A.9, page 34) report.)

That's all there is to it. The case will automatically be saved under a computer assigned number, known as the case #. This number will appear on reports, and is mentioned on the screen.

You might notice that the computer will suggest **In** after you **Initialize**, and **Report** after that. These are only suggestions; you can accept them by hitting the ***Enter*** key, or override them by typing something else.

I.I.1.c. Alternatively, you can create a computer case number without printing a report by doing §I.I.1.a above and then:

» COMMAND: **File — save the current case** (§II.B.5, page 39).

The case number will be reported to you. Later, to fill in the data when it becomes available:

» COMMAND: **Retrieve** case # ... (§II.A.10, page 35)

and continue with §I.I.1.b above.

I.I.2. Fixing Errors

The following commands are used to correct errors:

People — correct spelling	(§II.B.8, page 43)
Ethnic — respecify race(s)	(§II.A.3, page 27)
In — enter STR or serological phenotypes	(§II.A.4, page 28)
Delete phenotype data	(§II.A.1, page 27)

Suppose for example the child is misspelled on the report for case 1001. Use the sequence

COMMAND: **Retrieve** 1001	(§II.A.10, page 35)
COMMAND: **People**	(§II.B.8, page 43)
COMMAND: **File**	(§II.B.5, page 39)

(or COMMAND: **Report**, which will volunteer to file the changed data before it prints the report).

In a similar manner, you may retrieve a case and add further phenotype information, using **In — enter STR or serological phenotypes** or **In DNA**.

I.I.3. Various Racial Assumptions

COMMAND: **Ethnic** (§II.A.3, page 27)

lets you specify the races of the parties. It optionally forces recomputation of all systems entered via In - enter STR or serological phenotypes (but not DNA types imported from DNA·VIEW or entered with In DNA), which is often a convenient thing to do. For example, you might want to produce several versions of the report using various racial assumptions. **Ethnic** - **Report** - **Ethnic** - **Report** ... to do this.

See descriptions of the various commands for further details.

I.I.4. Report Versions and Formats

The paternity case report comes in two *versions*, and normally you will print out both versions for each case.

I.I.4.a. Laboratory report version

This is the compendious version, intended to be internal to the laboratory and not normally for outside distribution.

There are two slightly different formats of this report depending on whether you use the **Report** (§II.A.7, 33) or the **Immigration** command (§II.B.6, 41).

I.I.4.b. Client report version

The client report is minimal. It has the essential information about the testing, without extraneous details that would attract unnecessary questions. Send one copy to each interested party in the case, perhaps with a standard cover sheet that explains the nature and meaning of the testing.

There are three different client report *formats* — Nonexclusion (page 7), Exclusion (page 9), and No opinion ("Other").

I.I.4.b.i.) The *nonexclusion* client report (page 7) has the typing results and columns of numeric data, such as the paternity index. The columns that appear on the report can be customized using the **Option** PATER client report columns (§II.G.4.l, page 74).

I.I.4.b.ii.) The *exclusion* client report (page 9) has the typing results and no numbers. Each system that is inconsistent with paternity is noted with the word "inconsistent."

I.I.4.b.iii.) The *other* client report has just the genetic results.

The program will determine whether there are exclusions in the course of making its computations. Therefore, normally a Nonexclusion or Exclusion report is produced, as appropriate, as a matter of default. The user may over-ride the default such as to produce an `Other` format report either via the *Code* (§II.B.3) command, or at the moment of printing.

I.I.4.b.iv.) The *No opinion* client report has the typing results only.

If you want the No opinion report, you can elect it when you execute

COMMAND: **Report** (§II.A.7, page 33)

I.J. Entering data, 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.

I.J.1. Buttons

(new – 2010)

All modes make use of buttons (e.g. **Figure 4**) – 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:

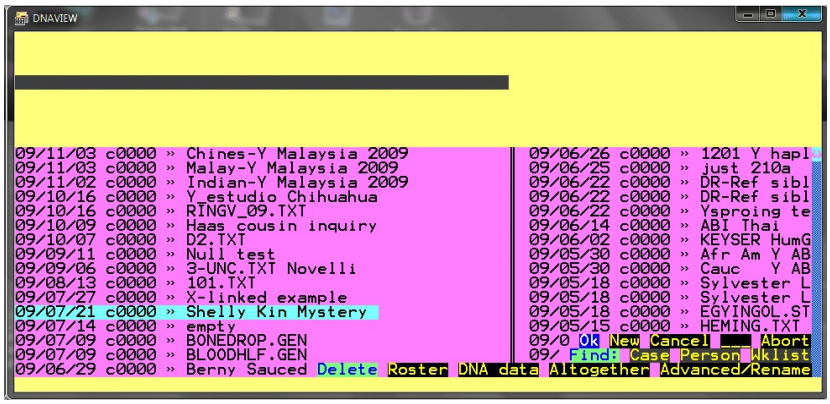


Figure 3 menu selection and buttons

I.J.1.a. Hot button

If one of the several buttons is highlit yellow, the others being white (OK CANCEL), then

I.J.1.a.i.) ***enter*** operates the yellow (hot) button (except ***ctrl-enter*** during multiline input)

I.J.1.a.ii.) ***tab*** and ***shift-tab*** cycle the yellow highlight among the buttons

I.J.1.b. Standard buttons and Key customs

I.J.1.b.i.) ***Esc*** operates the **Cancel** or similar button, and backs up one operation.

I.J.1.b.ii.) ***Ctrl-z*** operates undo, restoring the initial contents of a type-in area.

I.J.1.b.iii.) **?** is for help.

I.J.1.b.iv.) The button ____ erases the typed-in keystrokes so you can restart selection.

I.J.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.

I.J.1.d. Button help

The list of hot-keys is shown if you ***right-click*** on any button.

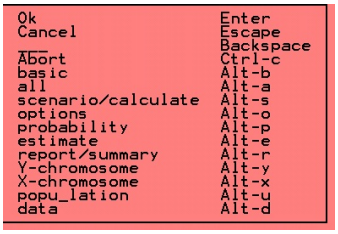


Figure 4 Example button hot-key cheat sheet

I.J.2. Selection from a list of choices

(enhanced – 2011)

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:

I.J.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*.

I.J.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 4** will be arranged in panels which are traversed with ***left-*** and ***right-arrow***.

I.J.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.

I.J.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.

I.J.2.b. **Confirm** with one of the buttons (or it's hotkey equivalent such as ***esc***).

I.J.3. Small Number Answer

When the program requires a small integer answer such as 0, 1, or 2 the selection is with buttons.

I.J.4. Yes/No prompts

Answer **`y`** or **`n`**; do NOT also strike ***Enter***.

I.J.5. 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:

I.J.5.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 §I.J.5.b.

I.J.5.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 keys to make the entry process efficient. (recent – 2007)

I.J.5.a.ii.) First among them is the *help* key: (recent – 2007)

F1 for a popup that documents the special keystrokes described below. 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.

I.J.5.a.iii.) Editing keystrokes (recent – 2007)

I.J.5.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.

```
Left-Dbl-Click   Exit -- accept window contents
Left-Click       Move cursor
Ctrl-C           Break -- abort to main menu
Ctrl-Z           Restore window to initial contents
Ctrl-Insert      Insert empty line above cursor
Ctrl-Delete      Delete line at cursor
Ctrl-a           Cut rectangle corner-to-cursor
Ctrl-v           Paste the cut rectangle
PageUp           Recall previous response
Ctrl-Up-Arrow    MENU of previous responses
PageDown         Recall next response
Ctrl-Dwn-Arrow   Recall next response
Right-Click      Clear window
Rgt-Dbl-Click    Clear window
Ctrl-Shift-Del   Delete current response from history menu
Alt-F6           HELP screens for program
F1               HELP editing keystrokes (this screen)
```

Figure 5 useful keystrokes during text entry (recent)

» ***Ctrl-z*** restores the text as it was before you began editing.

I.J.5.a.v.) **Navigational arrows** work as expected.

I.J.5.a.vi.) Memory (recent)

» ***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. (recent)

» ***Del*** deletes an item from the list of previous responses. (recent)

I.J.5.a.vii.) Copy-and-paste (recent – 2007)

» ***Single left-click*** to position the "mark" (by default at the beginning of the type-in area).

» ***Single right-click*** to capture a rectangular block with "mark" as the opposite corner. (Also repositions the "mark".)

» ***Single left-click*** again to position "mark" where you desire to paste.

» ***Ctrl-v*** to paste the captured rectangle with upper-left corner at "mark"s.

I.J.5.a.viii.) 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.

I.J.5.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 (§III.H.1), ***Esc***.

I.J.6. People & Accession Numbers

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.

I.J.6.a. Editing the Accession Number

The accession number is in two parts, separated by −. The first part is one to four two digits, and may be used as a year code (e.g. **`91`**). Type **`-`** or ***space*** to advance to the second part, which is up to five digits. Leading zeros are not significant. The year part cannot be 0; specifying 0 for the year part has the effect of deleting the accession number. The second part also cannot be 0.

Terminate the accession number entry with ***Enter*** or ***Tab*** to 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.

I.J.6.b. The Accession Race

The accession race is a single lower case letter. Hit **`?`** to pop up the list of races.

The accession race is the DNA·VIEW race (§I.E.3.a, page 14) used by DNA·VIEW for DNA computations and for classification of people into databases. It has no real function in PATER except that, when you later enter a PATER calculation race (§I.E.3.b, page 14) the program will remember the correspondance and in future will prompt you with the correct PATER race when the DNA·VIEW race is known to it.

I.J.6.c. The Accession Date

Dates are input as month, day, year, with today's date usually presented as the default. The month is accepted as a choice (§I.J.2, page 19) from a list of the months; day and year are numeric input (§I.J.5, page 20). Terminate selection for each field with ***Space*** to continue to the next field; terminate with ***Enter*** or ***Tab*** to accept the remaining default choices.

Accession dates are used only for documentation.

I.J.6.d. Entering accession numbers — example

The box below is the first step in initializing a new case, which happens either with the command **initialize**, or when the **retrieve** command is used with a previously non-existent case number. The process of entering the accession numbers is illustrated.

```
Case 0            race(s): -
Pat'y
M     Mother  92  -1235  -
                   OK EDIT DEL CANCEL ABORT
C      Child       COPY FROM A CASE GENERATE SEQUENTIAL
                   ▲ ▼ DONE UNDO ____ HISTORY ?
F Tested Man
```

Type **92** *Space* **1235** *Space* **c** *Enter* to define the mother as a Caucasian with today as accession date.

Eingabe: **92** *Leertaste* **1235** *Leertaste* **c** *Enter*, um die Mutter als Kaukasierin mit Eingabedatum von heute zu definieren (92 = Jahr; 1235 = laufende Nummer.)

```
    Mother  92-1235  c          September  9,   92
     Child  92           00/01/00
Tested Man              00/01/00
```

Type *Space* (for 92) **1236** *Space Space* (child race is -) *Enter* to define the child with today as accession date.

Eingabe: *Leertaste* (für 92) **1236** *Leertaste Leertaste* (Rasse des Kindes is immer -) *Enter*, um das Kind mit Eingabedatum von heute zu definieren.

```
    Mother  92-01235 c   92/09/09
     Child  92-1236  -          September 9,   92
Tested Man  92          00/01/00
```

Type *Space* **1234** *Space* **c** *Space Space Enter* to define the man as a Caucasian with today as accession date.

Eingabe: *Leertaste* **1234** *Leertaste* **c** *Leertaste Leertaste Enter*, um den untersuchten Mann als Kaukasier mit Eingabedatum von heute zu definieren.

```
    Mother  92-01235 c   92/09/09
     Child  92-01236 -   92/09/09
Tested Man  92-1234  c          September 9,    92
```

I.K. Files in the system

Here is general information about files created or used by the system:

\dnaview\pater\PATER.WS and PATREEn.SF (n=0,5,6,7,8) contain the paternity program. They are replaced by each new PATER version. The old versions of these files are sequestered in a directory with a name like

```
\Dnaview\Pater\Pre9.25
```

You might want to examine and eliminate such directories from time to time.

The files in \DNAVIEW\APLII are also program files that don't change. It is proprietary software Copyright 1983-1994 Manugistics, Inc.

\dnaview\CONFIG.APL is an Ascii file than has a few configuration parameters you may be directed to modify.

The files in \DNAVIEW\UPDATES are only used temporarily, by the *Update* command, when a new version is installed.

\dnaview\pater\GENES.SF contains all the data pertaining to genetic systems. The commands

HLA- specify splits

Change or create allele frequencies

New race - change or create race name

Create genetic system

change this file and only this file, so you can back off any changes from those commands by the precaution of making a backup copy e.g. **GENES.BAK** before making the changes, then reversing the procedure to back off the changes.

\dnaview\pater\cases\ (early PATER systems use \dnaview\pater\ for these files) contains files with names like CS1001.SF or C1234567.SF which contain cases. Up to 100 cases share a single file; the system automatically starts a new case file whenever necessary. CS1001.SF would contain cases from 1001 potentially up to 1100.

CS... files may be freely erased from the hard disc to gain storage space, and later copied back.

There are also several Paradox tables (\dnaview\pdox*.DB) that contain information about cases. However, the C*.SF file also usually contains a copy of such information.

\dnaview\pater\HELP.SF contains the help screens (i.e. the manual). \dnaview\DNAERROR.SF is normally empty, but is the repository for error reports (q.v.). \dnaview\DNACFG.SF has configuration information. \dnaview\DNAMAINT.SF is mainly part of DNA·VIEW, but is referenced for probe and enzyme names.

See also:

Error reports §I.H.2, page 16

II. Programs

Paternity programs are discussed in categories:

- II.A PATERNITY — routine casework,
- II.B PATERNITY — advanced casework,
- II.C GENETICS — genetic systems and allele frequencies,
- II.F SUMMARY — operations ranging over all cases,
- II.G MISCELLANEOUS,
- II.H LEAVE MENU — exit, and special facilities.

You may, optionally, configure the main command menu to reflect this organization. Select the command **Options**, *Command menu levels* (§II.G.4.b, page 70)

II.A. PATERNITY CASEWORK

The following subjects or COMMAND's pertain to the "current case," created via **Initialize**, **Retrieve**, or **Maternity**.

Delete	phenotype data	§II.A.1, page 27
Display	brief summary of active case	§II.A.2, page 27
Ethnic	respecify race(s); recompute PI	§II.A.3, page 27
Exclusion	report format when exclusion	§II.A.9, page 34
In	enter STR or serological phenotypes	§II.A.4, page 28
In DNA	edit DNA data from DNA·VIEW	§II.A.5, page 30
Initialize	create a case called case #0	§II.A.6, page 32
Report	print a paternity case report	§II.A.7, page 33
Reprt	display a paternity case report	§II.A.7, page 33
Retrieve	a paternity case by case number	§II.A.10, page 35

The above are the most commonly needed commands. See also the Advanced Casework Commands (§II.B, page 37).

II.A.1. COMMAND: **delete phenotype data**

lets you remove phenotype data for some system(s) entered in error.

See also:

editing HLA data	§II.A.4.c, page 29
entering DNA probe data	§II.A.5, page 30

Note: It is also possible to delete systems using **In — enter STR or serological phenotypes** or **In DNA probe data** by using the ***Backspace*** or the ***Delete*** key.

To delete both alleles at an HLA locus using **In — enter STR or serological phenotypes**:

Delete Comma Delete

II.A.2. COMMAND: **display this case**

gives a quick display to tell you which the current case is. Names are shown and a summary PI based on calculations so far. Note that there may be some phenotypes that have been entered but not yet calculated.

```
Tested man: (T) Ali Baba     5/6/91        Case 177v
Mother:     (T) Sheherazade  5/5/91        [Turkish Immigr #1]
Child:          Philip       5/5/91        October 30, 2008
PI=51.5, PE=98.51%

Continue typing to resume COMMAND:
```

A case number is shown as 0 if the case has not yet been filed.

The case number may be followed by the letter v. That signifies that there have been some changes or additions made that have not yet been filed.

II.A.3. COMMAND: **ethnic — respecify race(s)**

lets you specify the races appropriate to the various individuals to be used in calculations. It is called automatically during initialization of a case.

A race may be a number, or a combination of numbers indicating a mixed racial background. Thus 0 represents Caucasian, 3 is Asian, and 30 means half of each (which is not precisely the same as having one parent of each race). (Don't enter 03; leading 0's will be ignored). A mixture may have up to 8 digits.

You may enter 1, 2, or 3 races, all on the same line. One race would of course apply to alleged father, mother, and random man. If you give a second number, that is for the mother; the two men will still be computed with the same race. If a third race is specified, it will apply to the random man, and a special paragraph will be added to the report explaining what calculation is represented. Thus, you can specify all three races even if they are the same in order to get that paragraph.

See also:

Preparing reports	§I.I, page 17
initialize a new case	§II.A.6, page 32

II.A.4. COMMAND: **In — enter STR or serological phenotypes**

lets you enter, or change, the genetic data associated with a case. First the case must be current, either through the previous execution of

COMMAND: initialize a new case (§II.A.6, page 32), or

COMMAND: retrieve (for a previously filed case; §II.A.10, page 35).

Then a screen is presented with three columns for the three individuals, and any previously entered phenotypes displayed. You can now change those, or add data for new systems, as explained under Entering Phenotypes.

Calculations are not done during **in**, so you will not be warned of exclusions at this time.

See also:

entering phenotypes	§II.A.4.a, page 28
entering HLA data	§II.A.4.b, page 28
editing HLA data	§II.A.4.c, page 29
entering DNA probe data	§II.A.5, page 30
initialize a new case	§II.A.6, page 32
retrieve a previous case	§II.A.10, page 35
Create genetic system	§II.C.3, page 48

II.A.4.a. Entering phenotype data

These are rules for the COMMAND: **In — enter STR or serological phenotypes**.

II.A.4.a.i.) Type ***Alt-F6*** for help.

II.A.4.a.ii.) Choose (§I.J.1, page 24) a system from the green list. ***End*** to quit. (Space is an acceptable key to confirm a selection.)

II.A.4.a.iii.) Select (§I.J.1, page 24) the two (for a motherless case) or three phenotypes from the choice list into the windows provided. Confirm the choice normally with Space; confirming with Enter terminates input for this system.

II.A.4.a.iv.) HLA A locus and B locus are entered on separate lines, A first. For each individual at each locus, you enter (one or) two genes. If two, separate them with a comma. If you try to enter three (with a second comma), you get BEEP!'ed and have to start over, which gives a convenient way to get back to the first allele if you entered the wrong one.

You can type ***Enter*** while entering A locus data to skip immediately to the B locus.

II.A.4.b. More about entering HLA phenotypes

Editing HLA phenotype data, using the **in** program, is explained in more detail as follows:

II.A.4.b.i.) For a given locus and person, the phenotype consists of two alleles separated by a comma. However, if one of the alleles is blank, you don't have to type it in, and you can omit the comma. Thus

`7,-` and `7`

are equivalent. In case of `-,-` you can also type `-`.

