Panel Discussion: Biostatistics

Statistical Considerations to the Methods Used in Paternity Testing

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As requested by the organiser, there were three topics, which hat to be dealt with in this contribution:

1) The method of Chakraborty and Ryman

2) Two recent "critical" publications by M. Aickin (1) and C.C. Li and A. Chakravarti (2)

3) The "Pros and Cons" of the methods presented in this panel

The method of Chakraborty and Ryman is based on the well known paternity index X/Y and was originally introduced into paternity testing by Schulte-Mönting and Walter. The method uses the Neyman-Pearson principle to compare the conditional distribution functions for a given test statistic.
In this case the test statistic is the paternity index X/Y and the conditional distribution functions for both hypotheses can be explicitly derived on the basis of the gene frequencies of the systems used for testing. Consequently the conditional tail probabilities α and β for the errors of the first and second kind given a decision threshold

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can be calculated, but it must be stressed that the total error of this procedure, which is

$$(1-P_0)\ \alpha\ +\ P_0\ \beta$$

also is a function of the prior probability P_0 for paternity. α and β are error rates conditional on the subsets V (cases with the true father) and $\overline{V}$ (cases with a man falsely alleged) (Baur and Rittner (3)). Chakraborty and Ryman use for comparison not the subset $\overline{V}$ but - with no justification - the subset $N\overline{V}$ (non-excluded non-fathers), thus reducing one subset trough increase of the number of genetic systems tested. The consequence is a redefined conditional error rate α' which increases to a large relative value, while the absolute error equals

$$(1-P_0)\ \ \alpha'\ \ (1-P_{E)}$$

with P_E equal to the exclusion chance of the tested systems. It is apparent that α' can have a large numerical value while the absolute error is minimal. Furthermore α' errors from different tests are not comparable unless exactly the same battery of systems has been used for testing.

With regards to the discussion of the two critical papers by Aickin (1) and Li and Chakravarti (2) it must be stated that as a consequence of the legal situation the use of statistical methods in paternity testing is relatively new in the United States in comparison to Europe. Due to lack of knowledge of the literature from the last thirty years (admittedley not all available in english) it seems that methods are reinvented as in the case of the tail probabilities of Chakraborty and Ryman, which were already discussed by Hummel as well as Koller and introduced by Schulte-Mönting and Walter. More serious, though, is the creation

of doubt with regards to a well established method, namely the use of the paternity index X/Y as the only statistic containing all the genetical information at hand.
Aickin (1) in his paper correctly defines X/Y as the likelihood ratio conditional on the two alternative hypotheses

$$\frac{X}{Y} = \frac{L\ (\text{obs. phenotypes/fatherhood})}{L\ (\text{obs. phenotypes/non-fatherhood})}$$

His first objection states that this likelihood ratio can not distinguish between different men with identical phenotype. He draws the conclusion that all statements consequently correspond not to a specific man but to all males with this given phenotype. This objection would have its merrit, if paternity testing was performed by way of screening all men of the given population giving equal prior probability to all of them ignoring the information given from the mother. His second objection concerns the assumptions for the calculation of the denominator

$$Y = L\ (\text{obs. phenotypes/non-fatherhood})$$

First of all this conditional likelihood depends on the ethnic background of the true father, which may not be well defined. The effect caused by possibly differing genefrequencies in differing ethnical or geographical populations is small, though, if a sizeable number of systems has been tested (as Aickin states himself). Furthermore he would rather redefine Y on the basis of a population of "plausible fathers", which is a correct point, if knowledge about such a group exists. But given this knowledge we are no longer dealing with a one man case and proper statistical handling will yield the correct conditional likelihood(s). Aickin's third objection is the one least understandable. On the basis of a constructed example he argues that in case of a system with two common codominant alleles 1 and 2 and

a rare silent allele s and a case of

Child:	1	Mother:	1
P 1:	1	P 2:	2

the likelihood ratio strongly supports the hypothesis of paternity for putative father P1 on the basis of the given genotype-phenotype relations, although in some cases P2 could have genotype 2-s and be the father of a child 1-s. This objection is not acceptable, because the likelihood ratio correctly quantifies this likelihood in relation to a 1-1 homozygous child with a 1-1 homozygous father.

