

The Decision Theory of Paternity Disputes: Optimization Considerations Applied to Multilocus DNA Fingerprinting

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ABSTRACT: The solution of paternity disputes using results from scientific analyses is studied from a decision-theoretical viewpoint. Two alternative approaches to decision making, the so-called 'Bayes' and 'Minimax' strategies, are described and discussed. If prior probabilities of paternity are exactly known, then Bayes decisions are (a) independent of the source of evidence and (b) optimal with respect to average losses caused by wrong decisions. However, it is concluded that Minimax decisions, which depend upon the employed test system but not upon prior probabilities, are more appropriate in paternity cases if equal prior good will towards disclaimed children and alleged fathers is demanded. It is further demonstrated that, when major evidence about paternity comes from multilocus DNA fingerprinting, prior probabilities must be known quite accurately for Bayes decisions to be superior with respect to average losses. Finally, we are able to show that 'quasi' Bayes decision making, that is, adopting a neutral prior probability of 0.5 but leaving thresholds for decision making unchanged, coincides with Minimax decision making if multilocus DNA fingerprinting is employed.

KEYWORDS: pathology and biology, paternity disputes, DNA, DNA fingerprinting

If decisions must be made in the face of uncertainty, common sense tells us that we should acquire in advance as much information as possible about the object in question. In paternity cases, where the judge has to decide whether or not a given man shall be deemed to be the biological father of a child, genetic experts may contribute such information. Phenotypic features that are at least in part genetically determined present themselves in nonrandom relations between parents and their offspring. The question to answer is whether any observed phenotypic relationship represents a proof of paternity or whether it could also be due to chance alone. Any test system should aim to provide the judge with convincing evidence in favor of one of these two alternatives but usually an unequivocal answer cannot be given. Evidence is thus phrased in terms of probabilities, likelihoods and odds, allowing one at least to quantify the uncertainty left.

Methods and Results

The Likelihood Ratio

In what follows, we shall only deal with 'one-man-cases,' that is, cases where a decision has to be made merely between paternity and nonpaternity. Decision making with more

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than one putative father involved, or with incomplete information ('deficiency cases') will be subject to future work. The central quantity in which phenotypic information from individuals involved in a paternity case is condensed, is the 'likelihood ratio' or 'paternity index' $L = Y/X$. Here, X and Y denote the likelihoods, or conditional probabilities, of the observed phenotypic pattern under the assumption of paternity and nonpaternity, respectively. The likelihood ratio is thus a measure of how much more likely the observed phenotypes are if the defendant is, in fact, not the true father (for a detailed discussion of L , see [1,2]). A small value of L consequently argues in favor of paternity whereas a high value provides evidence against it. Thus, a major goal for rational decision making should lie in finding a meaningful threshold, L_o , such that paternity is rejected whenever L is larger than L_o , and a decision is made in favor of true paternity if L is smaller than L_o . It should be noted in this context that calculation of X is comparatively easy if an appropriate statistical model for the transmission of phenotypic features has been established. Likelihoods under the assumption of nonpaternity, however, are more problematic. The true likelihood Y would equal the average X for males from a population containing all other putative fathers. This figure can scarcely be determined in practice and must be approximated. This notwithstanding, and especially if rare or highly polymorphic characters are examined, estimates of the required population genetic parameters (for example, allele frequencies) may be rather inaccurate or difficult to obtain.

The Losses Bound up with Wrong Decisions

After all information relevant to a decision has been gathered, decision making depends upon the 'losses' that are assigned to possible wrong decisions [3,4]. (This principle cannot be denied although it may seem callous to talk about losses in the context of paternity disputes. Decision making does not depend upon probabilities or likelihoods alone, but also requires relative weighting of the different alternatives involved, in order to be optimal in some respect. To most of us, odds of 1:100 for a shower appear too small for taking an umbrella with us, whereas the same odds for a gun aimed at us being loaded puts us to flight). At least intuitively, the judge has to have an impression, too, of how much worse it would be to let a true father get off as opposed to deeming a defendant to be the father when in fact he is not. Only for the purposes of formal analysis shall we denote the losses bound up with these errors as A (rejecting paternity although true) and B (accepting paternity although false). Both figures are unitless, and we shall see later that only their relative ratio, that is, A/B , is essential for decision making.