II.A.4.b.ii.) The order of alleles is immaterial. 7,8 and 8,7 are in every respect equivalent. You might notice, however, that the program prefers to write -,7 rather than 7,-.

II.A.4.b.iii.) You may enter **7 , 7** if you wish. This is NOT the same as 7,- or just 7; the program will assume that you somehow know there is no blank present, and compute accordingly.

II.A.4.b.iv.) Type a ***Comma*** after the first allele to proceed with entering the second allele. Use ***Space*** or ***Enter*** to leave the second allele unchanged (or to have it be a blank in case of initial data entry).

II.A.4.c. Editing HLA Phenotypes

Assume HLA data has already been entered. Changing it is similar to initial entry, using **In — enter STR or serological phenotypes**, but there are some additional possibilities:

II.A.4.c.i.) To edit just one allele:

II.A.4.c.ii.) If it is the first one, type in the new allele, then ***Space*** or ***Enter*** to keep the same second allele.

II.A.4.c.iii.) If it is the second one, ***Comma*** past the first one, then type in the new allele. Then ***Space*** or ***Enter***.

II.A.4.c.iv.) To edit both alleles:

Type in the new 1st allele ***Comma*** then the new 2nd allele.

II.A.4.c.v.) To delete just one allele, edit it to a **-** as in #2, or

II.A.4.c.vi.) if it is the 1st allele, type ***Backspace Space*** or ***Backspace Enter*** to keep the same second allele;

II.A.4.c.vii.) if it is the 2nd allele, ***Comma*** past the first one, then ***Backspace*** then ***Space*** or ***Enter***.

II.A.4.c.viii.) To delete both alleles, use the **Delete** program. Or:

Backspace Comma Backspace Space

II.A.4.c.ix.) To back up to the 1st allele:

Enter extra ***Comma***s

See also

delete §II.A.1, page 27

II.A.5. COMMAND: **In DNA — enter or edit DNA data**

(**In DNA probe data** or **In DNA** for short)

```
     probe/enzyme   woman  child  oblig  man     X       Y       1-A

[1]  pS194/D7S107   5740   5740   5740   5740   0.5     0.1766  0.32201
        Pst I       6990   6990   6990   6990

[2]  pL336/D1S47    2760   2760          3440   0.25    0.04099 0.04057
        Pst I       3210   3120   3120   3120

[3]  TBQ7/D10S28    1230   1230          1780   0.25    0.01176 0.01173
        Hae III     950    1450   1450   1450

[4]                                              0       0       0
      TO EXIT:  Esc  or  Select blank probe
      Enter data as indicated; end each field with Space, Return, Tab, or
           control left and right cursor arrows to move to another field.
      Alt-F3 & Alt-F4 open up & delete a probe's worth of data.
      Control up and down cursor arrows move to previous, next probe's data.
            pS194/D7S107
            pL336/D1S47
            pYNH24/D2S44
            TBQ7/D10S28
            pCMM101/D14S13
```

Usually DNA data is imported from DNA·VIEW. However it is also possible to add or edit DNA probe data in PATER using this facility. The X and Y probabilities must be entered by the operator, rather than being computed by the program. The command **In DNA probe data** creates a screen form with two lines per probe, much as it will be on the report. Each pair of lines is numbered (e.g. [1]).

Simplified operation:

Most commonly you will fill in the fields in order and use *Enter* at the end of each field to proceed to the next one. Choose probe and enzyme, fill in the mother's, child's, non-contributed man's genes, and X and Y. The obligatory and man's paternal gene, and the fraction of non-excluded random men, will be filled in by the program. These fields are skipped over when you use *Enter*, so you don't have to bother with them.

The probe and restriction enzyme are chosen by selection from lists, in the usual manner of list selection.

The genes should be entered as integers, representing number of base pairs.

The X, Y, and 1-A values should be decimal values from 0 to 1. Note that if X is 0, you still should enter the correct Y value. The program introduces a default Y value of 0.00001 in this case to make sure you don't put 0, but you should still supply the correct value if you know it.

The value for 1-A is computed correctly from Y in most cases. In particular, the computation is correct when there is a mother, or even in a motherless case when the child is homozygous. In those cases, the program knows that it should update any previous value of 1-A whenever Y is specified.

But in the particular situation of a motherless case and heterozygous child, the program knows that it's computation is only approximate, so it will not override any non-zero value that has already been entered for 1-A.

There are some special rules for moving about among the fields that are mentioned on an on-screen help message, and are detailed here:

If you end each field entry — confirming a menu choice, or ending a numeric entry — with ***Enter***, the program advances to the next probable field, skipping over the obligatory gene column and the 1-A (random men eligible) field.

However, there are several other acceptable keys to end an entry:

Space moves to the very next field, skipping nothing.

Esc accepts the data so far entered, and ends DNA input.

Tab skips ahead 2 or 3 fields, or cycles back to the probe field.

Ctrl left and ***right arrow***s skip several fields to the left or right.

Ctrl up and ***down arrow***s move to the first field for the previous or next probe.

Alt F3 Accept the input into this field, and open up a pair of lines for a new set of probe fields above the current one.

Alt F4 Discard all the data for the current probe; discard the lines.

See also:

in — enter STR or serological phenotypes	§II.A.4, page 28
avuncular and incest table	§IV.D.1, page 117

II.A.6. COMMAND: **Initialize a new case**

II.A.6.a. Outline

accepts names, accession numbers, lawyers' or contacts' addresses, and races for a new case.

Initialize is roughly equivalent to **People** + **Ethnic**.

The computer case # is not assigned at this stage.

The usual next step is to enter phenotype data via **In** or **In DNA**.

II.A.6.b. Procedure

II.A.6.b.i.) Enter accession numbers for mother, child and father into the box.

II.A.6.b.ii.) Enter people's names into the form. You can add addresses, which will function as explained in **People**.

II.A.6.b.iii.) Enter numeric race codes, as explained in **Ethnic**. These are the PATER calculation race codes that control serological computations and STR computations performed by PATER.

At this point the case is temporarily numbered 0, and has not been saved.

II.A.6.b.iv.) You can assign any desired number with the **File** command.

II.A.6.b.v.) Implicit saving through **Report** or when exiting PATER — will assign the next sequential case number (which is also an option with **File**).

See also:

Accession numbers	§I.J.6, page 21
Example accession entry	§I.J.6.d, page 22
PATER calculation race	§I.E.3.b, page 14
Ethnic	§II.A.3, page 27
People	§II.B.8, page 43
In — enter STR or serological phenotypes	§II.A.4, page 28
In DNA— enter or edit DNA data	§II.A.5, page 30
Preparing reports	§I.I, page 17
Report	§II.A.7, page 33
File	§II.B.5, page 39

II.A.7. COMMAND: **Report — print a report**

II.A.8. COMMAND: **Reprt — display a report**

give you a report containing a summary of all calculations for the current case (the one you have just **Retrieve**'d (q.v.), or the one for which you have just been entering data).

COMMAND: **Reprt** is an abbreviated preview version which appears on the screen only.

COMMAND: **Report** produces either or both of two versions — the internal laboratory version (page 8), and the external version (pages 7, 9). The laboratory version has more numbers, more information, and less text. The external version may be in the inclusion (page 7), the exclusion (page 9), or the "No Opinion" format.

§II.G.4.l covers user customization of **Report**, inclusion of computation races for example. (**Reprt** always shows computation races.)

Both commands automatically invoke **File** (q.v.), so creating a computer case # and updating the filing codes if necessary.

II.A.8.a. Operation of Report

II.A.8.a.i.) Saving the case (also applies to Reprt)

If the report data includes changes that have not yet been save in the PATER files, then there will be a dialogue prompting you to save, as **File current case**.

II.A.8.a.ii.) Print the "laboratory report"

```
Print the laboratory version of the report? n
```

The laboratory version (page 8) is only for your internal records. It has too much information to give to a client, and it is not language-dependent.

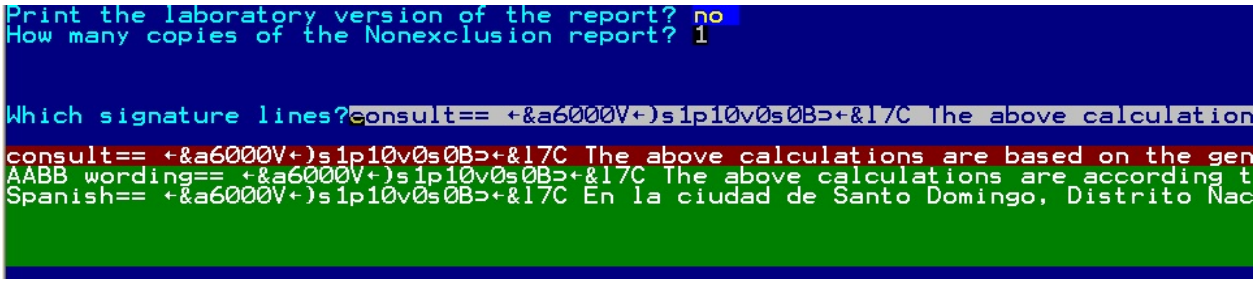
II.A.8.a.iii.) Print the "client report"

```
How many copies of the non-exclusion report? 1
```

Depending on the data and/or your choices when filing, the client report may be in any of several formats. See §I.I.4.

II.A.8.a.iv.) Selection of Logo and/or Signature line alternatives.

If you have alternative versions of either of these two parts of the report (see §II.G.4.k.i), then you will choose the desired alternative from a menu:



The most recently selected choice is listed first.

See also:

sample reports	pages 7, 8, 9
Preparing a report	§I.I, page 17
Retrieve	§II.A.10, page 35
Exclusion report	§II.A.9, page 34
File current case	§II.B.5, page 39

II.A.9. Exclusion report

The "exclusion" report is a simplified format of the external (for clients) version of the paternity report. It is the normal external version of the report when there are one or more systems with exclusions. It differs from the "inclusion" report in that it

1. omits paternity indices,
2. notes the systems with results inconsistent with paternity, and
3. contains a boilerplate explaining the opinion of non-paternity.

See page 9 for an example.

Normally the exclusion report is printed by the **Report — print a report** COMMAND when there are exclusions present. However, if desired a non-exclusion report can be prepared even when there are exclusions, or an exclusion report even when there are no exclusions, by editing the filing codes (q.v.) and overriding the default choice at the prompt *result?*

See also:

sample report	page 9
Report	§II.A.7, page 33
filing codes	§II.B.4, page 46
avuncular index	§IV.D.2, page 117

II.A.10. COMMAND: **Retrieve case #1001**

fetches the specified case from the disk, so that it becomes the current case. If the specified case doesn't exist, it will be created.

The information to be fetched comes from several places. PATER keeps complete information about each case in a file C*.SF. But there also may be relevant information in Paradox tables. The Paradox table information may be posted from various sources, including DNA·VIEW (for DNA calculations) or from an accessioning system written in Paradox. During the **Retrieve** operation, PATER offers to merge the Paradox table information into the case.

Reasons to **Retrieve** are

- » initialize a case with a specified number
- » make changes
- » prepare a report
- » or, just look at the case on the screen.

Here are the main steps in the retrieval operation

II.A.10.b. Supply case number.

Note the helpful buttons RECENT and SEARCH.

```
Retrieve case # 16079
```

The program is careful and considerate if there is unsaved work in progress on another case:

```
I have some unfiled data for


Tested man: (G) Edward W. Bicious     91-5306    9/10/91    Case 16080v
Mother:     (G) Samathana Eagle       91-5307   10/11/91    [RU88]
Child:          Fist of Steel         91-5309    9/10/91    17 NOV 1994
Shall I file it before proceeding? y
```

II.A.10.c. Display case structure, accession numbers.

```
Case 16079      race(s): c
     LA#123/Sup 99
      Mother  91-05307 c  91/10/11
    Child #1  91-05308 -  91/09/10
    Child #2  91-05309 -  91/09/10
    Child #3  91-01111 -  94/09/29
  Tested Man  91-05306 c  91/09/10
```

However, if the case number is a new number, an empty accession number block will be displayed, and you will be invited to create accession data into it exactly as with the command **Initialize**. The only difference from **Initialize** in this case is that a case number has been assigned to the new case.

II.A.10.d. Import DNA data from DNA·VIEW.

DNA·VIEW (or conceivably some other program) posts DNA results to a Paradox table, DNAAPPRO. **Retrieve** checks for relevant data.

```
 Case    |Probe |Enzyme   | Approved |Collected
16079-10|TBQ7  |Hae III  |
16079-10|EFD52 |Hae III  |
16079-20|TBQ7  |Hae III  |
16079-20|EFD52 |Hae III  |
```

Data is available as shown for two probes for each of two sub-cases corresponding to Child #1 and Child #2.

```
Include DNA·VIEW work-in-progress? y
```

By the answer **y** the DNA data will be merged into the information for this case, and will be saved into the C*.SF file when the modified case is subsequently filed.

If the case were later **Retrieve**d again, the prompts would be slightly different:

```
 Case    |Probe |Enzyme   | Approved |Collected
16079-10|TBQ7  |Hae III  |          |1994/11/17 08:45:47
16079-10|EFD52 |Hae III  |          |1994/11/17 08:45:47
16079-20|TBQ7  |Hae III  |          |1994/11/17 08:45:47
16079-20|EFD52 |Hae III  |          |1994/11/17 08:45:47

Refresh from DNA·VIEW data? n
```

The time stamps under "Collected" indicate that the data was already Retrieved once. The only reason for retrieving it again would be in case the saving of the modified case into the C*.SF file somehow failed to take place (e.g. through a computer failure).

II.A.10.e. Import other Paradox data

Names, addresses, and client reference codes can also be supplied to PATER from a program that writes them to Paradox tables. **Retrieve** checks to see if there is anything of that nature to import.

```
Retrieve addresses from accessioning? y
Update client ref code [LA#123/Sup 99] from accessioning? y
```

II.A.10.f. Select a sub-case

If there are multiple men, children, or women in the case, you must select because PATER will work only on one trio at a time. (The importing described in the previous steps does apply to all sub-cases in the case though, and they will all be updated subsequently when the case is refiled.)

```
Which child?

    Timy Vivol    Child #1
    Ehwood Vivol  Child #2
    Lani Teasdale Child #3
```

The sub-case is determined by menu selection for each role that is multiply represented.

II.A.10.g. Current case display

Retrieve puts a short report on the screen describing the current trio.

```
Tested man: (A/C)  Emmett G. Teasdale   91-5306    9/10/91  Case 16079-20v
Mother:     (A/C)  Sabrine Summer Vivol 91-5307   10/11/91  [LA#123/Sup 99]
Child:             Ehwood Vivol         91-5309    9/10/91
Filing codes: F    rN fC mC - t4 (PATER 9.16) t4 [LA#123/Sup 99]
PI=1040, PE=99.965%
```

See also:

Changing a report	§II.J.1..(2c), page 11
Initialize	§II.A.6, page 32
File Current Case	§II.B.5, page 39
C*.SF files	§II.L, page 19

II.B. ADVANCED PATERNITY CASEWORK

The following are subjects or COMMAND's pertain to the "current case," created via **Initialize**, **Retrieve**, or **Maternity**, which may not be used every day.

II.B.1. COMMAND: **Add person**

Sometimes there are extra children, possible other men, or even extra women. All the people can then be attached to the same Case Number. That way you can avoid duplicated data entry.

However, data input and reports are still based on trios.

See also

Extra people in a case	(§I.D)
retrieving from a multiple person case	(§II.A.10.f)

II.B.2. COMMAND: **Calculate current case**

tells the machine to go ahead and compute with the data you have put in so far using **In** or **In DNA**. It will tell you the overall ratio.

See also:

Entering STR or serological phenotypes	§II.A.4, page 28
Entering DNA probe data	§II.A.5, page 30

II.B.3. COMMAND: **Code — correct filing codes**

lets you modify the filing codes associated with a case. It is called automatically when a case is filed, such as when a report is printed if the case data has been altered. The screen is as below.

```
PATER "filing codes"
comment: tutorial test case

report inclusion/exclusion format
                    (test results suggest): Nonexclusion
                                      (is): Nonexclusion

NEXT>>  EDIT COMMENT  CHANGE REPORT FORMAT  ABORT
```

II.B.3.a. `comment` means the case comment. Any non-blank text is included in the paternity report just under the title line next to the case number. The EDIT COMMENT button lets you edit it.

II.B.3.b. The program guesses, usually correctly, based on the genetic analysis whether to print the `Exclusion` (page 9) or the `Nonexclusion` (page 7) format (§I.I.4) of the client/lawyer report version but you may choose to over-ride the default decision, perhaps to elect the `Other` format via the CHANGE REPORT button.

See also:

Exclusion report	§II.A.9, page 34
Report — print	§II.A.7, page 33

II.B.4. Detail of calculation

causes the computer to prepare a worksheet showing intermediate results of a paternity index calculation. This is helpful if you wish to reproduce the calculations manually, or if you want to verify that the calculation is being done correctly.

The worksheet can be directed to screen, printer, or both.

After the displaying to the screen, the machine pauses. You can then use the scrolling keys to view the entire display (since it may exceed the size of the screen).

Tabular values that are small but not zero are printed as εεεεε.

******** PLEASE NOTE ********

However, this information is NOT intended for dissemination outside of your laboratory. Please regard it as confidential, in the same way that the innards of the programs are confidential.

II.B.5. COMMAND: File current case

assigns a case # if none exists, and saves the case on the disc unless no changes have been made since a previous filing.

The **File** command gives you an opportunity to update and/or correct the filing codes before it files the case.

File will be rarely invoked because is usually called as a matter of routine by other commands, but occasionally you may want to make a quick change without printing a report, or to assign a case number out of sequence.

Example

COMMAND: **Retrieve** case # **1001**

COMMAND: **In — enter phenotypes** (and make some changes or additions)

COMMAND: **File** ... and you are given three options:

```
File this case by ...
                        using the same case number (1001)
                        letting the user pick any case number
                        Quit. Abort. Do nothing. Don't file.
```

A second example:

COMMAND: **Initialize**

COMMAND: **File** ... the options are slightly different:

```
File this case by ...
                        assigning the next sequential case number
                        letting the user pick any case number
                        Quit. Abort. Do nothing. Don't file.
```

The choice *letting the user pick any case number* includes the possibility of re-using a case number that is already in use. You will of course be warned before the save occurs in that case.

See also:

Retrieve a case	§II.A.10, page 35
In — enter phenotypes	§II.A.4, page 28

In DNA — enter or edit DNA data §II.A.5, page 30

II.B.6. COMMAND: **Immigration**

is a modified version of **Report**. The only difference is the chart of Brother (or Sister, if a maternity case) Indices, rather than Avuncular Chart, on the bottom of the laboratory version.

The Brother Index compares the Tested Man with his (untested) Brother as a possible father (as opposed to the avuncular chart, which compares the Brother with a Random Man.)

