THE BIAS OF THE DICHOTOMY EXCLUSION / NON-EXCLUSION AND THE EVIDENTIAL VALUE OF L (OR W)

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The aim of paternity expertises is to estimate the relative likelihood of mutually exclusive genealogical hypotheses, based on the results of a genetic analysis.

However, it is current practice to define it otherwise: the phenotypic evidence is the basis for a decision in terms of possible exclusion of one of the hypotheses; only if this aim is not attained, a statistical analysis is performed.

The purpose of this work is to demonstrate that this approach is not only grossly simplistic and inapplicable to a non negligible fraction of practical cases but also that it introduces implicitly a bias in the analysis of the problem, challenging the expert's neutrality.

DEFINITIONS

Let us call E to the **phenotypic evidence** obtained in paternity cases (blood group, enzymes, RFLPs, etc.). For these observations, a probability of occurrence can be calculated under the hypotheses of paternity (H1) or non-paternity (H2).

Thus a **paternity probability** (PP), has the form of a comparison between P(E/H1) and P(E/H2). For instance, in the **paternity index** (L), this comparison takes the simple form of a ratio.

The information flux in the process of paternity expertises is, therefore, the following:

(1)

$$\text{Phenotypic evidence} \xrightarrow{\text{statistical analysis}} 0 < \text{Paternity probability} < 1$$

Defining an **exclusion** as any E that contradicts the pre-established formal genetics hypothesis on the transmission rules, we will consider two types: a **1st. order exclusion** is said to occur when the hypothesis of paternity is not acceptable unless invoking mutation, whereas in the **2nd. order exclusion** the presence of a silent gene can also be presumed. When using the exclusion concept the information flux (1) is replaced by:

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Edited by W.R. Mayr

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(2)          ┌Exclusion (Non-paternity; PP=0)
Phenotypic   │
 evidence   -┤              statistical
             │               analysis
             └Non-exclusion ---------> Paternity Probability (0<PP<1)
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We will call **non-dichotomic** to the first approach and, obviously, **dichotomic** to the last. In the next section we will analyse the assumptions of each of them in order to be able to formulate an opinion on their relative convenience for paternity expertises.

DISCUSSION

The first question to rise when comparing the dichotomic and non-dichotomic approaches is to ask the legitimacy of accepting that an experimental result can assume probability values of one or zero under no matter what hypothesis. We obviously assume the only scientifically admissible attitude: that any explanation is always provisional.

It can be argued however that the reliability of some exclusions can be *practically* taken as one. This opinion would only be (even *practically*) acceptable if the dichotomic approach had the following properties: (a)reasonably error free; (b)not biased; (c)simplification of the analysis and (d)improvement of the understanding of the expert's opinion by laymen.

The first property is well known not to be observed; indeed, false exclusions (namely of the 2nd. order type) are intolerably frequent. Some authors, acknowledging this fact, advanced some rules to minimize the risk of excluding a true father. For instance, according to VALENTIN (1977) at least two exclusions must be observed, while CHAKRABORTY and RYMAN (1981) suggest that in doubtful cases a paternity probability should be calculated for the systems where paternity is considered compatible. In any case, a non-uniform statistic treatment of the data is advised.

Thus it seems that the only way to avoid a high risk of error is to stick to a statistically biased solution, where different weights are given to evidence classes (exclusion and non-exclusion results).

Concerning the simplification eventually introduced by a dichotomic approach, one must admit that in favourable cases, the number and/or the type of observed exclusions allow a fast and safe production of the expert's opinion. However in a far from negligible proportion of the cases the concept of exclusion produces the opposite effect, as outlined above.

Finally one could admit that even if the dichotomic approach was not, under the strict expert's point of view formally correct or practically advisable, it would be suitable for a satisfactory simplified version addressed to laymen. Unfortunately, in our opinion, this again is not true.

In fact, the formulation of the expert's opinion in terms of exclusion/non-exclusion has two serious drawbacks. On one hand, an opinion of exclusion of paternity, being given in categorical, non-probabilistic terms, gives the court the idea that the result is absolutely irrevocable. In more precise words, that no matter the progress in scientific knowledge or the future collection of further evidence on the case, the expert's opinion of exclusion would be the same, without gain or loss of confidence. Inversely, non-exclusion results being transmitted in a probabilistic form, it is legitimate to suppose that the court feels a rather different responsibility on the final decision in each type of cases.

Although this brief discussion would allow the immediate formulation of some conclusions, we think to be useful to devote a few lines to the recent controversies on the formulation of a statistic opinion.

Indeed, these controversies stem directly from the use of the dichotomic approach. We will take the proposition of LI and CHAKRAVARTY (1985) as the ultimate example of this strategy. These authors use a transformation of the *a posteriori* exclusion probability as a paternity probability (Pt), claiming to be superior to L or W. It has been already demonstrated that Pt is indeed poorer than L or W (BAUR *et al.*, 1986). We think that Pt is not only a less informative statistic when compared with L or W, but also that it is severely biased: the genetic information being only used in terms of exclusion/non-exclusion, (a) by definition, Pt is only usable in non-exclusion cases, preventing *a priori* its application to the opposite type of results. (b) all non-excluded men for the same mother/child pair have an equal Pt and (c) again by definition Pt cannot lower the established *a priori* value for paternity probability.

CONCLUSIONS

It is demonstrated that any methodology relying on the dichotomy exclusion/non-exclusion has serious and undesirable consequences in paternity expertises:

a. Formal inaccuracies

* non-uniformity of the statistic treatment of genetic data;
* incorrect assumption (or statement) that a paternity probability can assume a zero value (but not the unity).

b. Practical inadequacies

* creation and inability to cope with "difficult cases";
* risk of misunderstanding of the expert's role in paternity investigation.

The only way to avoid these problems is to stick to the time honoured methodology introduced by ESSEN-MÖLLER and not to use, at least when dealing with non-experts, the exclusion concept.

In practice that means a generalization of the proposal of GÜRTLER (1977) to the cases where non-conformity of the genealogical hypothesis with the genetic model cannot be alleviated by the presence of a silent gene but only by mutation. That is to say: L or W can be calculated in any cases and they are the only statistics that can deal in a uniform, unbiased manner with all kinds of genetic results.

Another corollary of this proposition is that a coherent efficiency criterion of genetic analysis for paternity investigation can no longer rely on the exclusion probability, but on the difference between the means of paternity probabilities among fathers and non-fathers. The description of the derivation of the algorithm is deferred to another paper.

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ACKNOWLEDGEMENT

This work was partially granted by Calouste Gulbenkian Foundation.