More serious in the way of confusion is the paper of Li and Chakravarti (2), whose mayor objection is based on their statement that the ratio X/Y is not a likelihood ratio, but merely a segregation probability over a weighted average of segregation probabilities written as

$$\frac{P(C/MF)}{P(C/M)}$$

The original definition of the likelihood ratio conditional on the two alternative hypotheses (which they state themselves) is again

$$\frac{P\ (\text{obs. phenotypes/fatherhood})}{P\ (\text{obs. phenotypes/non-fatherhood})}$$

which is equal to

$$\frac{P(A) \times P(M) \times P(C/MA=F)}{P(A) \times \left[P(M) \times P(C/M)\right]}$$

with A being the phenotype of the alleged man and A=F sig-

nifying the condition, that he is the father, whereas in the denominator he has been picked at random. Simple cancelation of P(A) and P(M) in this well defined likelihood ratio reduces the computational effort to evaluate

$$\frac{P(C/M\ A=F)}{P(C/M)}$$

but in no way changes anything with regards to its original property as likelihood ratio. Their next objection is to the fact that $X/Y > 1$ in a given example for all non-exclusion phenotypes. This is true, but they do not state that $X/Y=0$ for the exclusions. Their third objection that a true probability of paternity should monotonically increase with increasing number of tested systems and no exclusion only holds by expectation over all phenotype classes and definitely is no necessity for each single non-exclusion phenotype.

After this sequence of (non acceptable) objections they proceed to state, that on the basis of N previous court cases with a theoretical exclusion chance P_E and an observed number of exclusions N_E the prior probability P_O can be estimated by

$$\hat{P}_O = 1 - N_E/NP_E$$

There statement that this parameter "may be easily estimated.... and we are surprised that no such investigation has been made until recently" again shows ignorance of numerous such investigations carried out during the last 20 years and in addition ignores the more powerful estimation of P_O by way of expectation maximization applied to the posterior probability as a function of P_O.

Their final proclamation of a new "method" to calculate a posterior probability on the basis of exclusion - non exclusion by way of

$$P_t = \frac{P_o}{P_o + (1-P_o)\,(1-P_E)}$$

is equal to

$$P_t = \frac{1}{1 + \frac{1-P_o}{P_o}\;\frac{1-P_E}{1}}$$

which is immediately identified as the bayesian approach by Essen-Möller on the basis of less information than available. Basically all that is proposed, is to lump the most powerful knowledge of the possible phenotypes together into the two subclasses of exclusion phenotypes and non-exclusion phenotypes and use this reduced information in the well established Essen-Möller approach.

As to the discussion of the methods presented in this panel it has been already pointed out that the tail probabilities used in the Neyman-Pearson approach are conditional and can only be converted to an overall error rate by introduction of the same prior probabilities used in the bayesian approach. The use of a tail probability conditional on the varying subset of non-excluded non-fathers is strongly objected and it seems that the standardized paternity index of Martin uses exactly this approach. Both methods seem to be based on the believe, that the statistical argument begins after there has been no exclusion, which by no means is justified, because all likelihood arguments are based on all possible phenotypes.

The final argument concerns the use of the exclusion chance P_E only, because it is not based on a prior probability,

in contrast to the use of the paternity index X/Y, which may or may not be converted to a posterior probability W by way of the bayesian approach using either a standardized or an estimated prior probability.
Whereas the argument that the decision rule "all non-excluded men are fathers" has a small error rate of

$$(1-P_O)\ (1-P_E) < 1-P_E$$

relative to all cases, given a powerful battery of tests, it should become obvious from table 1, that the likelihood ratio X/Y as well as W have the same or better properties (because they use the total information) and are applicable for all conceivable situations.

Admittedly, in case of a high exclusion chance (I), there is little difference in the information provided by W as well as P_E. Contrary, though, with a low exclusion chance (II) for the given mother-child combination the parameter W, which is based on the total genetic evidence, is far more powerful to differentiate and consequently the amount of error comitted is smaller, if based on W. In case of several non-excluded men (III) P_E is of no information at all, because it is equal for all of them, and only W correctly quantifies the difference due to differing phenotypes. In case of complex family situations (putative father not available for testing) the correct calculation of P_E may be extremely complex (IV) in contrast to X/Y and W, and in many situations $P_E = 0$ due to the structure of the given data (V) as for the example of information only from one of the putative father's parents. In both cases (IV and V) the likelihood ratio X/Y and W correctly quantify the often powerful information at hand.

As a consequence of this comparison there is no doubt, that the statistic W (or equivalently X/Y), which are applicable

by definition to all conceivable problems, should be the basis of the judges decision.
As conclusion I would like to cite J. Morris' remark - "If it's not broken let's not fix it" - but from previous experience I have little hope that this will happen.

	Kind of case	P_E	P_E vs. W (X/Y,PI)
I	One man not excluded	high	$P_E \sim W$
II	One man not excluded	low	$P_E < W$
III	Several men not excluded	not informative	$P_E < W$
IV	"Family" cases	P_E "complex"	$P_E < W$
V	"Family" cases	$P_E = 0$	$P_E < W$

" $\sim$ " almost equally informative

" $<$ " less informative

Table 1 Comparison of P_E and W (X/Y,PI) in different situations

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