Example:

```
  Brother Indices
Degree (0=alleged father, 1=uncle, etc.) of
relation      0     1     2     3    10
vs. Brother   1     1.97  3.83  7.23 61.8
```

The "relation 1" column shows that the man is 1.97 times more plausible to have fathered this child than his brother is. The remaining columns help remind you of the meaning of the chart: the "relation 0" index is always 1 because this compares the man with himself; the "relation 10" index compares the man with a distant relative, so is nearly the same as the PI.

See also:

avuncular chart	§IV.D.1, page 117
avuncular index	§IV.D.2, page 117
motherless case	§IV.B.1, page 115

II.B.7. COMMAND: **Maternity**

allows you to create a maternity, rather than paternity, case. A "maternity" case means that the paternity of the father (if any) is assumed, and the woman is treated as an "alleged mother" to be compared to a "random woman."

You can invoke **Maternity** either before or after entering data for the case. You will be asked to choose whether to convert the present case to maternity. If that is your choice, it will however be marked as a "new case" (so that it will subsequently be filed with a new case number). Whether the present case is to be converted or a new case begun, you will be first be given an opportunity to save the present case if any unfiled changes have been made.

```
I have some unfiled data for
Tested man: (T) Ali Baba     5/6/91          Case 177v
Mother:     (T) Sheherazade  5/5/91
Child:          Phillip      5/5/91          October 1, 2008

Shall I file it before proceeding? n

Now the case is:
Father:       (T) Ali Baba     5/6/91         Case 177v
Tested woman: (T) Sheherazade  5/5/91
Child:            Phillip      5/5/91         October 7, 2008
        Choose an option, then Enter

      Start a brand new case
      Generate an additional case number with this
      data
```

If the present case is converted, then any data — name or phenotypes — already assigned to the "tested man" is assumed to apply to the father; data that had been entered for "mother" is kept for the "tested woman." That way you can easily produce both a paternity and a maternity report with the same data.

If the present case is not converted, then you will be prompted for "Father", "Woman", and "Child", initializing a new case.

Fatherless maternity cases are allowed. Just blank the father's name, or begin it with a **(**.

Note: Using the DNA·VIEW **Paternity Case** option *language is ...* to switch a case to maternity-language is likely to confuse PATER. Therefore don't use that feature. Always create maternity cases in only in PATER.

See also:

motherless	§IV.B.1, page 115
initialize	§II.A.6, page 32

II.B.8. COMMAND: **People — correct spelling**

lets you enter (or change) the name and sample id information for father, mother, and child, and to edit the addresses of the intended recipients of the report.

It also lets you change a case from motherless to with-mother or vice-versa.

The information is gathered into a screen form. Use the cursor keys (with or without ***Ctrl***) and the ***Enter*** key to maneuver around the screen.

If a particular field already contained information, it will disappear as soon as you start to type into the field.

To abort the editing process, enter a blank name for the Father.

Hit ***F5*** when you have completed editing.

Names may be entered **`Last, First`** or **`First Last`**. Use only one ***comma*** though. For example,

`Sandoval Jr, George`

and

`George Sandoval Jr.`

are acceptable.

`George Sandoval, Jr.`

will not work properly.

The first lines (names) from the addresses appear on external reports as a "cc:", and the entire addresses appear on the laboratory internal report version. The addresses can also automatically be printed onto envelopes.

See also:

motherless cases	§IV.B.1, page 115
address	§II.G.1, page 69

II.C. GENETICS

These commands deal with setting up genetic systems, allele frequency tables, and population statistics.

II.C.1. COMMAND: **Add an allele — modify genetic system**

lets you modify a genetic system previously created with **Create genetic system**.

You can add a new allele, or change the name of an existing allele.

You can only make one change per invocation of this command.

After adding an allele, it will have an allele frequency of 0. Use **Change or create allele frequencies**.

See also

In — enter STR or serological phenotypes	§II.A.4, page 28
Create genetic system	§II.C.3, page 48
Change or create frequencies	§II.C.2, page 46

II.C.2. COMMAND: **Change or create allele frequencies**

This command lets you change the frequency tables for any extant race, and any collection of genetic systems.

II.C.2.a. Race to edit?

If the race doesn't already exist, first use COMMAND: **New race**

II.C.2.b. Start from?

Copy frequencies (for systems to be specified) from another race, or start from 0.

II.C.2.c. Select systems:

as many as you like. ***Space*** after each one; ***Enter*** after the last one (or ***Backspace Enter*** when the list is complete).

II.C.2.d. Scale factor:

Choose a convenient power of 10 to avoid decimal fractions, such as 1000.

II.C.2.e. Enter frequencies

Frequencies will be presented for editing on a special screen with bracketed numbers (e.g. **[3]**) indicating the rows and columns. The row and column numbers are in turn explained by an (optional) on-screen legend. However, if you have a lot of alleles (e.g. in HLA), that legend could take up the whole screen. Therefore you are given an option to choose among:

> Just show the instructions and column/row names on the screen
> Print the instructions and column/row names. Don't waste space on the screen!
> Don't print or display. I don't need them.

The former frequencies (or zeros), scaled, are then presented on a *special editing screen* for entry or changing.

After making entries, type either

Ctrl-e to accept the changed data, or

Ctrl-q to ignore any changes and you will be presented with checktotals for the frequencies.

Normally the grand total should be about equal to the chosen scale factor.

II.C.2.f. Checktotals

Before filing the new frequency table, inspect the checktotals, which represent row and column sums, and choose from among the choices:

Edit some more - I want to change some of those numbers

if the numbers are nearly right;

Re-edit- I want to start editing with the original numbers

if the numbers are such a mess you want to try again from the beginning;

Abandon this system with no changes! Continue with the next system (if any).

to forget about changing or entering anything for this system;

Ok, those are the right numbers. I want to maybe scale, then file them.

if the changes are at least relatively (proportionately) correct. (Don't worry if you are off by a power of 10 for example. You can fix that in the next step.)

II.C.2.g. Scaling and filing

Assuming that you eventually answer *Ok* at §II.C.2.d, the program offers to scale the frequencies in up to three plausible ways.

```
Divide each frequency by what?
                                    10029; total will be 1
                                    1000; total will be 10.029
                                    10000; total will be 1.0029
```

These choices correspond to:

II.C.2.g.i.) whatever scale factor gives a total frequency of exactly 1;

II.C.2.g.ii.) the scale factor that you planned to use, at step II.C.2.e;

II.C.2.g.iii.) (if different) the power of 10 that most nearly scales the total to 1 — i.e. the scale factor that you probably did use, irrespective of your declared intent at step II.C.2.e.

See also:

editing HLA frequencies	§II.E.5.c, page 59
HLA — show complete frequency tables	§II.E.7, page 61
Define database size	§II.C.4, page 49
Document the allele frequencies in use	§II.C.5, page 52
New race	§II.F.2, page 63
choosing	§I.J.1, page 24

II.C.3. COMMAND: **Create genetic System**

lets you define a new system, for which you can then enter data using **In — enter STR or serological phenotypes** and which otherwise behaves like a codominant autosomal system with discrete alleles.

The procedure is straightforward. First you are asked for the new system name:

```
What is the name of the new STR system? THO1
```

Most of the alleles will be numbered alleles in a specified size range:

```
What is the smallest plausible allele # of THO1 ? 6
What is the largest plausible allele # of THO1 ? 12
```

but two extra catch-all alleles are added in case any larger or smaller fragments later appear. In addition, you can add or delete, one at a time, any more or fewer names that you want.

```
Defining the STR system THO1
Action?          Add an allele to those below
                 Take one away
                 Create the system
                 quit
6     7     8     9     10    11    12    <6    >12
```

II.C.3.a. *Add an allele*

```
Odd allele: 9.3
6     7     8     9     9.3   10    11    12    <6    >12
```

II.C.3.b. *Take one away*

```
Remove: <6
```

II.C.3.c. *Create the system*

The system is initially defined with frequencies of 0, so you must now use **Create or change allele frequencies** to define the frequencies.

Create genetic system is an experimental feature. It has some limitations: it can only be used to define codominant systems, and there is no good way to add additional alleles or change spellings after you have completed defining. (However, if you backed up the GENES.SF file you can discard your work and start over.) It may work differently in the future.

See also

In — enter STR or serological phenotypes	§II.A.4, page 28
Add an allele — modify genetic system	§II.C.1, page 45
Change or create frequencies	§II.C.2, page 46
Define database size	§II.C.4, page 49

II.C.4. COMMAND: **Define database size**

is an optional command. It affects the way that allele frequencies are determined.

Its main use is to assign a reasonable frequency to rare alleles, and to avoid a frequency of zero (which is interpreted as an "exclusion" by PATER when it is occurs as the frequency for the child's paternal allele, even if the tested man has it). An additional reason to use it is that it overcomes a small bias (a bias in the direction of over-stating the evidence for paternity) in the standard method of getting allele frequencies.

This command also helps you zero out some very small frequencies, that were so specified as an artifice before this command was available.

If this command is never used, or if the database size is set to "infinite", then the installed allele frequencies are used as-is.

However, if you specify a finite database size, then any allele whose frequency is desired is always "tossed in" to the database. This method is fair and accurate. In particular, it guarantees that a frequency of 0 will never occur. Also, it is convenient because, using it, it is no longer necessary to specify a non-zero value for the scores of HLA haplotypes that have never occurred.

II.C.4.a. Background

Suppose an allele or an HLA haplotype is quite rare, and was never observed in the database for a particular race, but that nonetheless it is observed in a particular case — as a shared allele between child and tested man.

If the database information is taken literally (as we used to do), then the allele is considered to be ïnfinitely rare and therefore gives an infinite paternity index.

II.C.4.b. Theory

That can't be right. One way to overcome the problem is to include the allele from the present case as if it were part of the database.

PATER has frequency tables which represent each allele frequency as a decimal number. Suppose that a particular allele Q, which occurs in the present case, occurred q times in a database of n. Then it is the number q/n that occurs in the database. After tossing in one additional Q, the frequency will be $(q+1)/(n+1)$, but there is no way to compute this from q/n alone; you also need to know n. Therefore, the **define database size** command allows you to specify the value of n for each database. This is new information that did not previously exist in PATER.

Of course n may not be known exactly for those databases that were entered long ago. However, an approximate value will do for most purposes.

For compatibility with the previous method of calculation, n needs to be infinite. Therefore, "infinite" is an allowable value for database size (sometimes typed in as `0`, which works as a convention since 0 is *not* an allowable database size). All database sizes are initially infinite, so if you never invoke this command the old performance is retained.

II.C.4.c. database sizes

To make defining database sizes more convenient, there is an "overall default size" as well as a database size that can be specified separately for every database (i.e. for every race/system combination). This information is created or modified with the **define database size** command.

The overall default can be any positive integer, or can be "infinity".

The size specified for each database can be any positive integer, or infinity, or "default" — meaning whatever the overall default is.

II.C.4.d. Operation — specify the overall default

```
The "default database size" is the presumed size of any database
for which you have not specified a particular size. "0" means infinite.

Default database size, in haploids or chromosomes (=2·# of people) 1000
```

II.C.4.e. Operation — select one or several genetic systems to work on

II.C.4.f. Operation — modify the genetic systems one at time

II.C.4.f.i.) race by race, you can set the database size moving by cursor from race to race. You may type "default", "infinity", or a number.

```
        Adjusting the database size for: Rh
Please enter the database size (in haploids) for any races below.
The database size influences frequency calculations in this way:
 If the database size is N, the minimum frequency is 1/(N+1)
Try the help screen "minimum frequency" for further explanation.
ESC when finished editing (maybe necessary several times)
```

	db size (=N) (default=1000)	# zero entries	# entries >0, <or= 1/N	total of freq <=1/N	total freq
Caucasian	default	5	0	0	1
Hispanic	default	5	0	0	1
Black	default	6	0	0	1
Asian	default	5	0	0	1
German	default	2	2	0.00046	1

Type ***esc*** when finished specifying database sizes.

II.C.4.f.ii.) Set small values to 0

The number 2 for the German race in the column `# of entries >0, <or= 1/N` means (in this case) that there are two Rh haplotypes with with frequency q such that $0<q<1/1000$. Such a frequency represents only one observation or even a fraction of an observation. Since even if the frequency value is specified as 0 in the database, the values that will be effective for casework is 1/1000, it probably makes sense to change these small values to 0.

Hence the offer (if there are any such small frequencies)

```
Shall I set to zero those frequencies that equal the minimum frequency?
```

If you give the answer **y**, the program returns to the previous step, II.C.4.f.i, giving you another chance to modify the database sizes.

II.C.4.f.iii.) Normalize the frequencies

```
Shall I normalize the total frequencies to sum to 1?
```

Removing a few small frequencies may have the effect of reducing the total frequency from 1 to a slightly smaller value. It may be preferable, instead, to distribute the deducted amounts to the remaining positive frequencies.

If you give the answer **y**, the program returns to step II.C.4.f.i.

II.C.4.f.iv.) Commit the changes to file

```
File the changes now? ('n' does not lose any changes)
```

If you give the answer **y**, the program proceeds to the next genetic system. If **n**, then it checks your intention:

```
More editing? ('n' will discard all the changes for this system)
```

If you give the answer **y**, the program returns to step II.C.4.f.i. If **n**, proceed to the next genetic system without saving any changes to this one.

II.C.5. COMMAND: **Document the allele frequencies in use**

lets you select a collection of genetic systems. The frequencies in use for calculations for those systems are presented on the screen or printed, at your option. All races are presented.

See also:

choosing tests	§I.J.1, page 24
HLA frequency tables	§II.E.5, page 58

II.D. Exact Tests for Independence

II.D.1. Discussion of Independence

Some forensic calculations — including paternity calculations — implicitly assume that alleles are randomly assorted through the population. In the absence of some mechanism to the contrary this is undoubtedly true, but it still needs to be checked.

Multiplication of paternity indices or of matching odds between loci is valid only to the extent that the loci are independent. Of course there is no particular question of genetic independence, but the issue is statistical or mathematical independence. Independence in this sense could in principle be violated by reason of non-random mating or some less likely biological causes such as disease association.

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 the observed variation to occur just because of sampling variation, even when the independence hypothesis holds?

The traditional way to answer the question was to compute a value called χ^2 which purports to measure the discrepancy between the observed data and the ideal expectation. The χ^2 theory holds that the larger the value, the less likely it is that the discrepancy is due to sampling variation. Moreover, according to the theory you can convert χ^2 to a ***p***-value — the probability, between 0 and 1, that a sample of the size chosen would have a larger χ^2 assuming independence.

Unfortunately the χ^2 theory falls down when dealing with data wherein some of the haplotypes have low expectation. In many cases a sample distribution that by any rational criterion is not particularly out of equilibrium nonetheless gives a large χ^2, and vice versa. Consequently and moreover, the usual formula for converting χ^2 to a ***p***-value doesn't work either. Nor is "pooling" of much help.

II.D.1.a. Exact test for independence of loci

There is, however, an excellent alternative approach. Instead of using the ad hod χ^2 statistic to measure the sample, there are "natural" functions that can be used. The alternative method is a so-called "exact test" as described in Zaykin, Shivotovsky, Weir (1995) Exact test for association between alleles at arbitrary numbers of loci, Genetica 96:169-178. The natural functions are chosen from one of the following:

II.D.1.a.i.) assuming that the sample will necessarily have the same allele counts as this sample does, what is the probability to observe exactly this set of (multi-locus) genotypes?

II.D.1.a.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 to observe exactly this set of multi-locus genotypes?

II.D.1.a.iii.) a hybrid — condition (1) for some loci, and condition (2) 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 (1) or (2) 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.

II.D.1.b. 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 (a) — you have the Guo & Thompson method.

Hence the PATER menu items **Independence of Loci** and **HW Exact Test** are the same program.

II.E. 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.

The program is operated through the menu shown in **5**. 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 an ASCII file.

Good for a HW test or a test of independence between loci:

3. Import data from an ASCII 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.

II.E.1. Type in phenotype counts for a Hardy-Weinberg check

Select the first menu option from **5** to enter a triangular array of numbers such as Figure 2. 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 labelled 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 2. 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 (II.E.3).

II.E.2. Monte Carlo calculation

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 2, 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.

Strike **n** to stop the Monte-Carlo run, or hit any key to interrupt it whereupon you are asked Resume Monte-Carlo run? and you may type **n** to return to the menu of **5**.

II.E.3. Ascii export, or import a triangle of phenotype counts

II.E.3.a. *Export*

prompts you for a DOS file name to which to save an ASCII text file of the report buffer, just as the **ASCII** command from the main menu.

II.E.3.b. *Import*

allows you to retrieve to rework a triangle of numbers for a HW check that were previously saved in a named ASCII 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.

II.E.4. Import columnar genotype data

is the way to input data for a test of independence of loci.

```
DONOR NoVWA VWA THO1THO1F13 F13 FES FES     <<< first line can be titles
                                            <<< blank lines are ignored
  1     14  17  7   9   5   7   10  11      <<< genotypes for person 1
  3     15  19  7   8   5   15  10  11      <<< genotypes for person 2
  41    14  18  6   7   7   7   10  10
  43    18  19 9.3 9.3  7   7   10  11
```

Figure 8 Genotype file format

Using this option, you specify an ascii file containing the genotypes for a list of people in columnar form as shown in **Figure 8**.

II.E.4.a. Input file format

The format rules are rather permissive:

i. You can use any name (not containing spaces) you like for the alleles, and not just numbers.

ii. The columns of data are separated by one or more columns of spaces.

iii. The first row is interpreted as titles, and doesn't have to have spaces indicating column separation. Blank rows will be ignored.

iv. Each person is represented by one row.

v. Each locus is represented by a pair of consecutive columns.

vi. Extra columns — e.g. for sample identification — won't hurt because you will always specify which columns you want to work with.

vii. If the input includes the ***tab*** character, it will be resolved into spaces on the basis of tab stops every 8th character position.

II.E.4.b. Selecting columns

A few sample rows of the input file are displayed on the screen with different colors representing the different columns. Each column is also labelled with a small letter at the top: ***a, b, ...*** To specify the columns to work with, answer the prompt by giving the letters for both columns of all the loci of interest. Thus you give four letters, or six, etc. (If you are choosing data for a Hardy-Weinberg analysis, then just give two letters.)

In the case that you choose only 1 locus, 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 section II.E.1 if you wish.

II.E.4.c. Specify kind of test

There are two possible ways to consider each locus, as discussed in the Genetica paper:

i. *as a collection of alleles* — allelic shuffling, discussed in II.D.1.a(1). 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.

   This method is chosen automatically if only one locus is selected under II.E.4.b.

ii. *as a collection of genotypes* — phenotypic shuffling, discussed in II.D.1.a(2). 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.

II.E.4.d. Summary statistics

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:

II.E.4.d.i.) observed allelic associations (e.g. # people with A1 and B2)

II.E.4.d.ii.) expected numbers of each allelic association

II.E.4.d.iii.) excess of observed over expected = observed - expected

II.E.4.d.iv.) relative excess = (observed-expected) / expected

II.E.4.d.v.) approximate variation from expected, in standard deviations

= (observed-expected)^2 / max(expected,2)

This tables are only offered in the case of comparing just two loci due to the difficulty of presenting 3-dimensional 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. Normally table (v) is the most significant — particularly large values in that table are most likely to mean trouble.