The Prior Probability of Paternity

In a paternity dispute there is commonly further, though unquantifiable evidence that might need to be considered in decision making. If, for example, the defendant is able to convince the judge that he had not seen the mother in the year before the birth of the child, then even a very small likelihood ratio can be outweighed by this fact. Consideration of such points is equivalent to the question about how to deal with prior probabilities in the process of decision making. Prior probabilities for paternity, P , and nonpaternity, $1-P$, may either be purely intuitive or may stem from empirical studies. Although empirical figures appear to be scientifically substantiated, their relevance to an individual case and their reliability may be questioned as much as for intuitive predicates.

Bayes and Minimax Decisions

Optimization of decision making requires certain criteria. If one and the same kind of decision has to be made rather often, then it appears logical to require that the *average* loss caused by wrong decisions should be minimal. If, however, the decision is made only once or seldomly then we shall probably be more interested in keeping the *maximum* loss expected from different scenarios as small as possible. A life insurance company, with its premium well adjusted to the prior probability of claim, will usually conclude a contract with any interested client. Although the company's loss is large in an individual case, their business runs rather profitably because the overall number of clients is large. A single client, although facing the same prior probabilities, will also decide in favor of the contract because his loss would be unbearable in case of a mishap.

The previously mentioned different requirements are fulfilled by two strategies studied in statistical decision theory, the 'Bayes' and the 'Minimax' principle. The essential difference between these two approaches is that decision making according to the Bayes strategy involves prior probabilities whereas the Minimax strategy does not. The question of whether decisions about paternity cases should be made according to Bayes or Minimax principles is of both theoretical and practical interest, and it will be demonstrated in the following sections under which circumstances each approach would be the more appropriate. The implications of these considerations will then be discussed in detail for the application of multilocus DNA fingerprinting in paternity testing. Here, as we shall see, adopting equal prior probabilities will usually convert Bayesian into Minimax decisions.

The Optimal Thresholds for Decision Making

Let L_o be a given threshold for the likelihood ratio. For the sake of simplicity, we shall abbreviate the probability of $L > L_o$ for true fathers as $\alpha(L_o)$, and the probability of $L < L_o$ for non-fathers as $\beta(L_o)$. These figures are usually referred to as the 'error rates' bound up with the threshold L_o . If we decide in favor of paternity whenever L is smaller than L_o , and against paternity if $L > L_o$, then the loss expected among cases where true fathers are tested is

$$v_1 = A \times \alpha(L_o)$$

and

$$v_2 = B \times \beta(L_o)$$

for nonfathers.

Decision making according to the Minimax principle means that the maximum of both, v_1 and v_2 , should be minimal. This is achieved exactly when $v_1 = v_2$, because increasing L_o in the case of equality would reduce v_1 but increase v_2 and, therefore, the maximum of the two. Similarly, any decrease of L_o would reduce v_2 but increase v_1 , so that the optimal threshold for Minimax decisions is implicitly defined by

$$A \times \alpha(L_o) = B \times \beta(L_o)$$

or

$$A/B = \beta(L_o)/\alpha(L_o).$$

This decision rule is also intuitively apparent. If unjustified rejection of paternity is regarded as 100 times less harmful than deeming a man to be the father when he actually is not, then it also seems fair to let 100 true fathers get off before doing a single nonfather an injustice, irrespective of prior probabilities. In practice, exact determination of L_o is feasible for a ratio A/B if the distributions of L , given paternity and nonpaternity, are known. If L further follows a continuous, or approximately continuous distribution, then a unique L_o can be found.

