

**Inference in Cases of Disputed Paternity**

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### Abstract

Blood testing for hereditary factors is being used increasingly in paternity cases to infer that a particular man is the father. Geneticists calculate a probability of paternity using Bayes's theorem, making various assumptions about the genetic factors used and the other evidence in the case. These assumptions are criticized and the role of Bayes's theorem in a legal setting is discussed. The role of the forensic statistician in helping a court combine quantitative genetic evidence with nongenetic evidence is described. The effects of statistical errors and laboratory errors are discussed.

Key words: Bayes theorem, Blood groups, Blood tests, Errors in testing, Genetic polymorphism, Genotype, Human leukocyte antigen, Independence, Likelihood ratios, Paternity, Paternity index, Phenotype, Plausibility of paternity, Probability of exclusion, Probability of paternity, Reference population, Reporting accuracy, Random man, Silent gene

## §1. Introduction

There are two important kinds of evidence in cases of disputed paternity: blood tests for hereditary factors and testimony concerning sexual intercourse between the mother and alleged father. If blood tests exclude an alleged father then the case usually proceeds no further. If an alleged father is not excluded then the question of sexual intercourse becomes central. In many courts the particulars of blood tests play an increasingly important role in this setting.

Given the genetic makeup of parents, that of an offspring has a particular probability distribution. The problem of inference in cases of disputed paternity is to decide which genetic structure produced a given result. So it is a typical problem in statistical inference.

The basic statistical tool for problems involving "inverse probabilities" is Bayes's theorem. While its use for scientific inference is controversial among statisticians, it has been readily adopted by geneticists for purposes of genetic counseling and by some in cases of disputed paternity. Its application will be discussed in Sections 3 and 4. The approach we describe is used by many authors and, in particular, Salmon and Salmon (1980).

The procedure followed varies considerably from one laboratory to another. The following is a scenario that some facilities follow in obtaining and reporting results of blood tests. First, a blood sample is taken from the alleged father. At the same time he is identified and photographed; his thumbprint may be taken. When the samples are drawn from the mother and child, the mother is asked to verify that

the man in the photograph is indeed the alleged father.

The child, mother, and alleged father are compared with respect to various blood group genetic systems. The systems chosen vary greatly from one laboratory to another, and we have records of cases which show that different systems can be chosen by the same laboratory. Most states employ tissue-typing for human leukocyte antigen (HLA). This latter possibility will be discussed in Section 6, but our examples deal with red blood cell antigens and enzymes and serum proteins.

The testing laboratory prepares a report with its findings. This report (which is similar to Table 9 in Section 4) lists the various blood factors tested and gives measures of "likelihood of paternity". A main purpose of this paper is to elucidate and critically examine the assumptions and ensuing calculations in such a report. This is done in Sections 4 and 5. Another purpose is to describe an appropriate method of presentation for quantitative evidence in the presence of other kinds of evidence.

The focus of this article is the role of the forensic statistician in paternity cases, particularly as it applies to educating lawyers and communicating with juries. This role revolves around the use of Bayes's theorem: How are likelihoods calculated? Are blood group factors and other genetic polymorphisms independent? What is the effect of classification errors? How are prior probabilities assessed and interpreted? How should nongenetic evidence be combined with genetic polymorphisms? What are appropriate reference populations? The paper is written mainly

for statisticians but the discussion is kept at a level appropriate for many nonstatisticians. Some of the ideas and criticisms of the usual approach are similar to those of Ellman and Kaye (1979) and Aickin and Kay (1982).

A short summary of the necessary genetical background is given in Section 2. The interested reader is referred to Elandt-Johnson (1971) for a much more extensive presentation.

## §2. Background Genetics

Until recently, genetic testing was used in paternity suits to exclude an alleged father. If the testing procedure did not exclude it was regarded as largely irrelevant. These tests involved mainly the red cell antigens of the blood.

These antigens are inherited substances present on the surface of the cells that have the capacity to induce the production of other substances termed antibodies. Antibodies in turn react with antigens; it is assumed that the serum of an individual in which the red blood cells are suspended, does not possess nor can produce antibodies against its own antigens. So a blood group system is a property of the individual's serum by which the antigen is recognized.

Consider a hypothetical system, say GH. An individual having blood group G then will not produce anti-G (antibody) but can possess or produce anti-H, say, an antibody to an alternative blood factor H belonging to the same system. A battery of diagnostic tests are available to determine an individual's blood group.

The logical basis for an exclusionary result depends on a relatively simple genetic construct. An individual's genotype, or

hereditary configuration, for a particular inherited antigen consists of two out of a number of alternative forms of the hereditary unit, called alleles, one from each parent. For example, for the simplest system with only two possible alleles, say, G and H, an individual's genotype will be one of GG, GH, and HH. If the parents are GG and HH then all offspring must be GH, a G from the first parent and an H from the second. On the other hand, if both parents are GH then the child can be any of the three genotypes. Since each child inherits one of its two letters (alleles) from each parent, certain men can be logically excluded if the mother's and child's genotypes are known. Table 1 lists these exclusions. No alleged father of genotype GH can be logically excluded under any genotypic combination of mother and child.