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.

II.E.5. HLA frequency tables

II.E.5.a. HLA frequency commands

The HLA data that you edit is the complete, reference frequency tables. The frequencies used by the calculational programs are a (usually) condensed version. The condensing depends on which splits you call, and is specified to the computer via

COMMAND: **HLA — specify splits** (§II.E.6, page 61)

The complete tables can be displayed with

COMMAND: **HLA — show complete frequency tables** (§II.E.7, page 61)

or the condensed ones with

COMMAND: **Document the allele frequencies in use** (§II.C.5, page 52)

The tables may be changed with

COMMAND: **Change or create allele frequencies** (§II.C.2, page 46)

and the changes will automatically be reflected in the condensed tables.

II.E.5.b. HLA Frequencies — Discussion

The originally intended use of the HLA frequency mechanism in the program is that you should specify the splits that your laboratory actually calls, because only in this way can the power of exclusion be calculated correctly. Then if you decide NOT to call a certain split for an exceptional case, you redefine the split table temporarily.

In order for this mechanism to work correctly, the frequencies should be entered for the split antigens, and entered as 0 for the corresponding broad antigen.

If you do this, the frequencies should sum to 1 for All antigens or for Narrow Specificities, but not for "Broad Antigens."

However, if you are not concerned about power of exclusion (or incest index, which is also affected), you can enter frequencies for all antigens, broad as well as narrow, and set up the split table so that there are not splits — every antigen just means itself. This is simpler in a way, because you will never have to change the split table for case work.

If the frequencies are set up in this way, they will sum to 1 for "Narrow specificities" and for "Broad Antigens" (assuming the splits are temporarily defined realistically), but never for "All antigens."

See also:

check totals	§II.C.2.f, page 46
splits in HLA	§II.E.6, page 61
Change or create frequencies	§II.C.2, page 46
about HLA frequencies	§II.E.5, page 58

II.E.5.c. Editing HLA Frequencies — Procedure

The command **Change or create allele frequencies** is used to edit frequency tables for other STR or serological systems as well as HLA. The command offers a number of options designed to make entering a table of frequencies more convenient. These options are particularly useful in the case of HLA tables because HLA tables are so large.

Suppose for example that you want to enter new HLA frequencies for the race Caucasian using the tables published by Terasaki. Here are the recommended steps.

II.E.5.c.i.) Create a temporary fictitious race as a repository for work-in-progress.

COMMAND: **New race**

Race you are replacing? **other**

(New) name for race: **Temp**

II.E.5.c.ii.) Create frequencies for the ficticious race

COMMAND: **Change or create frequencies**

Change frequencies for race: **Temp**

start editing from? **zero**

Specify test(s) **HLA** ***Enter***

Scale factor for Temp HLA frequencies? **100000**

The Terasaki HLA tables are published with a scale factor of 1000, so that is another logical choice. 100000 saves you entering any decimal points.

Edit which A locus antigens? **All**

Edit which B locus antigens? **All**

Enter data into the special editing portion of the screen as detailed in §II.C.2.e.

The program will probably make scaling offers as described in §II.C.2.g. Choose the exact power of 10 scaling factor (100000), resulting in a non-round total. Otherwise, all the numbers that you have entered will be slightly adjusted and you will not recognize them in the future!

II.E.5.c.iii.) Proof the results as follows:

COMMAND: **Specify HLA splits**

Define the splits in HLA the same way they are defined by the source tables that you will be typing in. The (only) reason for this is that the terms **Narrow specificities** in step 3d and **Broad antigens** mentioned in step 3b are relative to the definitions you create here.

COMMAND: **Create or change frequencies** ...

Edit which A locus antigens? **All**

Edit which B locus antigens? **Broad antigens**

Then exit from the editing screen with ***Ctrl-e*** or ***Ctrl-q***, and compare the checktotals for the A locus with Terasaki's. Note any major discrepancies (more than 0.30 parts in 1000), perhaps using the B locus sums as a very rough aid to help find the positions in error — and correct them (**Edit**).

Re-edit, but this time with **Broad** A Locus antigens and **All** B locus antigens.

As a further check, select **Narrow specificities** for both loci, and look at the checktotals. They should be close, but a bit low. The reason is that some people have B5 but neither B51 nor B52, for example. Save the accurate, proofread frequency table.

II.E.5.c.iv.) [optional, but theoretically preferable] Edit again, this time selecting **All** antigens at both loci. Change to 0 all those broad antigens for which there is split data as well — e.g. set A9 and B5 to 0, but do not set A2 or blank frequencies to 0. The result should be the same checktotals as in step 3d, and the overall total will be close to 1.

» If you OMIT this step, so that frequencies do not sum to 1, here are the consequences:

i. The program's calculations of Random Men Eligible and Power of Exclusion will not be reliable because the program will overestimate the chance of homozygosity when apparent blanks are involved.

ii. The calculations of incest indices will not be accurate.

Those are not crucial numbers, however.

iii. For casework calculation you must define the splits in HLA so that everything is a narrow specificity. (Otherwise the effective frequency for antigens that have splits will be about twice too high.) This could be a convenience — see 4b.

» If you INCLUDE this step, then for casework calculations you must define the splits in HLA to reflect your actual laboratory practice. If you vary the practice for a single case, then change the splits table, calculate the case, then change the table back.

In any event, the internal laboratory version of the case reports will contain a note that mentions any relevant splitting assumptions that affect the calculation of that case.

II.E.5.c.v.) Install the new frequencies:

COMMAND: **Change or create frequencies**

Change frequencies for race: **Caucasian**

start editing from? **Temp**

Specify test(s) **HLA** *Enter*

Then get an editing screen, ***Ctrl-q***, and accept the Checktotals via Ok.

II.E.5.c.vi.) Use

COMMAND: **Specify HLA splits**

to create the correct definition of splits for casework — note the remarks above in step 4.

See also:

check totals	§II.C.2.f, page 46
splits in HLA	§II.E.6, page 61
Change or create frequencies	§II.C.2, page 46
about HLA frequencies	§II.E.5, page 58

II.E.6. COMMAND: **HLA — specify splits**

This command enables you to specify which splits you can call in HLA. You will be asked to create two tables, one for the A-locus and one for the B-locus. Then the computer will create a condensed version of the HLA frequency tables according to your specifications.

Example: Suppose you have entered A25 and A26 data (via change frequencies), and have entered A10 frequencies as 0. If you map each allele to itself:

A10 **A10**

A25 **A25**

A26 **A26**,

then the splits A25 and A26 will be available to you. (Using A10 would cause an exclusion, since it "doesn't exist".)

To unsplit, don't change the frequency tables. Just re-**HLA — specify splits**:

A10 **A10**

A25 **A10**

A26 **A10**.

Now A10 will have the combined frequencies for A25 and A26, and if it is used for a case the notation `A10=(10,25,26)` will appear at the bottom of the printed report. (A25 and A26 become invalid responses to **In — enter STR or serological phenotypes**.)

See also:

Change or create allele frequencies §II.C.2, page 46

II.E.7. COMMAND: **HLA — show complete frequency tables**

This command lists the HLA reference data for a single race at a time. The output can be directed to the screen or printer at your option.

See also:

HLA frequency tables §II.E.5, page 58

Document frequencies in use §II.C.5, page 52

II.E.8. COMMAND: **Recombination % in HLA is calculated at 0.01**

shows the current value assumed for the recombination rate between the A and B loci in HLA (as a fraction, NOT as a %).

You can change it by typing in the new value. The changed value will be saved on disk, and will apply until you change it again.

The HLA calculation is always done twice — once with recombination at 0, and once with recombination at either 0.008 or whatever value you have specified, if not 0.

The result based on your specified value appears in the report. The result of the alternate calculation is shown on the screen, and is mentioned on the bottom of the laboratory report.

II.F. SUMMARY

These COMMAND's pertain to the multiplicity of cases, as opposed to a single case:

II.F.1. COMMAND: **Avuncular from case #... to case # ...**

summarizes the specified case range with regard to avuncular indices. A double column report is printed, 110 cases to a page. Cases are sorted according to the ratio PI/AI (so exclusion cases are shown at the end). Given equal PI/AI, cases are sorted by AI. Cases with HLA not tested are omitted.

Example:

CASE	PI	PI/AI	AI
172	555	9.4	59.3
136	96.0	4.7	20.6
135	91.7	3.8	27.1
171			15.4
142			3.77
147			1.86

See also:

what is an avuncular index? §IV.D.2, page 117

interpreting the avuncular index §IV.D.3, page 117

II.F.2. COMMAND: **New race — create or change race name**

This command lets you create a new race, or change the spelling of an old one.

Frequencies are not automatically created; they still have to be added via the command **Change or create allele frequencies** (q.v.).

See also:

change frequencies §II.C.2, page 46

II.F.3. COMMAND: **Summary, a box per case**

prints a very condensed summary of specified information for a selected range of cases.

First you specify case numbers to include, e.g.

summarize from case # **130** to **145**

The data you can choose to include for each case includes:

Case # and filing codes

Some or all of the names and/or sample numbers.

Phenotype data for specified STR or serological tests and individuals. (DNA data imported from DNA·VIEW, or created with In DNA, cannot be included).

For each case a small summary (a few inches by a few inches) will be prepared. These will be printed in multi-column format.

Example:

```
139. CWISC rE fC mC  *m
t2
 Daddio
 (none)
 Leetle Feetle
              Mo   Ba   Fa

 ABO          AB   B
 Rh
 A locus      3,11 1,2
 B locus      -,7  5,7
```

```
140. CUTAH rN fC mC *m t0 /139 [broth
 Sam Rayburn
 Clara Burton
 Fitzgerald A. Burton
         Mo  Ba  Fa
 ABO     B   B   AB
 Rh
 A locus 1,2 1,2 3,11
 B locus 5,7 5,7 -,7
```

```
141. F NIX rE fP mC  *f*m
t9 [Hawaiian "
 William Payton Hough
 Kimberly A Williams
 Nicholas Hough
         Mo   Ba   Fa

 ABO     O    O    O
 Rh      DCce DCce DCce
 A locus 2,11 -,11 -,9
 B locus 7,40 -,40 35,12
```

```
142. F NIX rN fH mC *f*m t16
[Original b
 Benjamin V. Hough
 Kimberly A Williams
 Nicholas Hough
         Mo   Ba   Fa
 ABO     O    O    A2
 Rh      DCce DCce DCe
 A locus 2,11 -,11 11,29
 B locus 7,40 -,40 8,40
```

See also:

filing codes §II.B.4, page 46

II.F.4. COMMAND: **Summary, one line per case**

The intent of this report is a marketing tool summarizing the case work that you have done per client (e.g. per county).

It is a 1 or 2 line per case summary of the specified cases based on the filing codes.

You are asked to select a range of cases for the report:

from case # **140** to # **180**.

The report shows the filing codes, paternity index, and exclusions.

The cases are sorted according to source (columns 2-5 of the filing code). The listing begins with a new page for each source. You are given the opportunity to select a subset of the pages to print, or to print them all.

Within each each source the report is sorted by paternity index.

Example:

```
                        4 Paternity Cases Performed for RIV
Case Tested                Prob   Classification
     man                          codes

172. Bicious, Edward       0.998  F RIV rN fH mH -  t8 /171 (PATER 3.1)
137. Juno the Magnificent  0.996  F RIV rE fH mH -  t6 (PATER 3.0)
171. Bicious, Lowell       0      F RIV rE fH mH -  t13 [ref: AG9876] (PATER 3.1)

       Exclusions:  MNSs, Gc, pL336/D1S47, YNZ22/D17S5

147. Alfredo Gonzales      0      F RIV rE fH mC -  t3 (PATER 3.1)
       Exclusions:  ABO, Rh, MNSs, Kell, Gc, HLA
```

See also:

filing codes §II.B.3, page 38

II.F.5. COMMAND: **Cull phenotype frequencies from cases**

For specified range of cases and any collection of STR or serological tests (but excluding HLA and DNA types imported from DNA·VIEW or created with In DNA), compute and list by race, for each specified race code, the number occurrences of each phenotype.

Data for children is not used.

Data for extra men or women is not used (i.e. only the first of several).

Data for father or mother corresponding to *f or *m in the filing code is not used.

The race codes used are the ones provided in the filing codes.

See also:

filing code description §II.B.4, page 46

II.G. MISCELLANEOUS

These COMMAND's have little in common.

Address	laserjet envelope addresser	§II.G.1, page 69
Export	ascii file of case & accession	
Flush network buffer		
Manual	print out help screens	§II.G.2, page 69
Noise	adjust victory music length	§II.G.3, page 69
Options	printer, titles, menu style	§II.G.4, page 70
Play music	Bach, Kern, Mozart	§II.G.5, page 78
Printer setup	margins, type size	§II.G.7, page 79
Update	adapt to new program version	§II.G.8, page 81

II.G.1. COMMAND: **Address**

The **Address** command addresses envelopes. It will prompts you for case numbers. For each case, it prints one envelope for each address found, the addresses having been supplied on the **Initialize** screen, or changed using **People**. There is a pause before each address, in case you have to load the printer.

After each case is processed, the next sequential case is suggested as a default. That way, you can print lots of envelopes by just pressing ***Enter*** repeatedly.

Before using **Address**, you may want to invoke **Printer setup** to adjust the left and top margin settings.

See also:

Printer setup	§II.G.7, page 79
People	§II.B.8, page 43
Initialize	§II.A.6, page 32

II.G.2. COMMAND: **Manual**

lets you print out the Help screens. The **Manual** menu gives you these options:

1. New screens. You give a date. Any screen added or changed after that date will be printed. This is especially useful to get information newer than this printed manual when you receive a new version of PATER.

   The date can be Y M D, like **`1995 6 14`**, or shorter, like **`1`** to print everything newer than the year 1 (i.e. everything).

2. Specific screens. You give the screen numbers. In order to know the screen numbers, it is a good idea first to print out:

3. The directory page (which is screen #1).

The printed manual will have copious cross references, with page numbers.

The page numbering is historical — you might want to reorder the pages into a logical order.

See also:

Printer setup	§II.G.7, page 79

II.G.3. COMMAND: **Noise**

The program normally plays a song fragment after making a paternity index computation. The length of the selection can be adjusted.

Quiet = minimum music

Medium = short music

Long = possibly the whole song

Whatever selection you make stays in effect until you change it.

II.G.4. COMMAND: **Options**

This command lets you modify a few configuration options. Select the option you want to change from the menu shown in **Figure 9**. The possibilities include:

II.G.4.a. Box drawing characters (recent)

Three alternative drawing choices are available for box borders (e.g. when PATER puts a box around a result:

II.G.4.a.i.) 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:

II.G.4.a.ii.) Symbols **+ - | >** as an approximate way to draw box borders, or

II.G.4.a.iii.) (none) – leave the borders off.

II.G.4.b. Command Menu Levels

II.G.4.a Box drawing characters
II.G.4.b Command menu style
II.G.4.c Date order
II.G.4.d Decimal symbol
II.G.4.e Locus name style
II.G.4.f Name tag style
II.G.4.g PATER printer
II.G.4.h PATER output to: [LPT1 etc.]
II.G.4.h PATER report font, margins
II.G.4.j PATER logo (heading)
II.G.4.k PATER signature line
II.G.4.l PATER client/lab report columns
II.G.4.m PATER report, sort alleles?
II.G.4.n Keyboard

Figure 9 Options

You may request that the main Command Menu be displayed as one flat list, or in a hierarchical form.

The 1 level menu lists all thirty plus PATER commands. This is probably quicker for experienced userd.

The 2 level menu lists the half dozen major categories at the top level; after you select one of those you would be given a second menu containing the commands in the selected category.

II.G.4.c. Date order: 3/18/2011 (recent – 2007)

Nominate whether dates are displayed American (month-day-year), European (day-month-year), or Asian (year-month-day) style.

II.G.4.d. Decimal symbol: . (dot) (recent – 2007)

For international compatibility, you may select dot or comma.

II.G.4.e. Locus name style like: D16S539 STR (recent – 2007)

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.

II.G.4.f. 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. To change the accession date, you must (temporarily if you like) change this to DATE. Then, you must open the case *in DNA·VIEW*, edit people, and ***space*** until you get to the date fields.

II.G.4.g. PATER Printer

PATER needs a printer mainly for the parentage report. PATER acknowledges the printer options of **Figure 9**. Except for **(none)**, whether and how the report is printed also depends on the PATER output (§II.G.4.h) choice.

II.G.4.g.i.) **Laser** refers to a laserjet printer, and is the preferred choice because it is the best supported. This choice will also work for a **deskjet printer**. If you use this choice, printer setup commands are sent to the printer each time you invoke PATER. Therefore you would not usually need to use the **Printer Setup** command. Also for this reason the program will "hang" (for 60 seconds) on startup if the printer is not attached to the computer.

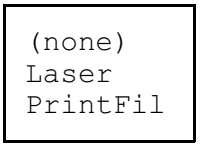


Figure 10 Printers

II.G.4.g.ii.) **(none)** will create an text report with no printer control codes presented on-screen in a sort of print-preview window, from where you can save it as a file.

II.G.4.g.iii.) **PrintFil** is experimental and probably not necessary. Even if using the **PrintFil** software for printing, recommended procedure:

» Assume the **PrintFil** software is installed and configured to (a) capture LPT1, (b) print to some Laserjet -compatible printer

» Set PATER Printer: Laser (!)

» Set PATER output to: LPT1

» If you wish to customize the PATER logo (§II.G.4.j) appearance re font size etc., do so using the HP PCL language per §II.G.4.j.ii).

II.G.4.h. PATER output to: [LPT1 etc.]

Lets you select output destination from LPT1, LPT2, screen, file, etc; allowable width in which to print. For details see §II.G.7.a.

II.G.4.i. PATER report font, margins

Same as §II.G.7.c.

II.G.4.j. PATER Logo

The PATER Logo means the title lines at the top of a paternity report. You can customize them using this option. If you want you can have multiple different versions of the Logo. See §II.G.4.k.i for the method.

