International Congress Series 1288 (2006) 474-476





LR-calculation of any kinship situation using a new graphical interface: Generate two or more hypotheses, draw the family trees and assign the DNA-profiles to person symbols

T. Dajda, M. Jung *

bj-diagnostik GmbH, Kerkrader Str. 11, 35394 Giessen, Germany

Abstract. We developed a graphical interface to allow an intuitive construction of alternative family trees defined as hypotheses. The novelty is that the family tree/hypothesis can be constructed like with a graphic design programme. The LR formulas and numerical results are given by the kinship algorithm which is applied to alternative family tree hypotheses. Person symbols are dropped with the computer mouse and the connection lines between the symbols are also set with a mouse. Null alleles or mutations can be treated. A simulation module allows calculations for any kinship scenario. The markers used and the number of persons can be varied in order to study the influence on the LR-value. It is a powerful tool for the planning of a relatedness DNA-analysis and is most useful for deficiency cases (how many markers, which persons should be tested). © 2005 Elsevier B.V. All rights reserved.

Keywords: DNA; Relatedness testing; Likelihood ratio; Kinship; Mutation; Silent null allele; Simulation; Deficiency case

1. Introduction

The software PaternityIndex is structured in two modules: one for calculation and one for simulation of kinship cases. The innovative part is a graphical interface, written in Java, with which hands-on time spent constructing the various hypotheses and the assignment of DNA-profiles to person symbols is greatly reduced. Hypotheses and DNA-profile operations can be executed intuitively by the user. This simplicity helps to avoid syntax errors in the set of hypotheses. DNA-profiles can be loaded from ASCII files or

^{*} Corresponding author. Tel.: +49 641 9446053; fax: +49 641 4994139. *E-mail address:* m.jung@bj-diagnostik.de (M. Jung).

 $^{0531\}text{-}5131/$ \otimes 2005 Elsevier B.V. All rights reserved. doi:10.1016/j.ics.2005.08.016



Fig. 1. A: Person symbol dropped with a mouse, B: symbols connected with a mouse, C: allele assignment.

from a database. DNA-profile ASCII files are generated by DNA-fragment analysis software. Using such files directly for calculation reduces the risk of typing errors during manual data entry. The software uses the kinship algorithm derived by Brenner [1]. The results are given in numerical values and algebraic formulas for each marker. Several alternative hypotheses can be compared with one click. The LR-value for the most probable hypotheses will then be determined in a pairwise hypothesis comparison.

2. Construction of hypotheses

The first steps of setting up a hypothesis are described in Fig. 1A–C. After the family tree is set up, one assigns the DNA profiles via *Person properties* to the symbols and gets a menu as in Fig. 1C. Data can be entered manually via drop down menus or automatically read from a file or database.

3. Example: Paper Challenge ISFG Paternity Testing Workshop 2004

DNA-data was given for BR, SI, M and Child to find out if AF can be the father of Child. The grandparents GM and GF had to be introduced to define BR, SI and AF as full siblings. Calculation for H2 was not under question in the paper challenge of the workshop but can easily be introduced into the scenario just by copying H1 and adding the parent connection line from BR to Child as in H2. The graphical setup of the hypothesis' family trees is given in Fig. 2.

Choosing the function *compare hypotheses* leads to the following result in Fig. 3. H2 is the least likely hypothesis and BR with 4 inconsistencies on THO1, D2S1338, TPOX and D5S818 can be excluded from paternity.



Fig. 2. Three hypotheses H0, H1 and H2, no DNA-data for GM, GF and AF (Alleged Father) was available.

omparison of hypothese Hypotheses' probabiliti	es Hypotheses' com	marison		1		
Compared hypotheses	Hypothesis probability	Exclusions on markers	LR vs. least likely	Probability vs. least likely		
H1 1. Alternative Hyp. H2 2. Alternative Hyp.	9.039899087932733 0.0	[NONE] [TH01, D2S1338, TPO	1			
Fi Fi	orce null allele		Prior likeli	hood:		
	0.0		0.5			
	Re	compute all Close	9			

Fig. 3. Comparison of H0 and H1 against the least likely hypothesis H2.

Markers 0 D8S1179 D21S11 D7S820 CSF1PO D3S1358	0 1	File: BR BR_04	P: BR	File: SI	P: SI			1		
D8S1179 D21S11 D7S820 CSF1P0 D3S1358		BR_04	12 14			File: M	P: M	File: Child	P: Child	Likelihood Ratio
D21S11 D7S820 CSF1P0 D3S1358			14 14	SI_04	12 13	M_04	14 14	Child_04	14 14	1.369941259
D7S820 CSF1P0 D3S1358		BR_04	31 32	SI_04	30 31	M_04	31 32	Child_04	31 32	6.418488499
CSF1P0 D3S1359		BR_04	11 12	SI_04	11 12	M_04	8 12	Child_04	8 1 2	1.414277968
D3S1359		BR_04	10 11	SI_04	11 12	M_04	10 11	Child_04	10 11	1.215387284
0001000		BR_04	15 16	SI_04	15 18	M_04	17 18	Child_04	15 17	1.483415881
TH01		BR_04	9.3 9.3	SI_04	8 9.3	M_04	67	Child_04	68	2.398349176
D13S317		BR_04	12 13	SI_04	11 12	M_04	12 12	Child_04	12 12	1.329649232
D16S539 <	< >	BR_04	8 9	SI_04	8 9	M_04	11 11	Child_04	8 11	17.407855678
D2S1338		BR_04	19 20	SI_04	19 20	M_04	19 20	Child_04	17 20	0.436127764
D19S433		BR 04	12 15	SI_04	13 14	M 04	13 16	Child 04	12 13	3.846153846
WVA		BR_04	15 17	SI_04	14 15	M_04	16 17	Child_04	15 16	3.061431029
TPOX		BR_04	88	SI_04	8 11	M_04	8 8	Child_04	8 11	1.167995274
D18S51		BR_04	12 22	SI_04	16 18	M_04	16 17	Child_04	17 22	39.682539683
D5S818		BR 04	9 10	SI_04	8 10	M_04	9 11	Child 04	11 11	0.219106047
FGA		BR_04	21 23	SI_04	21 22	M_04	23 23	Child_04	21 23	1.953249981
	-		1		<u>п</u>					•
Mutation	inter -	8.196	75%	90%	95%	99%	99.9%	99.99% 9	9 99955	10.000000 10.00
🔲 Force null allele		Prior likelihood:			Total likelihood ratio:			Total probability:		
		0.5		126782.549938958			99.999211254%			

Fig. 4. Numerical result for H0 against H1, it is most probable that AF is the father of Child.

Although AF could not be tested, it is highly probable that AF is the father of Child when testing H0 versus H1 as given in Fig. 4. The scenario in Fig. 2 now can easily be modified e.g. to study the influence on the quality of the result by removing SI, BR, or M from the family tree.

4. Concluding remarks

This paper describes a new graphical interface for DNA-relatedness testing. It is a tool to carry out calculations for complex kinship scenarios in very short time. But as a function of persons and loci tested, the question is how strong the support from an achieved LR value is. The interpretation of the LR value to the customer may consume much more time and effort than calculating the LR especially in case of smal LR values. In such cases the calculation and simulation functions of PaternityIndex can be used to plan the further extension of the analysis, such as to decide which other persons from the family tree and/or how many other markers should be typed to increase the LR value.

Reference

[1] C.H. Brenner, Symbolic kinship program, Genetics 145 (1997) 535-542.