When deciding according to the Bayes principle, we want to determine L_o such that the average loss

$$\begin{aligned} v' &= P \times v_1 + (1 - P) \times v_2 \\ &= P \times A \times \alpha(L_o) + (1 - P) \times B \times \beta(L_o) \end{aligned}$$

is minimal. $\alpha(L_o)$ was defined as the probability of $L > L_o$ for true fathers and, therefore, equals the sum of X values taken over all imaginable cases with $L > L_o$. Consequently,

$$P \times v_1 = P \times A \times \alpha(L_o) = \Sigma P \times A \times X$$

where summation is over all cases with $L > L_o$, and

$$(1 - P) \times v_2 = (1 - p) \times B \times \beta(L_o) = \Sigma(1 - P) \times B \times Y$$

where summation is over all cases with $L < L_o$. This means that cases in which a decision is made in favor of paternity contribute

$$(1 - P) \times B \times Y$$

to v' , whereas cases in which decisions is made against paternity contribute

$$P \times A \times X$$

Casewise contribution to v' would hence be smallest if a decision were made in favor of paternity for cases with

$$(1 - P) \times B \times Y < P \times A \times X$$

and against paternity for cases with

$$P \times A \times X < (1 - P) \times B \times Y.$$

These criteria are equivalent to

$$\begin{aligned} L &= Y/X < P \times A / [(1 - P) \times B] \text{ and} \\ L &= Y/X > P \times A / [(1 - P) \times B]. \end{aligned}$$

Hence, we should take

$$L_o = P \times A / [(1 - P) \times B].$$

The optimal threshold for decision making following Bayes principles is, thus, independent of the likelihood distributions, but depends only on the losses and the prior probability of paternity.

Minimax and Bayes thresholds, and therefore average losses bound up with the two strategies, usually coincide for only one prior probability, say P_o , depending on the likelihood distributions. Since the average loss is always larger for Minimax than for Bayes decisions, the Bayes decision for P_o yields, of course, the maximum average loss of all prior probabilities. Therefore, a Minimax decision can be regarded as a Bayes (and therefore optimal) decision under a worst case scenario, that is, assuming the most disadvantageous, or 'worst,' prior probability P_o . As evident from the name, the MINimum loss (from the Bayes decision) attains its MAXimum for P_o .

The Probability of Paternity

The Bayes criterion

$$L = Y/X < L_o = P \times A / [(1 - P) \times B]$$

is equivalent to

$$W = P \times X / [P \times X + (1 - P) \times Y] > B / (A + B)$$

where W equals the posterior probability of paternity, calculated from Bayes' formula. W can be interpreted as the proportion of true fathers seen by the judge among a large series of identical cases. A Bayes decision can thus be made on the basis of the posterior probability of paternity alone. If W exceeds a certain limit, depending on the relative loss ratio only, then decision should be made in favor of paternity, otherwise paternity must be rejected.

It should be noted that the probabilistic interpretation of W mentioned above is only admissible if P equals the true prior probability of paternity. This, however, cannot be claimed in general for the commonly used standardized or 'Essen-Möller' probability of paternity, W_s , calculated on the basis of $P = 0.5$ [5] W_s as such is a transformation of the likelihood ratio, and decision making strategies based solely on W_s and thresholds that are independent of the test system will usually be less than optimal. Considerations as carried out above for Bayes decisions do not obviously apply unless, in fact, $P = 0.5$. On the other hand, decisions based on W_s can also not be expected to coincide with Minimax decisions: The loss ratio A/B is independent of the test system, so that the threshold for a Minimax decision will actually depend on the likelihood distributions and, thus, the analytical methods employed.

Decisions Based on Multilocus DNA Fingerprints

Whether a Bayes decision is optimal depends on knowledge about the true prior probability of paternity. It may well be that the average losses bound up with Bayes decisions are larger than those of the corresponding Minimax decisions if the estimate of the prior probability is too inaccurate. This phenomenon shall now be examined in detail for decisions made on the basis of so-called 'multilocus DNA fingerprints.'