You will get a special editing screen with these instructions at the top:

```
Full Screen Alpha Editing                    (Ctrl-h for complete editing rules)
Ctrl-e when done to save the edited result, or Ctrl-q to abandon changes.
Surrogates: { and } can be used to enter Esc and Bell
```

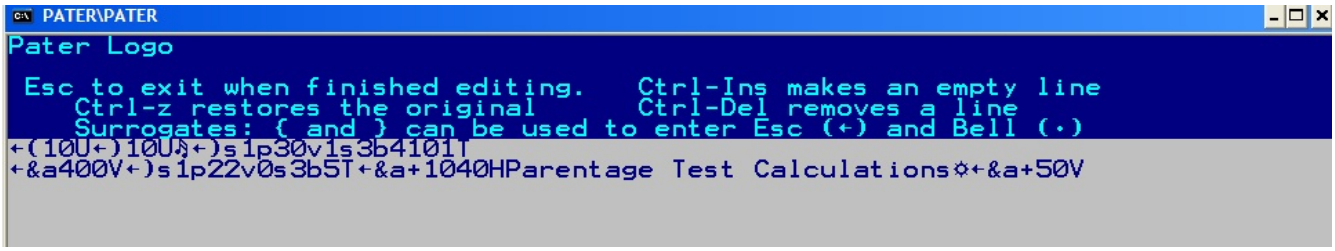
followed by the current text of the logo which you can modify:

```
PATERNITY REPORT
Dr. Cary Middlecoff, Director
(510)644-1112
```

If you would like some of the text lines to be in a large or special typeface, you can insert the appropriate printer control codes as described in your printer manual. Using an HP-compatible printer, use "secondary font" for the logo, then revert to "primary font" at the end.

The easiest way to achieve a balanced centering of lines in that case is to put a horizontal positioning code at the beginning of each line of the logo. Thus you defeat the program's futile attempt to center the logo by counting characters — futile because the program is not sophisticated enough to consider that characters may have different widths.

II.G.4.j.i.) Example –



creates approximately the appearance:

Parentage Test Calculations

II.G.4.j.ii.) Some remarks about Hewlett Packard Printer Control Language (PCL)

Reference: # HP PCL/PJL Reference (PCL 5 Printer Language) - Technical Reference Manual Part II (bpl13211)394 pages, 3.8MB, from http://h20000.www2.hp.com/bc/docs/support/SupportManual/bpl13211/bpl13211.pdf)

PCL is recognized by HP and many other printers. It is a system of control phrases that select fonts, page positioning, etc.

PCL phrases begin with an Escape character (denoted ←) and end with a capital letter. There are also a few single-character controls.

←(10U	selects PC-8 as the code page for the primary font. “Code page” is an assignment of characters to the 256 ascii positions. Some others are: 8U for Roman-8, 1E for UK, 0S for Swedish, 1G for German, 5S for Portuguese.
←)10U	selects PC-8 as the code page for the secondary font.
♫	shift out to (start using) secondary font
←)s1p30v1s3b4101T	secondary font proportional, 30 points, italic, medium weight, CG Times
←&a400V	absolute vertical position, 400 decipoints (400/720 inches) from the top of the “logical page”
←)s1p22v0s3b5T	secondary font proportional, 22 points, upright, medium weight, ?font
←&a+1040H	relative horizontal position, 1040 decipoints right of current position
☼	shift in to (start using) primary font
←&a+50V	relative vertical position, down 50 decipoints from current position

HP PCL5 Font Strings for Non-Fixed & Fixed Pitch Fonts		
Category	Description	ASCII (dec)
Prefix to setting font	primary font	←(s
	secondary font	←)s
Spacing Variations	Fixed spacing	0p
	Variable spacing	1p

Point Size Variations (for variable pitch fonts)	6 point	6v
	8 point	8v
	9 point	9v
	10 point	10v
	12 point	12v
Primary Pitch Variations (for fixed pitch fonts)	10.0 CPI	10.0h
	12.0 CPI	12.0h
	13.3 CPI	13.3h
	15.0 CPI	15.0h
	16.0 CPI	16.0h
	17.1 CPI	17.1h
	20.0 CPI	20.0h
Primary Style Variations	Solid Upright	0s
	Italic	1s
Stroke Weight Variations	Medium (book or text)	0b
	Semi Bold	1b
	Bold	3b
	Semi Light	-1b
Typeface Family Variations (variable pitch)	Albertus	4362T
	Antique Olive	4168T
	Arial	16602T
	CG Times	4101T
	CG Omega	4113T
	Clarendon Condensed	4140T
	Coronet	4116T
	Garamond	4197T
	Marigold	4297T
	Times New Roman	16901T
	Univers	4148T
Typeface Family Variations (fixed pitch)	LinePrinter	0T
	Courier	4099T
	CourierPS (?)	24579T
	Letter Gothic	4102T

II.G.4.k. PATER signature line

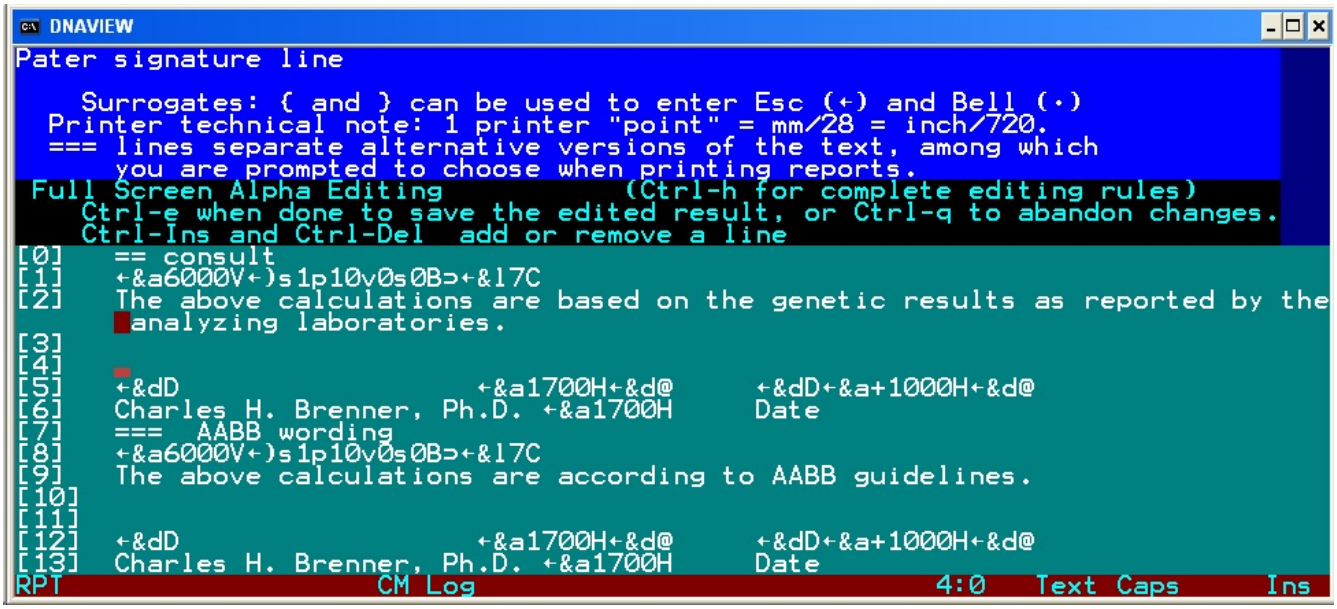
The PATER signature line is the boilerplate at the bottom of the paternity report. Changing it is similar to changing the logo (II.G.4.j).

II.G.4.k.i.) Alternative versions (applies to either signature line or Logo)

New feature for reports in various languages etc

If you have more than one then each time you print a report you will select the desired version from a menu.

» Separate the various versions with a line beginning **===** (at least two = signs). Note that you can also include a comment on the separator line if you wish.



Each alternative has to have all necessary printer codes. (See the discussion at §II.G.4.j.ii.)

Copy/Paste. Therefore it is important to know how to copy/paste! That depends on which editing screen the program uses. Assuming the screen shown in the above figure, the method is

Move the cursor to the first line to copy. ***Ctrl-t*** (t="tag")

Arrow to the last line to copy. ***Ctrl-t*** again.

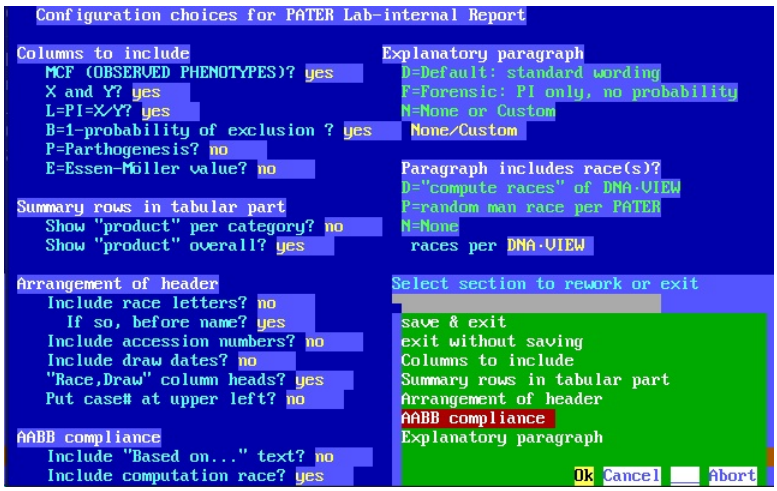
Arrow to the line above where you want to put the copy. ***Ctrl-c*** to paste a copy. (Or ***ctrl-m*** would *move* instead.)

In any case, the screen tells you how to find the *help* instructions.

II.G.4.l. PATER client/lab report customization

These options allow you to customize the printed paternity reports. The customization includes more than just specifying which columns should be included so they are slightly misnamed. You customize by answering a series of short answer questions.

You can ***enter*** through the many options. ***Enter*** all the way to the end to save.



If you went too fast or want to go back and change use the menu in the corner to select the paragraph at which to begin.

II.G.4.l.i.) Columns to include

Choose desired columns to comprise the tabular part of the report, e.g. `MCF, X and Y, L` for the genetic types and paternity index calculation.

All the computational columns are omitted from an exclusion report.

» B=1-probability of exclusion? **n**

This statistic (=1-A, A=probability to exclude a random man) has no logical purpose because L is the correct statistic. But it is requested by some courts.

B, rather than the more commonly cited A, is presented in the tabular form because you can multiply several B's together to get an overall B. The exclusion probability A is given in the summary, however (provided this column is requested).

» Include P=Parthenogenesis? **n**

This statistic has no independent interest. It is only calculated as a step in determining the incest indices that are given at the bottom of the laboratory report.

» Include E=Essen-Möller value? **n**

This is another statistic of historical interest only. The definition is

$$E=10-\log(X/Y).$$

It was formerly used in Germany as a step in hand-computation of W.

II.G.4.l.ii.) Show "product" per category, overall?

Should partial or overall products be included in the tabular part? Probably not. If DNA is the only category (i.e. no serological systems; see §II.H.1.d), then there is no point in product/category. The overall product will be mentioned in the summary anyway.

II.G.4.l.iii.) Arrangement of header at the top of the client-format report.

Include race letters? If so, before name? ... accession numbers? draw dates? "Race,Draw" column heads?

Customize the name rows at the top of the report. “Column head” means for example that the word “race” appears above the column (if present) with letter codes for races.

II.G.4.l.iv.) AABB compliance

Include "Based on..." text? **yes** means *exclusion* reports should include a sentence listing the loci that evidence parentage inconsistency (see page 9).

Include computation race? **yes** to include in (normally) the laboratory report version (page 8) the letter code for the population ("race") that DNA·VIEW used for calculation of each DNA locus.

Note that the on-screen Reprt (§II.A.8) always includes the computation race.

II.G.4.l.v.) Include explanatory paragraph?

The default “explanatory paragraph”, is a paragraph at the end of the report similar to:

> The observed combination of genetic markers of the involved parties is at least 17000 times more characteristic of paternity by Reverend Dimmesdale than of paternity by an untested, unrelated man.

None or custom if you have a custom wording in order to obtain your laboratory-specific explanation.

II.G.4.l.vi.) Choices re race names in the “explanatory paragraph” at the bottom of the report

» DNA·VIEW (D). This choice means that the one or more “compute races” associated with the case will all be listed as the population considered for the random man. The race spellings

– the words associated with the race letters – are set in DNA·VIEW using **Housekeeping**, Maintenance, race coding (change).

» PATER (P). This choice means that just one race will be listed. That race is normally the race actually used for the computation – C for the example above.

However it is possible to change it. Retrieve the case in PATER, then from **Casework** choose Ethnic. There you can specify a number for the "random man" race.

The race spellings – the words associated with the PATER race number – are set in PATER. In the **Summary** menu choose New Race. The races in the race menu are numbered 0, 1, ... Choose the race of interest and you can type a new spelling, then hit ***enter.***

II.G.4.m. PATER report, sort alleles?

This option controls the order in which the DNA alleles of a heterozygous person are printed on the report.

The standard order (if you elect not to sort), is determined by the order in which you typed them in with **in DNA**, or by the order that DNA·VIEW prefers (which generally puts the child alleles in the order maternal, paternal, and matches up the parental orders to correspond).

If you elect to "sort" the alleles, you can further elect whether they will be in numerically increasing, or decreasing order.

II.G.4.n. 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).

II.G.4.o. Language

You can select among English, German, Spanish or Portuguese — other languages on request — for the paternity report. Some of the menus, explanations, and report paragraphs also have multi-lingual options. Choose which language you prefer.

II.G.4.p. Numlock on/off

Reverse the default state of the "numlock" key.

II.G.4.q. PATER character translation

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. However, most of the display symbols are available in the printer symbol set, but are merely in different positions. 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 are the ones that might change. 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 II.G.4.o). If a character is currently listed in the table as untranslated, the likely translation will be suggested as a default.

Character translation will be used for example in conjunction with the HP Roman 8 symbol set (§II.G.7.c.ii), page 80).

II.G.5. COMMAND: **Play music**

plays music selections.

II.G.6. COMMAND: **Search for a name from case # ... to case # ...**

This command is to help you find the case number when you only know the name (or part of the spelling of the name). You give a range of case numbers to search within, and the letters in the name to search on. The program displays the case # and names when it finds a match, and volunteers to continue the search in case that isn't the case you were looking for. It is rather slow.

II.G.7. COMMAND: **Printer setup**

This command accesses these four options governing report output:

II.G.7.a	Output to printer, screen, or file	Druckerausgabe auf Bildschirm, oder Datei
II.G.7.b	Select printer type, report titles, etc	Wählen von Druckertyp, Überschriften, usw
II.G.7.c	Printer setup (font, margins)	Drucker einrichten (Schrifttyp, Ränder)
II.G.7.d	Specify character substitutions	Ersatzzeichen definieren

II.G.7.a. Output destination

Output to printer (& line widths), to screen, or to file

Druckerausgabe (& Zeilenlänge), auf Bildschirm, oder Datei

Menu selection from the options shown at the right control the destination for future output from other commands. Output that normally goes to a printer can alternatively be sent to the screen or to a designated file.

Output that normally goes to the screen — operator messages, **Display**, **Reprt** — is not affected.

II.G.7.a.i.) *local printer* gives the further choice of specifying how wide to make the title and free-text regions of the paternity reports

```
Number of print columns for Inclusion report text: 80
Number of print columns for Exclusion report text: 80
Print column width in which to center report title: 75
Number of print columns for Laboratory report text: 83
```

II.G.7.a.ii.) Output to file (or to file; *convert spaces to tab*)

You may specify any DOS filename, including a "qualified" name (i.e. in some other drive or directory). Output from **Report** and most programs will spool to that file until you again invoke **Printer Setup**, or leave PATER.

The most common use of this feature, especially using *convert spaces to tab*, is to import the Paternity Report into a word processor so that you can tailor it to your preference.

II.G.7.a.iii.) Output to numbered files

If you want to make one DOS file per case, there is a special way to do it. Give a filename consisting only of numbers. Then PATER will change the number to the number of the current report. This means, if you create several reports for different case numbers, PATER will automatically make a different numbered file for each one.

The file name must be just digits, with no extension — i.e. such as **`12345`** but not **`12345.rpt`**. There can also be a path or drive — **`A:\reports\12345`** counts as "only numbers" for this purpose. Note also that the file name you specify might not be used at all. It's really just a template to indicate that the "numbered file" convention should be adopted.

II.G.7.b. Select printer etc.

This menu choice from **Printer Setup** is identical to the main menu choice **Options**.

Select printer type, report titles, etc (="Options" in main menu)

Wählen von Druckertyp, Überschriften, usw (="Options" im Hauptmenü)

II.G.7.c. Printer setup

Printer setup (font, margins)

Drucker einrichten (Schrifttyp, Ränder)

Depending on the type of printer you have, there are various ways you can control the appearance of the paternity report. Whatever configuration you select will be filed, and thus presented to you as the default next time you invoke this program. For a laserjet, a sample choice dialogue is:

```
LASERJET setup
    Page orientation 11.5 inch long paper (U.S. portrait)[1]
    Type style Courier elite (10 point=12 characters/inch)
    Vertical lines/inch (proportional=7.2) 6.5
   Note: margin settings may be padded by printer behavior
    Left margin (inches, usu 0.5-1.5) 0.7
    Top margin (inches, usu 0-2) 0.4
    Number of copies 1
    Symbol set HP Roman 8 (included all accented chars, diff ASCII codes)
```

"Symbol set" means the correspondence between characters and ascii code. Your choice of symbol set pertains to the primary font, which is always used for the tabular part of the paternity. If you choose to select the secondary font for the title and/or signature sections of the paternity reports, you can insert secondary symbol set code if desired. The choices are

II.G.7.c.i.) PC-8 (ASCII codes same as display; with line draw chars) is the symbol set used on the display. For example, **ü** is 129 and **é** is 130. Using this option you won't need any character substitutions.

II.G.7.c.ii.) HP Roman 8 (includes all accented chars, different ASCII codes) puts the accented characters in different positions. For example, **ü** is printed as character 207 and **é** is 197.

Also, the line drawing characters are omitted in favor of extra accented characters. These translations must be mapped using the *character substitution* facility.

II.G.7.d. Character substitutions

This selection is identical to **Options**, *PATER character translation*.

Specify character substitutions (ASCII code translations)

Ersatzzeichen definieren (ASCII-Code Übersetzen)

See also:

Options	§II.G.4, page 70
PATER character substitution	§II.G.4.q, page 76
choosing	§I.J.1, page 24

[1] You don't normally have to set the printer to landscape in order to get the internal laboratory report (§II.A.7, page 33) to print sideways; the **Report** program will temporarily invoke landscape if needed.

II.G.8. COMMAND: **Update**

The **Update** command is used to help install a new version of PATER. Improvements and modifications to PATER consist of an update installation file such as `setupPATER13.59update.exe`. After executing the installer (per §119), the next time you start PATER you will be forced to invoke the **Update** command before the full command menu will appear.

Update usually needs to copy in some data file changes so that your data files will become compatible with the new PATER program release, and perhaps make some other changes for compatibility with the new release.

(The update files have been copied into the \DNAVIEW\UPDATES directory by INSTALL as part of the update installation. Once the **Update** command has been performed those files are no longer necessary.)

When prompted

> Please type Enter or Y to allow the update to proceed

you should type ***Enter*** or **`Y`**, as requested.

Invoking **Update** an extra time is harmless.