For the generation of a DNA fingerprint, an individual's DNA is digested using specific restriction enzymes, size separated by gel electrophoresis, and hybridized to a comparatively short, labelled DNA probe. An autoradiographic band appears at a specific position of the DNA fingerprint if cleavage of the DNA yields at least one fragment of

corresponding electrophoretic mobility that hybridizes to the probe used. Among the test systems applied to DNA typing in paternity disputes are single locus probes, pin pointing only one site of variability in the human genome, and multilocus probes, capable of detecting polymorphism at many DNA loci at a time. Results of multilocus DNA fingerprinting can be regarded as a series of informational bits, depending upon either absence or presence of a band at the analyzed positions of an autoradiograph. While typing results for single locus probes can be analyzed as codominant Mendelian traits, formal genetic analysis of hybridization patterns derived by multilocus probes is more complicated.

Recently, we have presented a new approach to the statistical analysis of multilocus DNA fingerprints, allowing the calculation of joint likelihoods, X and Y , for the multilocus DNA fingerprints of mother, child, and putative father [6]. These likelihoods depend mainly on the number of separable gel positions and on the probability, x , of presence of a band at each of them. Two sets of parameters rather typical of the real life situation with multilocus DNA fingerprints are $x = 0.25$ on 60 positions [7], and $x = 0.15$ on 120 positions [8]. Likelihood distributions and error rates assuming these parameters were derived from the simulation of 5 000 000 cases of paternity and nonpaternity, respectively. Simulation had to be employed here because exact calculation or even approximation of the likelihood distributions is scarcely feasible at their extremes. The results are presented in Fig. 1 in the form of cumulative distribution functions for the \log_{10} of the likelihood ratio L . Distribution functions corresponding to paternity were generated by including in the trios those fathers that were utilized to simulate the offspring DNA fingerprints. Nonpaternity was simulated by including another individual with a 'randomly' generated DNA fingerprint. For $x = 0.25$ on 60 positions, 99.73% of fathers as opposed to 0.63% of non-fathers exhibit a \log -likelihood ratio less than zero, corresponding to $L = 1$. For $x = 0.15$ on 120 positions, these rates improve to 99.93% and 0.08%, respectively.

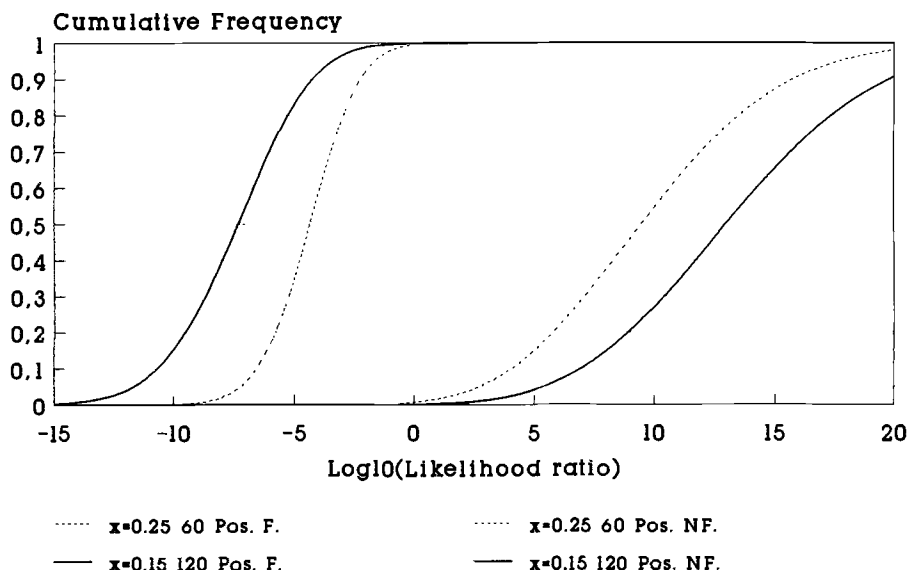


FIG. 1—The phenotype likelihoods for multilocus DNA fingerprints of father-mother-child trios. The cumulative distribution of the \log_{10} of the likelihood ratio Y/X (nonpaternity vs. paternity) is given. Left curves refer to trios with true fathers included (F.); the two curves on the right side refer to cases where nonfathers (that is, 'random' individuals) are included in the trio (NF.). x : band-sharing probability, Pos.: separable gel positions.