II.H. LEAVE MENU

The selection **Leave Menu** from the (hierarchical form of) the Command menu includes these options:

Quit from PATER		(§I.F.5, page 16)
DNA·VIEW	Switch to DNA·VIEW	
æ tools	Special facilities	(§II.H.1, page 82)

II.H.1. COMMAND: **æ tools**

The COMMAND menu item and command **æ tools** contains some special facilities. The ones not mentioned here are used only under supervision in order to make emergency bug fixes. The symbol **æ** is intentionally not typeable; select **æ tools** either via ***Backspace***, ***up arrow*** (since it is at the end of the menu), or by ***f2 Space*** **t**.

When through with **æ tools**, ***f3*** to return to the COMMAND menu. Here is one occasionally useful tool:

II.H.1.a. FileCompress (***Shft-F4***)

Sometimes some PATER files accumulate unnecessary dead space. FileCompress can be used, without supervision, to recover the space. Once **æ tools** are selected, ***Shft-F4*** invokes **FileCompress**.

A list is shown of all PATER 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 during the compression. Interrupting compression might cause a minor problem.

II.H.1.b. Other tools — Push_Me (***f1***)

The **æ tools** menu includes an item called **Push_Me** (***f1***). Press ***f1*** for the **Push_me** menu which includes:

New HLA (***f4***) allows adding new HLA alleles and changing the allele names.

Serology (***f2***, §II.H.1.b, page 83) lets you examine & to some extent edit the serological or STR systems.

æ tools: Serology (***f1 f2***)

```
Serological test
Categories of tests
DNA test name
Enzyme name
```

Figure 14 Serology top level menu

Serology is a **æ tool** rather than a command because it is experimental (i.e. not guaranteed to work in all cases) and not thoroughly documented. It has the following uses:

Change the spellings as they appear on the report:

i. of genetic systems,

ii. of "categories" i.e. "Red cell antigens",

iii. of a DNA probe or a restriction enzyme.

iv. Reclassify a genetic system (into a different category).

v. Put a genetic system into the category #0 so that it won't appear as a choice by **In — enter phenotype data**.

vi. Examine the genetic structure/statistics for a genetic system.

vii. Remove/Restore DNA probes/enzymes from the in-DNA menus.

To access these facilities, select **æ tools** from the COMMAND menu. Then type Serology (or ***f1 f2***) in response to the prompt:

```
Select tool, or F3 to resume COMMAND:
```

Serology *Enter*

and you will see the menu shown in **Figure 14**.

II.H.1.c. Serological test

Select this option to see a complete list of serological tests. It will also clarify part of PATER's organization of serological tests. Immediately you get a list of all serological systems (including STR's that you have created using **Create genetic System**).

```
Level 2: Modify or examine which test name? (Enter->top level)

ABO         ( 0]  ABO       [1] Red Cell Antigens )
Rh          ( 1]  Rh        [1] Red Cell Antigens )
MNSs        ( 2]  MNSs      [1] Red Cell Antigens )
Kell        ( 3]  Kell      [1] Red Cell Antigens )
Duffy       ( 4]  Duffy     [1] Red Cell Antigens )
Kidd        ( 5]  Kidd      [1] Red Cell Antigens )
GPT         ( 6]  GPT       [3] Red Cell Enzymes )
Gc          ( 7]  Gc        [2] Serum Proteins )
Bf          ( 8]  Bf        [5] Complement Factors )
Hp          ( 9]  Hp        [2] Serum Proteins )
AcP         ( 10] AcP       [3] Red Cell Enzymes )
DQα         ( 11] DQα       [4] HLA )
6-PGD       ( 12] 6-PGD     [3] Red Cell Enzymes )
ADA         ( 13] ADA       [3] Red Cell Enzymes )
PGM1        ( 14] PGM1      [3] Red Cell Enzymes )
EsD         ( 15] EsD       [3] Red Cell Enzymes )
Glo         ( 16] Glo       [3] Red Cell Enzymes )
```

Figure 15 Serological test list

Every serological system has two names in PATER. The short "internal" name is used for menus and you can't change it. The up to 16 char "external" name is the one that appears on the report, which you can change. The list in **Figure 15** shows the external name first, and the internal name + category in parentheses. The half-bracketed number is the internal test number. The bracketed number, before the category, is the category number which is explained below (II.H.1.d).

Select no test to return to the top level menu.

Select any test to

i. change the spelling of the external name.

```
New name (Enter if no change):

 6-PGD
6-PGD       ( 12] 6-PGD     [3] Red Cell Enzymes )
```

(Just hit ***Enter*** if you do not wish to change it.)

ii. change the category to which the test is assigned

iii. view the genetic definition of the system as understood by PATER (doesn't work for HLA)

```
***  TEST:      6-PGD
ALCEL, PHENOS, AMT
var var 1+1=1 var   1     B   B   2+2=2 B     1     A   A   4+4=4 A     1
B   var 2+1=3 B-var 1     A   B   4+2=6 AB    1
A   var 4+1=5 A-var 1     var A   1+4=5 A-var 1
var B   1+2=3 B-var 1     B   A   2+4=6 AB    1
ALELS (not used in application)
[0]   A
[1]   B
[2]   var
DOM (not used in application); 1st col dominates 2nd
Frequencies (by race):
var 0.00 0.00 0.00 0.00 0.00   A   0.98 0.98 0.96 0.90 0.98
B   0.02 0.02 0.04 0.09 0.02
ave allele freq    0.965 0.951 0.915 0.827 0.957
ave phenotype freq 0.931 0.906 0.841 0.698 0.917
```

Figure 17 Genetic definition of 6-PGD

Example: **var B 1+2=3 B-var** means that the **var** and **B** genes (internal codes 1 and 2) in combination produce the **B-var** phenotype (internal code 3).

The "ave freq"'s represent averages over a population, not just averages over the set of numeric frequencies. That is, they are weighted according to expected occurrence in the population assuming Hardy-Weinberg equilibrium.

i. show *Exclusion power* statistics — as phenotypes, or as paternity systems.

ii. show *Race discrimination power*:

displays statistics as shown in **Figure 17**. The second chart is the interesting one, but to help understand it the first chart, phenotype frequencies, are shown. Note from the first chart that "a-b-", meaning the Duffy blank gene, is common among Blacks but non-existent with other races.

Mostly because of this extreme phenotype, the typical Black phenotype is far rarer — about 57 times rarer — among Caucasians than it is among Blacks.

On the other hand, if we evaluate the Duffy phenotypes of a number of Caucasian's, they will typically be about 4 times rarer among Blacks than among Caucasians.

One way interesting way to consider these statistics — albeit perhaps only academically interesting — is this. Suppose it is desired to guess the race of the donor of a bloodstain. If the stain is twice as common,

Phenotype frequencies for Duffy

	Cauca	Hispa	Black	Asian	Germa
a-b-			**0.567**	0.001	
a-b+	0.358	0.204	0.223	0.015	0.312
a+b-	0.175	0.32	0.179	0.816	0.211
a+b+	0.466	0.476	0.03	0.168	0.477

Underestimation of phenotype frequency by using wrong race

	Cauca	Hispa	Black	Asian	Germa
Cauca		1.09	***4.21***	3.87	1.01
Hispa	1.09		4.37	2.08	1.05
Black	***57.4***	58.4		55.62	57.25
Asian	2.83	1.74	4.4		2.43
Germa	1.01	1.05	4.27	3.21	

Row = actual race of a sample

Column = alternate database race used for frequency computation

Entry = expected, typical ratio of the apparent odds against a phenotype match by using the wrong race to the odds using the actual race. "Expected" is in the sense of geometric mean, so these numbers can be combined between systems by multiplication. The table can be interpreted as predicting how much information the system will supply for guessing the race of a stain donor.

Figure 18 Duffy as a predictor of race

for example, among one race as among another, then all other things being equal it is twice as likely that it comes from the race in which it is common. Therefore Duffy is on average a very useful test for making a race discrimination between Black and any other race.

II.H.1.d. Categories of test

The category names (e.g. "Red cell enzymes") appear on the paternity report as section titles: within each category-section are listed the test results and calculations for that category.

II.H.1.d.i.) **Category 0 ("not used") is a pseudo-category**. If there is some test that you never use, you can put it in this category and that will prevent it appearing in the menu of tests presented by In - enter phenotypes. (On any particular case report each test appears only if data was entered for that test.)

II.H.1.d.ii.) **Categories 1 through 8 are arbitrary**. Any serological or STR test defined in PATER, whose data is entered using In - enter phenotypes, can be assigned n to any one of these categories.

II.H.1.d.iii.) **Categories 9 and 10 are special**, and are used specifically for DNA data. Such data is usually imported from DNA·VIEW, but alternatively and equivalently it may be entered using in DNA. Any DNA systems that are 8 defined as STR systems[2] are listed on the PATER report under category 9. Other DNA systems, normally RFLP systems, are listed under category 10. You should never assign any of the PATER systems — that is, systems for which data is entered using In - enter phenotypes — to either of these categories.

```
Level 2:  Modify or examine which
     category? (Enter->top level)
Note:  0] means "not in use";
       9] is PCR tests from DNA·-
          VIEW or in DNA
      10] is RFLP tests from DNA·
          VIEW; K triggers Kbase
          format.

  0]   (test not used)
  1]   Red Cell Antigens
  2]   Serum Proteins
  3]   Red Cell Enzymes
  4]   HLA
  5]   Complement Factors
  6]   Example Calculation
  7]   PCR Polymarker test
  8]   PCR Gel tests
  9]   STR alleles
  10]  DNA Probes (Kbase)
```

The description for categories 9 and 10, as for any category, can be modified by the user. There is one important point about the description for category 10 however: the presence or absence of the capital letter **K** is used as a signal to format DNA sizes as kilobases rather than bases.

Category 8 is the category to which STR systems are automatically assigned when you create them with Create genetic System (Section II.C.3). It is permitted to reassign them, however.

Once you select a category, you are given a chance to change the name. Press ***Enter*** to leave it as is. Then you are given a shortened list of tests, showing just those in the selected category, and operation proceeds as in section II.H.1.c.

II.H.1.e. DNA test name

[2] PATER distinguishes category 9 and category 10 according to whether or not there is a non-0 repeat amount specified in the PCR parameters (specified in DNA·VIEW using the command **Maintenance**, Probe/locus parameters.

DNA tests (category 10) refers to DNA data imported from DNA·VIEW (or, rarely, input through In DNA) and are treated quite separately from STR tests in PATER or serological tests. The methods are documented on the screen.

II.H.1.f. Enzyme name

Enzymes can be re-spelled or inactivated in a similar manner.

III. Kinship

III.A. Conceptual Overview

Figure 20 Kinship chapter index

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)[3], and some examples illustrating the method are presented in §III.I.6 and throughout this chapter.

This chapter, III, shows the steps using Kinship and other commands to analyze a typical kinship problem. Section III.I explains some special considerations: the need for a realistic δ (Section III.I.1), and explains such tricky situations as twins (Section III.I.3) and missing persons (Section III.I.4).

III.B. Kinship in PATER

III.B.1. Two versions of Kinship

There are two versions of the Symbolic Kinship Program in DNA·VIEW – Kinship and the Case option Immigration/kinship. PATER has only Kinship. (The PATER command Immigration is not related to the Symbolic Kinship Program.) For compatibility (mostly of documentation), a comparison is outlined here.

[3] Symbolic Kinship Program, Genetics 145:535-542

III.B.1.a. Kinship

is the original version, and is still essential in certain situations:

III.B.1.a.i.) to obtain the formula for a hypothetical problem, without having any actual data;

III.B.1.a.ii.) systems with null alleles;

III.B.1.a.iii.) X-linked systems;

III.B.1.a.iv.) loci that are not defined as DNA loci;

III.B.1.a.v.) to verify that the input is correct (use "parsing and hints");

III.B.1.a.vi.) if you don't have the full DNA·VIEW system, but only Abridged, Toolbox, or PATER.

III.B.1.b. immigration/kinship (DNA·VIEW only)

It is essentially an automatic version of Kinship which requires less user attention and time in most cases because

III.B.1.b.i.) A user interaction is not necessary for each locus separately.

III.B.1.b.ii.) The program automatically determines the genotype patterns; you don't type them in.

III.B.1.b.iii.) The program automatically looks up the allele frequencies and calculates the likelihood ratios.

III.B.2. Limitations of both versions

The Kinship program (either version)

III.B.2.a.i.) computes only a simple loci, and does not understand haplotype systems such as Rh or HLA;

III.B.2.a.ii.) assumes the same population group for all individuals;

III.B.2.a.iii.) does not handle mutations particularly well.

Further development work may cater to mutations. The other limitations are not annoying in a practical sense.

III.B.3. Limitations of the PATER version

III.B.3.a.i.) A stand-alone program, Kinship does not directly interact with other parts of PATER or DNA·VIEW such as allele frequency tables. Allele frequencies need to by typed in by the user each time.

III.B.3.a.ii.) The user needs to type in the genotype patterns, and since these are different for each locus separate entry is needed for each locus.

The program does, though, make a summary calculation covering all loci.

III.C. The Kinship language

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.

The script defines the two *scenarios* that are to be compared:

III.C.1.a. Principal scenario — The expected pedigree of a set of relationships among the people.

III.C.1.b. Alternate scenario — An alternate pedigree or way they may be related.

It may include:

III.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

III.C.1.d. People — some typed, some may be untyped.

and, only in the case of the stand-along Kinship version:

III.C.1.e. Phenotypes — for some or all of the people.

III.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.

III.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.

III.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`**.

III.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`**

III.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!

III.C.2.d.i.) Simple but verbose way to designate alternatives

Put the **`/`** symbol in front of each line of the alternative scenario.

III.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** **/C : M + Joe**	**C : M + F/Joe**
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** **/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** **/ 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** **/ F : M + C**	**C/F : M + F/C**

Advanced syntax rules (shortcuts):

III.C.2.e. **Statement separator**

III.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.

III.C.2.e.ii.) When using the "verbose" syntax method (**/** at the beginning of alternative scenario lines), there is just one **/** 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
```

III.C.2.e.iii.) **More attractive alternative separators** (recent)

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**

III.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.

III.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 a discussion, see §III.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 un-named person?	**C : M + F** **/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** **/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** **/ ? : M+F**	**C/? : M+F**
avuncular	... that **F** is the uncle of **C**, as opposed to being unrelated?	**C : M + Bro** **Bro, F : ?+?** **/ C : M + ?**	**C : M + Bro** **Bro, F/? : ?+?**
incest	... that **C**'s father and uncle are the same person? ("Mom" assumed to be untyped.)	**C : Mom+U** **Mom, U : ?+?** **/ ; empty line!**	**C : Mom+U/?** **Mom, U : ?+?**
sib-ship	... that **A** and **B** are full siblings, as opposed to being unrelated? (Parents not typed.)	**A, B : ?+?** **/ ; empty line!**	**A, B/? : ?+?**

III.C.3. How to learn the Kinship Language

The Kinship program computes a likelihood ratio comparing two different pedigrees as explanations for the same genetic data.

III.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 21**. Alternatively (H_0, **Figure 22**), some "Other" body may be the missing child.

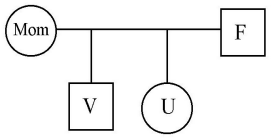


Figure 21 full sibling

H_1: The sample named V represents the victim who is the child of reference F and brother of reference U.

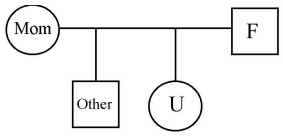


Figure 22 unrelated

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` `U : Mom + F`	`Other : Mom + F` `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.

III.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.

III.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.

III.C.3.b. **Two pedigree transcriptions**

Under each picture – on the left and right sides of the page – transcribe the pictures into linear notation.

III.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.

III.C.3.b.ii.) Do not use the **?** or **,** shortcuts for either of the transcriptions.

III.C.3.c. Two ways to enter the pair of pedigrees

III.C.3.c.i.) Simple way

Type in every line of the primary hypotheses	`V : Mom + F` `U : Mom + F`
Type in each line of the alternative hypotheses preceded by a slash (/)	`/ Other : Mom + F` `/ U : Mom + F`

» There one possible small complication: If the alternative hypothesis is *empty* – that is, if the alternative hypothesis is that no one is related to anyone else – you must still but in at least one line beginning with a slash. It can be otherwise blank, or can contain a comment, or can contain a dummy relationship.

III.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 III.C.3.b.

This method gives a more concise formulation but sometimes merging seems tricky.

The (either) result of III.C.3.c.ii may be typed into the program.

III.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.

III.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:

III.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
```

III.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:

III.C.3.e.iii.) The letter **o** can only be used for a null allele.

III.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 III.H.2.a).

III.C.3.e.v.) For an X-linked system, male types should be written with a dash, i.e. **r-**.

III.C.4. Kinship Semantics, clarification, and examples

III.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*). Conversely, one may write **Johnny : ? + Dave** even though Johnny has only one possible mother.

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.

III.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 (§III.C.3.c.i), one concise (§III.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.

III.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
/
```

).

III.C.5.a. Why one hypothesis?

III.C.5.a.i.) when the alternative is "no one is related."

III.C.6. Prior probabilitiy

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].[4]

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.

III.D. Multiple hypotheses in Kinship (recent)

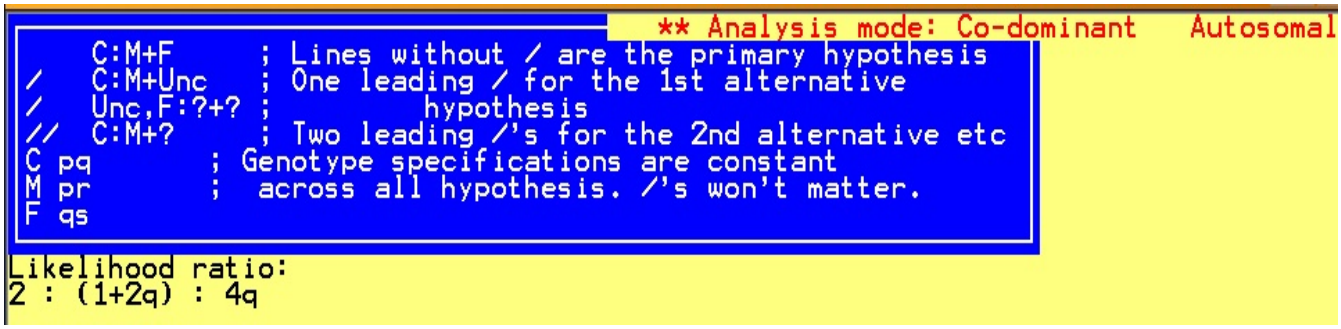
A Kinship scenario may compare more than two hypotheses.

III.D.1. Multiple hypothesis syntax (recent)

III.D.1.a. example

The input syntax shown here is based on the "simple method." Example:

[4] 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.



The three hypotheses are: The man F is the father; the man F is the uncle; the man F is unrelated.

III.D.1.b. rules

III.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.

III.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 §III.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.

III.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*.

III.D.2. Multiple hypothesis result (recent)

The result is a list of the relative likelihoods corresponding to the several hypotheses. For example:

```
2 : (1+2q) : 4q.
```

These correspond to the hypotheses *primary*, *1st alternative, 2nd alternative*.

III.D.2.a. Interpretation of multiple likelihoods

III.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,

» $1/2q = 2:4q$ is the LR by which the evidence supports the primary hypothesis over the 2nd alternate;

» $½+1/4q = (1+2q):4q$ is the LR by which the evidence supports the 1st alternate hypothesis over the 2nd alternate;

» $2/(1+2q) = 2:(1+2q)$ is the LR by which the evidence supports the primary hypothesis over the 1st alternate.

III.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.

III.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.

III.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*****≠***Edward*.

III.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 §III.D.2.a.ii.

III.D.2.e. Posterior probabilities (recent)

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.

III.E. The Kinship command (stand-alone Kinship program)

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 Kinship's calculation is an algebraic formula for the likelihood ratio of the relative probabilities of the two pedigrees. If you supply the frequencies, it will then evaluate the formula.

III.F. Kinship cases

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 frequencies 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 frequencies, 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 §III.C.

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

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 (=Reprint)

III.G. Overview of using (stand-alone) Kinship

Kinship and its menus are designed to be convenient for several different possibilities of working through a kinship problem.

III.G.1. a new problem

To work through a completely fresh problem, the typical order of operations is:

III.G.1.a. preparation

Lay out the genotypes for each locus and assign letters in a consistent way to each allele.

III.G.1.b. Clear complete case

III.G.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 §III.H.11.

III.G.1.d. Find formula for locus

III.G.1.e. Evaluate formula. (Supply relevant allele frequencies as requested.)

III.G.1.f. Post this locus to the complete case

III.G.1.f.i.) Post to what row? Select Locus or Serological test or arbitrary as desired.

III.G.1.f.ii.) Choose a name and the pedigree/locus is posted under that name within the complete case.

III.G.1.g. File or fetch a complete case. For safety, file the case so far to disk.

III.G.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 (§III.H.13).

III.G.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:

III.G.2.a. preparation (per §III.G.1.a)

III.G.2.b. File or fetch a complete case. Retrieve the model that will be modified.

III.G.2.c. Rework, reorder, or delete a locus

III.G.2.c.i.) Clear all the old likelihood ratios by moving the red bar to the first locus, then hitting **0** once per line.

III.G.2.c.ii.) Select the [next] locus to rework.

III.G.2.d. Edit pedigree/locus. Change the definition as necessary. Find formula, Evaluate, Post, File (using a new name) as per the steps in §III.G.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 (§III.H.13).

III.H. Using the manual Kinship Command

III.H.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 §III.C. For example, the pedigree diagram **Figure 24**, **Figure 25** 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:

III.H.1	Type in a new pedigree/locus
	Read pedigree/locus from *.KIN file
	Save pedigree/locus as a *.KIN file
III.G.2.d	Edit pedigree/locus
III.H.3	Find formula for locus
III.H.4	Algebraically simplify formula
III.H.5	Evaluate formula
III.H.6	Display pedigree/locus
III.H.13.b.iv	Insert pedigree/locus in report
III.G.2.c	Rework, reorder, or delete a locus
III.H.7	Post this locus to the complete case
III.H.8	Display case summary
	Clear complete case
III.H.13.b.i	Clear the report
III.H.13.b.ii	Insert case summary in the report
III.H.13.a	Print the case summary
III.H.10	File or fetch complete case
III.H.13	Print report (=Reprint)
	quit
III.H.2.a	Silent alleles: not allowed
III.H.2.b	Locus is: autosomal
III.H.2.c	Parsing & hints are: not shown

Figure 24 Kinship options

```
; Example reconstruction case
; locus D3S1358
Child pr
Mom pq
Auntie pr
Granny pr
Child : Mom + Allegedfather / ?
Allegedfather, Auntie : Granny + Gramps
```

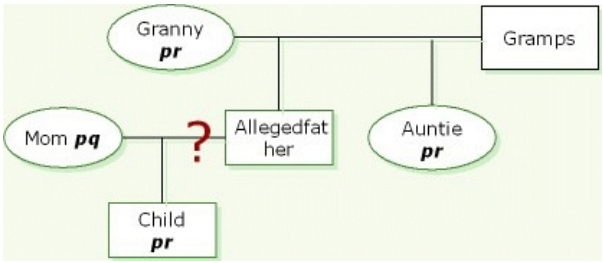


Figure 25 Deficiency case pedigree

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 saved to disk by the ***ctrl-e*** action, but only held temporarily for analysis.

III.H.2. Set option toggles

Several of the menu items are toggles that control the way the analysis is performed.

III.H.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 §III.I.5.

If the ***o*** allele is specifically mentioned in the scenario definitions, then the analysis will automatically allow silent alleles.

III.H.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*.

III.H.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.

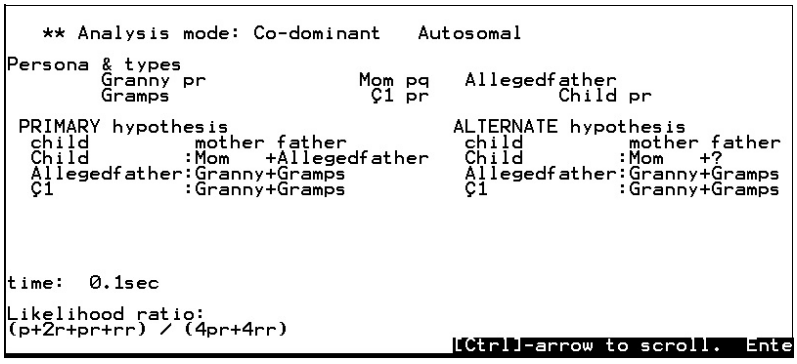


Figure 26 Parsing output

Figure 26 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 (§III.H.3) and evaluate formula (§III.H.5).

III.H.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*`+2`*r*`+`*pr*`+`*rr*`)/(4`*pr*`+4`*rr*`).`

Symbols *p, r* etc. are used to denote gene frequencies corresponding to the symbols `p, r` etc. that designate genes on the input.

III.H.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 (§III.H.2.c) is turned on, the display is enhanced and also includes estimated time to finish.

III.H.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.

III.H.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.

III.H.5. Evaluate formula

Select the Evaluate formula option in order to evaluate the algebraic expression with numeric gene frequency values, which are obtained from the "reconstruction" table (?, page ?).

Evaluate formula creates a form on the screen in which you can type allele frequencies, 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, strike ***esc***.

For the example above and locus D3S1358, the result is

```
Using p=0.113 r=0.223    value = 2.12
```

If Parsing & hints (§III.H.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 frequency value is *anti*-conservative in the sense of exaggerating the strength of the evidence favoring the principal scenario.

III.H.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 27**.

III.H.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

III.H.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.

III.H.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.

III.H.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.

III.H.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, §III.G.2.c), modifying it, then putting it back.

III.H.7.e. no option (***backspace enter***)

has the effect of escaping from the Post this locus to the complete case submenu without doing anything.

III.H.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 27**.

III.H.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.

III.H.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

III.H.10.a. (a new case)

to create a new complete case entry in the kinship case library.

III.H.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.

III.H.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:

III.H.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.

III.H.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 III.H.10.a.i above.

III.H.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.

III.H.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 24**, **Figure 25** 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 III.H.3 etc. The formula for this pedigree/locus will be given as:

```
Likelihood ratio:   (1+r) / (4r)
```

and the result of step III.H.5 is

```
Using  q=0.258     value = 1.22
```

III.H.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 frequencies 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 §III.G.2 works well in such a case.

III.H.13. Print the results

The computations are displayed on the screen as you work. They are also saved to the report buffer, which you can print in the usual way from the main menu using Reprint.

However, the 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:

III.H.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-per-

```
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 27 Kinship case summary

locus summary of the case including the overall likelihood ratio.

III.H.13.b. Make a report with full detail

III.H.13.b.i.) Clear the report — empties the report log

III.H.13.b.ii.) Insert case summary in the report — puts the case summary, e.g. III.H.13.a as the top of the report-in-progress.

III.H.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.

III.H.13.b.iv.) Insert pedigree/locus in report puts the kinship file, the likelihood ratio formula and value, and documentation as the chosen allele frequency 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 III.H.13.b.iii.

III.H.13.b.v.) Print report (=Reprint) after all pedigree/loci have been added to the report.

III.H.14. Explaining the result

Classically people considered three statistics concerning paternity:

III.H.14.a. 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.

III.H.14.b. W=probability

> In the paternity case, W=probability of paternity. The present context may require different words of course.

III.H.14.c. LR=L=likelihood ratio

> This is the only statistic that is calculated and reported by **Kinship**.

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.

III.H.14.d. Paternity question; moderate evidence

> For the example case of the preceding few sections comparing paternity versus nonpaternity and with likelihood ratio computed in section 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."

III.H.14.e. 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."

III.I. Kinship — Special Notes

III.I.1. Choosing δ (RFLP)

Important note: The calculation will be much more likely to be useful if the gene frequencies are small. If fact it may be absolutely essential to switch to a **realistic** value for δ (for purposes of calculating frequencies) rather than the conservative value that you use for ordinary casework.

Here's an illuminating example. A brother and a sister want to prove their relationship (versus unrelated). Even if we determine their DNA types at many loci, there will be several loci (about ¼ of the loci in fact) where they will share no alleles. Each of these loci contributes a likelihood ratio of ¼ — evidence against relationship. At half the loci they will share one allele Q and the likelihood ratio will be $^{1}/_{8\mathbf{P(Q)}}$ (only ¼ of the typical amount in a paternity trio case). If a realistic value for **P(**Q**)**=0.02, then the likelihood ratio contribution is a factor of about 6 and eventually the 6's will overwhelm the ¼'s. But, if your δ is generous and you consequently estimate **P(**Q**)**=0.08, say, then the ¼ factors may well dominate, which would be misleading.

If the value of `range÷δ` between mother and child in casework happens nearly always to be under 25%, then I think that the true, realistic value for δ is 25% of the (conservative) value in use for casework. Such a smaller δ would result in frequencies typically 4 times smaller also, so if the client (i.e. in the deficiency case) simply wants my best estimate of the truth I would use the smaller frequencies.

III.I.2. Comparing three or more scenarios

Kinship only compares two pedigrees or scenarios at a time. However, since the result is in the form of a likelihood ratio making a comparison among three or scenarios is quite straightforward.

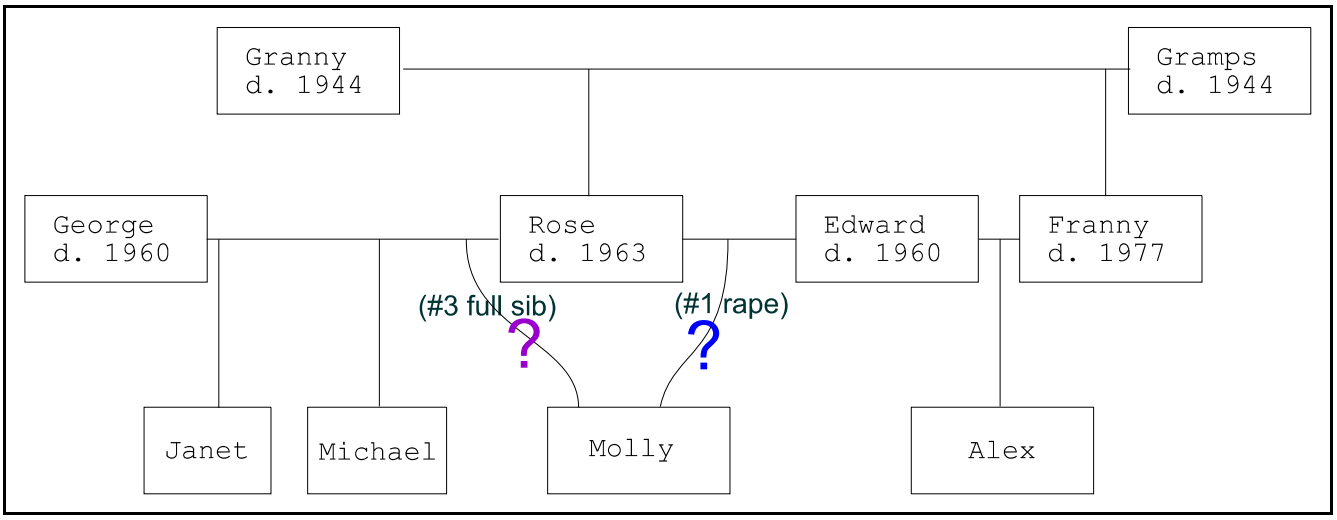


Figure 28 The case of Molly

To illustrate the logic, consider the case in **Figure 28**. It is desired to decide the parentage of Molly based on genetic types that are available for the four children in the last generation. These relationships are undisputed:

```
Rose, Franny   :   Granny + Gramps
Janet, Michael     :   Rose + George
Alex           :   Franny + Edward
```

and there will also be genetic information which can either be interspersed in the above lines, or can be entered as additional lines.

In addition, we would like to consider these possibilities for Molly:

Scenario #1 (rape)	`Molly : Rose + Edward`
Scenario #2 (stranger)	`Molly : ? + ?`
Scenario #3 (full sib)	`Molly : Rose + George`

Each operation of Kinship always computes and compares *two* scenarios. To combine #1 and #2, write the line

```
Maureen    :   Rose / ? + Edward / ?
```

and add this to the undisputed lines above. After choosing find formula and evaluate formula, you will end up with a likelihood ratio, such as 50, which would mean that #1 is 50 times better than #2 as an explanation for the genetic results.

Next, you may choose to compare #1 and #3 by using instead the line

```
Maureen        :   Rose + Edward / George
```

Suppose you get a likelihood ratio of 5 in this case, meaning that #1 is only 5 times better an explanation than is #3.

Now if you wish, you can also make a computation comparing #3 and #2 by using instead the line

```
Maureen        :   Rose / ?    + George /   ?
```

but it's hardly worth the trouble, because from the preceding comparisons we already know that #1 compares to #2 as 50:1, and #1 compares to #3 as 50:10. Therefore likelihood ratio comparing #3 to #2 will certainly be 10:1 = 10.

III.I.3. 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: the **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.

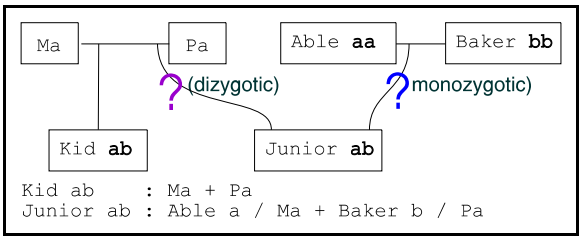


Figure 29 Monozygotic vs. dizygotic twins

Figure 29 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 dizogotic 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 29**. Finally, combining (3) and (4) in legal notation:

```
Kid ab : Ma + Pa
Junior ab : Able aa / Ma + Baker bb/ Pa
```

III.I.4. Missing persons

A person who is reported missing has both descendants and ancestors that can be typed, as in **Figure 29**. 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 34** for another example.

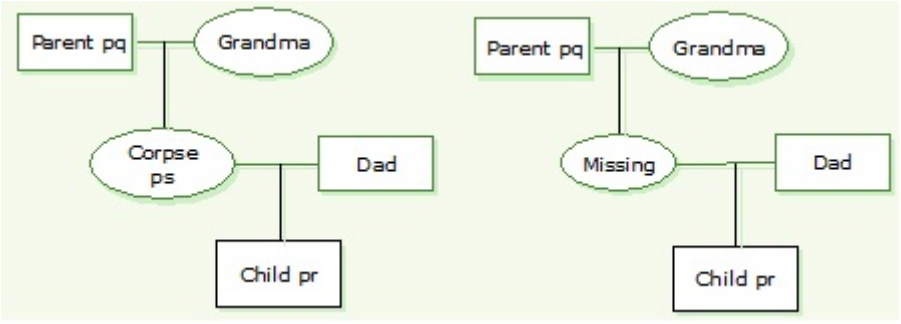


Figure 30 Missing person solution

III.I.5. 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).
```

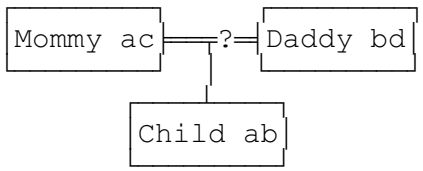
This can be factored as (*o*/(2*o*+*q*)) / (*p*+*o*). Compare the numerator and denominator with the formulas in sections XI.C.6.c.ii and XI.C.6.b.

III.I.6. Kinship Examples

These examples are to be found in the \DNAVIEW\EXAMPLES directory, so you can quickly run them using **Kinship**.

The most familiar paternity situation, expressed in "Kinship" notation:

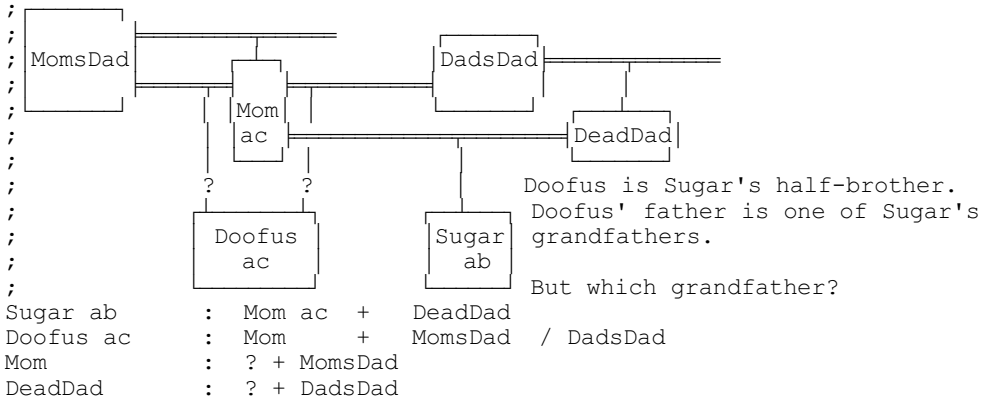
```
;  This file defines a simple paternity trio with heterozygous people.
;  Anything after the semicolon is a comment.
;  People's names begin with a capital letter.
;  Phenotypes use lower case letters.
;  / separates alternate possible parents. ? means an unknown person.
;  + means mother on the left, father on the right.
;  : means child on the left, parents on the right.