If a judge pretends to know the correct prior probability and tries to make optimal (that is, Bayes) decisions, then a threshold of 0.99 for the probability of paternity is equivalent to an implicitly assigned loss ratio of 1:99. In general, if Z is the threshold for W , then the assumption of optimality implies

$$A/B = Z/(1 - Z).$$

Let us adopt $A/B = 1:99$ which corresponds to a commonly accepted limit of 0.99 for W [5]. The appropriate Minimax thresholds, calculated from the likelihood distributions such that

$$A/B = \beta(L_o)/\alpha(L_o),$$

are $L_o = 0.0117$ for $x = 0.25$, and $L_o = 0.0132$ for $x = 0.15$. Use of these thresholds is equivalent to adopting limits of 0.9884 and 0.9870, respectively, for the standardized probability of paternity, W_s . This means that paternity should be accepted in a Minimax decision if W_s exceeds 0.9884 in the case of $x = 0.25$ on 60 positions, or if W_s exceeds 0.9870 for $x = 0.15$ on 120 positions. It should be noted that both thresholds are rather close to 0.99, the limit of W assumed for optimal decision making here. This phenomenon reflects that both 'worst' prior probabilities, $P_o = 0.538$ for $x = 0.25$ and $P_o = 0.566$ for $x = 0.15$, are close to 0.5, which in turn is used for the calculation of W_s . The above-mentioned findings have a rather curious implication. If the decision (i) is based upon W_s as derived from multilocus DNA fingerprints, and (ii) if 0.99 is regarded as a suitable threshold for W in cases where the true prior probability of paternity is known, then a Minimax decision is made, in fact, if this threshold is applied to W_s .

We next want to demonstrate how accurate the prior probability of paternity is required to be known, in order for the Bayes decision to be superior to the Minimax decision. Therefore average losses bound up with Bayes and Minimax decisions on the basis of multilocus DNA fingerprints were calculated, now assuming $A = 1$ and $B = 99$. Average losses were derived for a variety of true vs. assumed prior probabilities, respectively, and the results are presented in Fig. 2. Shaded regions correspond to pairs of prior probabilities for which the Bayes criterion including the assumed prior probability yields smaller average losses than the Minimax criterion. White areas represent pairs for which the Minimax decision is superior with respect to average losses. The straight line dividing both sections corresponds to the 'worst' prior probability, which is a rather logical property. Obviously, the average loss is proportional to the difference between assumed and true prior probabilities. Thus, if the true prior probability is smaller than P_o , assuming P larger than P_o yields average losses larger than the Minimax strategy. A similar argument applies if the true prior probability is larger than P_o . Although actual values of losses had to be adopted for these considerations, the results depend again only on the relative ratio A/B .

As can be inferred from Fig. 2, the prior probability of paternity must be known quite accurately for a Bayes decision to be superior. This is especially the case if the true value is close to the 'worst' prior probability. The latter, as has been demonstrated above, depends on both the likelihood distribution (that is, the test procedure) and the loss ratio. If the loss ratio increases, so does the 'worst' prior probability. For example, if $A/B = 0.999$ then P_o is 0.593 ($x = 0.25$ on 60 positions) and 0.714 ($x = 0.15$ on 120 positions), respectively.