Child ab : Mommy ac + Daddy bd / ?
```



Likelihood ratio: 1 / 2b

Figure 31 TRIO.KIN

A peculiar incest situation:



Likelihood ratio: (2+2a+2c) / (3a+3c)

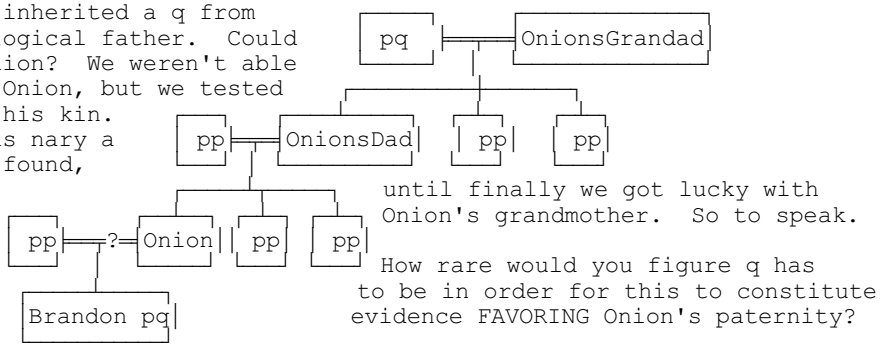
Do Algebra

Likelihood ratio: (2/3)(1 + 1/(a+c))

Figure 32 PUMP.KIN

Many relatives tested:

```
;  Brandon inherited a q from
;  his biological father.  Could
;  it be Onion?  We weren't able
;  to test Onion, but we tested
;  lots of his kin.
;  There was nary a
;  q to be found,
;
;
;
;
;
;
Brandon pq : pp + Onion / ?   ; "pp" means definitely homozygous for "p"
           ; You don't need to put a name for Brandon's mom.
Onion, pp, pp : pp + OnionsDad ; Onion & two pp sisters have same parents.
OnionsDad, pp : pq + OnionsGrandad  ;  Onion's grandma finally has a q!
```



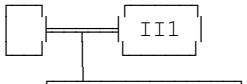

```
;                                  until finally we got lucky with
;                                  Onion's grandmother.  So to speak.
;
;                                  How rare would you figure q has
;                                 to be in order for this to constitute
;                                 evidence FAVORING Onion's paternity?
```

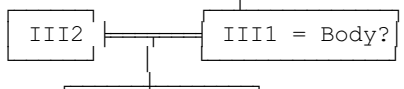
Likelihood ratio: (1+p+q) / (24q+16pq)

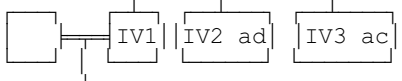
Figure 33 ONIONS.KIN

A missing person:

```
;MISSING PERSON Is the body the same as person III1 from the family tree?
Body ac              ; a body is found & typed.
                                          ;
                                          ;
                                          ;
Body / III1          :   ?  +  II1        ;
                                          ;
IV1, IV2 ad, IV3 ac : III2 + Body / III1;
                                       ;
                                       ;
                                       ;
                                       ;
V1 ab                :   ?  +  IV1;
                                       ;
;   The example is from a paper by Egelund & Mostad
```







Likelihood ratio: (2a+c) / (4ac+24aac)

Figure 34 MISSING.KIN

IV. Special Protocols and Irregular Cases

IV.A. Discussion

Paternity cases (including motherless) and maternity cases (including fatherless) are handled automatically by the paternity system. Besides that, an avuncular index is calculated for each case. Also, the **immigration** command in PATER (§II.B.6, page 41) (not to be confused with the immigration version of **Kinship** in DNA·VIEW) makes an additional uncle computation that is sometimes of interest.

Other irregular cases are best computed using **Kinship** (Chapter 1).

IV.B. Motherless Cases

Sometimes the mother is not available for testing. In such a case the program will automatically perform the correct analysis based on the phenotypes of the child and the tested man.

IV.B.1. Creating a motherless case

To signal that a case is a motherless case, either leave the mother's name blank when entering people's names, or enter a remark in parentheses in place of the mother's name, e.g.

```
Mother  (not tested)
```

Having done that, the **in** and **in DNA** commands will now only request phenotypes for the child and the tested man.

If you create a case WITH a mother, and later wish to change it to motherLESS, just invoke **people** and remove the mother's name. The case will be recalculated automatically when you prepare a report. However, any phenotypes entered for the mother will be recalled by the program if you later decide to revert the case to WITH mother.

See also

People	§II.B.8, page 43
In — enter STR or serological phenotypes	§II.A.4, page 28
entering DNA probe data	§II.A.5, page 30

IV.C. Standard Cases

IV.C.1. Motherless Case

H_1	mother unknown	H_0	mother unknown
	Man is the father		father unknown

See §IV.B.1, page 115.

IV.C.2. Paternity Case

H_1	Woman is the mother	H_0	Woman is the mother
	Man is the father		father unknown

See §I.I, page 17.

IV.C.3. Maternity Case

H_1	Woman is the mother	H_0	mother unknown
	Man is the father		Man is the father

Invoke **maternity** (§II.B.7, page 42) either before or after entering the data. Print the report.

Create an ordinary paternity case, but leave the mother's name blank (§IV.B.1, page 115).

IV.C.4. Fatherless Case

H_1	Woman is the mother	H_0	mother unknown
	father unknown		father unknown

Ask for a **maternity** (§II.B.7, page 42) case, and leave the father's name blank.

IV.D. The Avuncular Index

IV.D.1. The Avuncular and Incest Index Chart

The laboratory (internal) version of the paternity report includes a table that indicates the plausibility that the true father is a blood relative either of the accused man or of the woman — the "avuncular" and "incest" indices, respectively.

The columns of this chart refer to "degree of relation" — 0 meaning genetically identical, 1 meaning sharing 1/2 of the genes, relatives of degree 2 share 1/4 of their genes, and so on.

The 1st row ("avuncular") refers to paternity by relatives of the tested man: himself, his brother (i.e. unclehood by the tested man), his uncle, etc.

The 2nd row ("incest") considers possible paternity by blood relatives of the woman: a genetic clone ("parthenogenesis"), her father or brother, etc. This row is of course omitted in a motherless case.

See also

DNA probes (§II.A.5, page 30)

IV.D.2. The Avuncular Index

Whenever there is an exclusion in a case, the computer reports what I call the avuncular index, or AI. (When a paternity index or PI is mentioned in exclusion cases, it refers to non-excluding systems; otherwise it would always be 0).

The AI is defined as

X' = the probability that this man's brother (or father or son) would sire such a child, given this woman

divided by

Y' = Y = the probability that a random man would sire such a child in combination with this woman.

X' is computed system by system via the formula

$$X' = (X + Y) / 2.$$

The ratios for the individual systems are then multiplied together.

See also

avuncular chart on laboratory report §IV.D.1, page 117

IV.D.3. Interpreting the Avuncular Index

Suppose an alleged father turns out to be excluded. It follows that the woman had intercourse with at least one other man, which in all probability means at least two men.

Under these assumptions alone, the probability that she had intercourse with a brother of the accused is perhaps 1% to 10% (derived from part guessing and part common sense). Converting % to odds, that is 1:99 or 1:9.

Now suppose we compute the avuncular index and it is 9, meaning the tested man looks 9 times more like an uncle than a random man would. Multiplying this likelihood ratio times the prior odds of 1:99 or 1:9 gives posterior odds of 9:99 or 9:9 that a brother is the actual father.

Under such circumstances, it is likely that the woman can shed some light on the matter. If no relative exists, then 1% is already too high a figure. But if she in fact agrees that there is a highly eligible brother, then we privately reassess his prior odds at 2:1 (the odds for a typical accused man).

Combining 2:1 with the avuncular index of 9:1, we expect a likelihood ratio of 18 if the brother should appear for testing. (That is, we reckon to exclude him 1 time in 19, and include him very strongly the rest of the time.)

V. APPENDIX — INSTALLATION AND UPDATE

V.A. Installation

The computer must be a PC, a Pentium for example. Mac with a Windows emulator doesn't work.

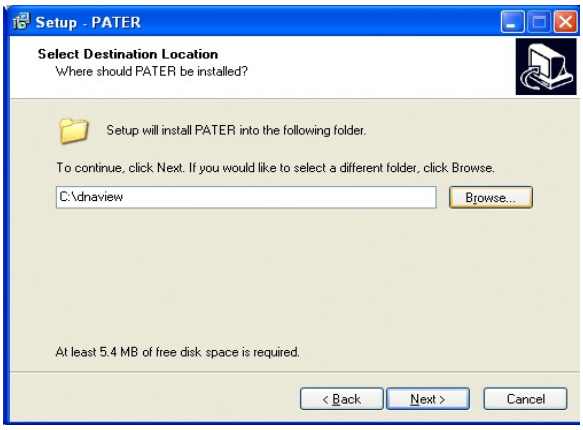
PATER consists of one CD, or a file obtained from the Internet. The installation file has a name like `SetupPATER....EXE`. If using an installation CD, setup should autostart, but you can browse to the file (**My Computer** etc.) and click on it if not.

V.A.1. Answer the prompts.

Password

There may or may not be a password..

V.A.1.a. **Select Destination Location**



V.A.1.a.i.) If updating or if adding PATER to an existing DNA·VIEW system, choose the same directory as was chosen for initial PATER or DNA·VIEW installation.

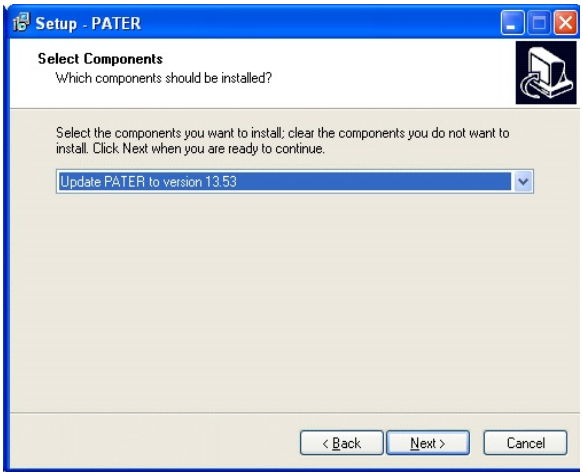
WARNING: Sometimes the installer suggests a location with an extra "dnaview", such as `C:\dnaview\dnaview`. Don't be fooled!

V.A.1.a.ii.) For a **new installation**, anywhere you like. Examples:

» Traditionally `C:\dnaview`.

» Typical Windows style would be `C:\Programs and Files\dnaview`.

» For installation to a **network server**, see §V.C. Note that there must be a mapped drive letter.



Click **Next**.

V.A.1.b. **Select components**

The component choices depend on whether the

installation setup file is for update or is an original PATER installation.

In total there are 3 possible options:

V.A.1.b.i.) Choose **Original (my lab) PATER stand-alone installation** if you have neither PATER nor DNA·VIEW.

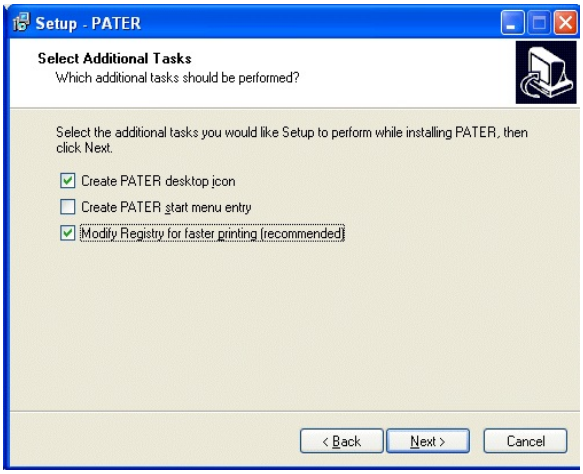
V.A.1.b.ii.) Choose **Add (my lab) PATER to existing DNAVIEW system** if you have DNA·VIEW but not PATER. All DNA·VIEW data will remain intact.

V.A.1.b.iii.) Choose **Update PATER to version xx** to install a new version. All your case data will remain intact.

V.A.1.c. **Start menu folder**

This refers to the program group where PATER will be found when you click START, All Programs. I usually check the box **Don't create a start menu folder** because I always use a desktop icon to start the program. If you do wish a **Start** menu entry use the group name `DnaView`.

Click **Next**.



V.A.1.d. **Select additional tasks**

If you already have a desktop icon folder that you don't want to change, put a checkmark in the box "Don't create any icons."

V.A.1.d.i.) `Create PATER desktop icon`. Very handy – an icon on your desktop to click to start PATER.

The installation creates a desktop folder called **DnaView Icons** with several icons, particularly the main program startup icon **PATER**. Drag or copy it directly to the desktop if you prefer. The icons are discussed below in §V.A.3.

V.A.1.d.ii.) `Create PATER start menu entry`. Less handy but some prefer it.

Click the obvious buttons to complete installation.

V.A.2. 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.

V.A.3. Icons

The installation produces a folder called **Dnaview Icons** on the desktop with a collection of icons for PATER (and for DNA·VIEW if present).

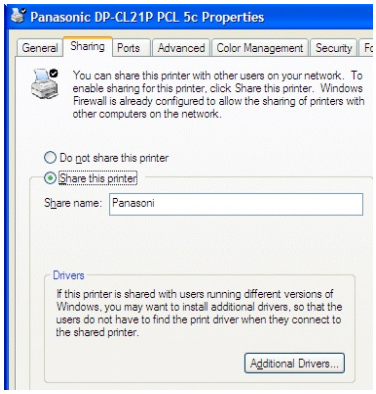
V.A.3.a. Icon functions

V.A.3.a.i.) **PATER** – starts PATER

V.A.3.a.ii.) **Connect USB printer**

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.



V.A.3.a.iii.) Print icon customization

» Choose the network or USB printer you wish to print to.

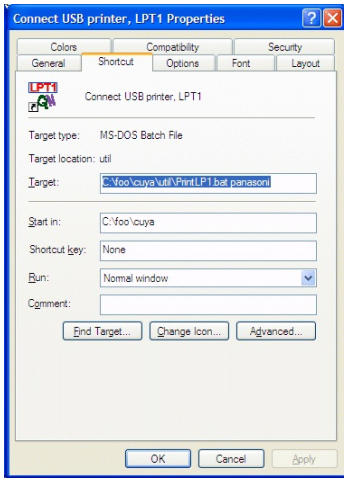
» Determine its "Share name". The easiest situation is if the printer can be shared through your own computer. Open **Printers and Faxes** from or near the Control Panel, right click on the printer you want to use, and choose Properties. Choose the Sharing tab. Choose the option **Share this printer** if possible. Note the Share name. If there is none, add one of your choosing, preferably without spaces. If the name is just an IP address (like `122.34.35.129`), you must create a regular name instead.

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 §V.A.4.



V.A.3.a.iv.) 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.

V.A.3.b. The **Repair** icon offers a choice of some maintenance processes.

V.A.3.c. **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.

V.A.4. Customizing the icons

(recent – 2007)

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:

V.A.4.a. Right click an icon and choose **properties**

V.A.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**.

V.A.4.a.ii.) enable mouse for menu. Click the **options** tab and make sure that **Quick edit mode** is *not* checked.

V.A.5. Start PATER

Execute (or allow to execute) the command *Update* (§II.G.8) from the **Miscellaneous** menu.

V.A.6. Installation — Check

V.A.6.a. Numlock status

See §II.G.4.p for the *Option* to toggle the numlock status.

V.A.6.b. Printing

See §II.G.4.g and §II.G.4.h for *Option*s to specify the printer and the printer port, and §V.A.3.a.ii for printing under Windows.

V.A.6.c. **Extended or expanded memory** for PATER under Windows

Especially if you experience a WS FULL error message from PATER, 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 PATER will be allocated.

```
wssize=20000
```

is a modestly small allocation (representing 20 megabytes) considering the amount of memory most computers have available. The minimum acceptable amount is `2000`; the maximum is about `500000` (half a gigabyte).

V.A.6.d. **File handles**

PATER requires at least 70 of a system resource called "file handles." Otherwise the program may produce an error message `Not enough file handles`.

V.A.6.d.i.) Windows NT/XP/2000

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 installation folder (possibly named `C:\DNAVIEW`.)

The normal copy of `Config.NT` is located in the system directory — usually at `\WinNT\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!

V.A.6.e. Windows XP/NT/2000 startup glitch (recent)

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:

V.A.6.e.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`.

V.A.7. Printing from PATER (in Windows)

Printing from DNA·VIEW or PATER when running under Windows may require a trick.

V.A.7.a. Printing to an LPT port

> PATER only knows how to print to LPT1 or LPT2 (§II.G.4.h), which are traditional names for plugs on the back of the computer where printers used to be plugged in. PATER 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.

V.A.7.b. Printing to a network or USB printer from Windows XP (2000, NT, Vista)

> See **Connect USB printer**, §V.A.3.a.ii. (For the technically included, it invokes a `NET USE` command.)

V.A.7.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 (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 §II.G.4.h.
>
> Choose the path for the desired printer – e.g. \\YourServerName\PrinterName
>
> Check "Reconnect at logon"
>
> Click on OK

V.B. Running PATER under Windows

requires a 32 bit Windows system, or a virtualizer (such as VMWARE) or DOSBOX.

V.B.1. Startup

Usually click the **PATER** icon or choose PATER from the start menu.

V.B.2. Hardware security key

If you have a hardware security key, it must be plugged into the LPT1 port in the back of the computer for PATER to start or to run. If there is a printer normally plugged into LPT1, unplug the printer, plug in the security key and then plug the printer into the security key.

V.B.3. Annoying message

If, when trying to start PATER, Windows complains about COM1 and asks "**Close or Ignore?**", click "Ignore" and everything will work fine. This probably only happens on older PATER installations. See §V.A.6.e for the fix.

V.B.3.a. **Windows Lore**. Did you know?

When PATER is running, you can use ***alt-enter*** to toggle between full- and windowed- screen.

Alt-tab cycles among running Windows processes, including PATER.

V.B.3.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.

V.B.3.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".

V.C. PATER and Networks (recent)

PATER can be installed on the server and thereby accessed from every workstation.

V.C.1. Installation of PATER to the server

However, PATER 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 PATER 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\programs\DnaView\import`.

V.C.1.a. Establish a drive letter

First, you will need to define a drive letter – using the Windows facility called **Map network drive** – that equates to a folder on the server. Suppose the drive letter `P:` is available. For the example above, there are three possibilities:

map `P:` to		**Destination location** for PATER installation	comments
entire server drive	`\\servoir\`	`P:\programs\DnaView`	Entire server drive is accessible to work-stations
just PATER	`\\servoir\programs\DnaView`	`P:\`	Consumes a drive letter for just one application
in between ➜	`\\servoir\programs\`	`P:\DnaView` ➜	Normal compromise

Choose one of the above patterns – probably not the first one – and (from a workstation)

V.C.1.a.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**

V.C.1.b. Install the program

Run the PATER installer from any workstation after having mapped the network drive as above.

V.C.1.b.i.) Select the **destination location** (§V.A.1.a) following an example from the table above.

That completes installation to the first workstation

V.C.2. Install to other workstations

V.C.2.a. Map network drive as above for the next workstation

V.C.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 PATER installation file (§V.A.1.d.i).

V.C.3. File sharing among workstations

When several users run PATER 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**.

See the DNA·VIEW manual for enabling file locking and other networking customization. If you don't have DNA·VIEW then please call for help.

V.D. Moving PATER

Move the entire **DNAVIEW** folder in which **DNAVIEW\PATER** resides.

Use the same installer that updates **PATER** to create startup icons for the new location. (Optionally uncheck the part that would update, and be sure to check the option for startup icons.)

V.E. Web conference

is 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.

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