Conclusions

The use of prior probabilities in decision making about paternity cases has been a subject of theoretical debate [9-13]. We set out in this paper to apply statistical decision

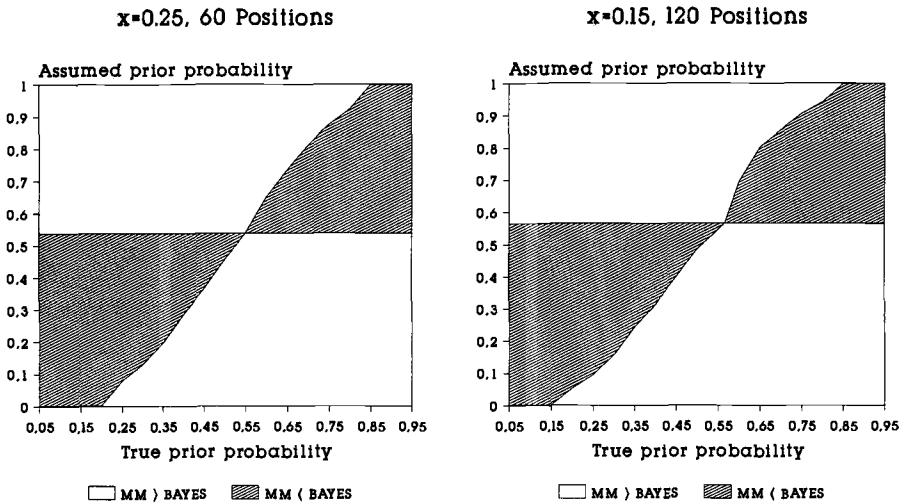


FIG. 2—Decision making in paternity cases using multilocus DNA fingerprints. Shaded regions correspond to pairs of true and assumed prior probability of paternity, respectively, for which average losses bound up with Bayes decisions are smaller than with Minimax decisions ($MM < BAYES$). x : band-sharing probability.

theory in order to study this problem. This approach requires that, at least intuitively, a judge must be aware of how much worse it is to do a non-father injustice than letting a true father get off. These relative losses must form the basis of decision making. The considerations outlined above show that, in case the prior probabilities of paternity were exactly known, they must be considered if the judge intends to minimize the average losses bound up with wrong decisions.

However, keeping the average losses at their minimum is mainly in the interest of the court, but this may not be so for the children seeking their fathers or the defendants denying paternity. An alleged father, for example, may justly ask why he should be at a disadvantage as compared to others only because prior odds in favor of paternity are higher under certain circumstances which apply to his case. Modifying results from laboratory testing by prior valuations means falsifying their probative weight. It cannot seriously be claimed that a knife says more about a case of murder, merely because it has been found in a sinister residential area. Minimax criteria do not pay regard to prior probabilities but 'let the facts tell their own tales,' and therefore appear to be more appropriate. In order to let the judge account for whatever his own implicit loss ratio A'/B' is, the results experts provide to the court should also include error rates, $\alpha(L_a)$ and $\beta(L_a)$ in addition to the actual L_a . For example, if

$$\beta(L_a)/\alpha(L_a) > A'/B' = \beta(L_o)/\alpha(L_o)$$

then this implies $L_a > L_o$, and a Minimax decision would be made against paternity. However, whether a request of equal prior good will towards all disclaimed children and all alleged fathers is legitimate cannot be answered by theoretical considerations.

Decisions based on Minimax criteria are not optimal with respect to average losses, unless the true prior probability is close to the so-called 'worst' prior probability. However, we have demonstrated that for a broad range of realistic prior probabilities [14-16] this may actually be the case. Thus, even if 'average' justice is required in the sense of small average losses, Minimax decision making is appropriate. Finally we noted

that, if multilocus DNA fingerprints are employed for paternity testing, adopting equal prior probabilities to take (intuitively) a standpoint of neutrality is equivalent to making a Minimax decision. Thus, at least for this test system, a discussion as to whether assuming $P = 0.5$ is justified for the calculation of paternity probabilities is meaningless. Whether such a statement also holds for other test procedures remains to be determined.

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