Table 1: GH Paternal Exclusion

<u>Child's Genotype</u>	<u>Mother's Genotype</u>	<u>Excluded Father's Genotype</u>
GG	GG	HH
GH	GG	GG
HH	GG	Mother excluded!
GG	GH	HH
GH	GH	None
HH	GH	GG
GG	HH	Mother excluded!
GH	HH	HH
HH	HH	GG

When the alleles are codominant, as in the MN red cell antigen case, it is possible to establish the genotype of any subject. When one allele dominates another it is only possible for the test to establish the phenotype--the physical expression of a

genotype which may be influenced by environmental conditions, in this case the presence or absence of the dominant allele.

The ABO red blood antigen system is basically a three allele system (though modern methods are able to discern at least two variants of the A allele,  $A_1$  and  $A_2$ ). It is a mixed system since A and B are codominant and both dominate O. Hence an individual whose phenotype is A has genotype either AO or AA; similarly for B. However, the phenotype of the codominant case AB and also that of the recessive case O completely determine the corresponding genotypes AB and OO.

As an example consider the following phenotypic frequencies<sup>1</sup> for the ABO system in a sample of  $n=6004$  white Californians reported by Grunbaum, et al. (1978):

$$n_O = 2891, \quad n_A = 2149, \quad n_B = 724, \quad n_{AB} = 240.$$

Let  $p_O$ ,  $p_A$ ,  $p_B$ ,  $p_{AB}$  be the population phenotypic relative frequencies and  $g_O$ ,  $g_A$ ,  $g_B$  the allelic frequencies. Then using the Hardy-Weinberg law:<sup>2</sup>

$$(g_O + g_A + g_B)^2 = p_O + p_A + p_B + p_{AB},$$

it follows that

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<sup>1</sup>They actually report relative frequencies to three decimals--the frequencies we give are approximated from their figures.

<sup>2</sup>This law depends on the assumption of random mating which, strictly speaking, assigns to each individual of one sex in a population an equal chance of being a partner to a given mate of the opposite sex which is interpreted as a mating between unrelated individuals. The transmission of the inherited units, one from each parent, is then presumed to be statistically independent of the particular blood systems to which the law is applied.

$$p_0 = g_0^2$$

$$p_A = g_A^2 + 2g_0g_A$$

$$p_B = g_B^2 + 2g_0g_B$$

$$p_{AB} = 2g_Ag_B$$

One estimate of  $p_0$  is  $n_0/n$  and in turn  $g_0$  can be estimated to be  $\sqrt{n_0/n}$ . But these estimates are not efficient. Assuming the sample is random, the likelihood function of  $g_0, g_A, g_B$  (where  $g_0 + g_A + g_B = 1$ ) is proportional to

$$g_0^{2n_0} (g_A^2 + 2g_0g_A)^{n_A} (g_B^2 + 2g_0g_B)^{n_B} (g_Ag_B)^{n_{AB}}.$$

Methods for numerically determining the maximum likelihood estimates of the various allelic, genotypic, and phenotypic frequencies are readily available; c.f. Elandt-Johnson (1971). The maximum likelihood estimates of the allelic relative frequencies for the above data are  $\hat{g}_0 = 0.692$ ,  $\hat{g}_A = 0.224$ , and  $\hat{g}_B = 0.084$ . The corresponding estimates of genotypic and phenotypic relative frequencies are given in Table 2. (These phenotypic estimates differ from those of Grunbaum, et al. (1978) who apparently used maximum likelihood estimates for the allelic frequencies but the sample proportions for the phenotypic frequencies.)



Table 2: ABO System Proportions  
(Estimates for a White California Population)

Phenotype	O	A		B		AB
Genotype	OO	AO	AA	BO	BB	AB
Genotypic Frequency	$\frac{2}{g_O}$ (0.479)	$2g_Og_A$ (0.310)	$\frac{2}{g_A}$ (0.050)	$2g_Og_B$ (0.116)	$\frac{2}{g_B}$ (0.007)	$2g_Ag_B$ 0.038
Phenotypic Frequency	0.479	0.360		0.123		0.038

Logical phenotypic exclusions for the ABO system are given in Table 3. This table also gives estimates of the proportion of men who are excluded from paternity on the basis of ABO blood type.

Table 3: ABO System Paternal Exclusion

Child's Phenotype	Mother's Phenotype	Excluded Paternal Phenotypes	Proportion of Excluded Males	Estimate for White Californians
O	O	AB	$P_{AB}$	0.038
A	O	O, B	$P_O + P_B$	0.062
B	O	O, A	$P_O + P_A$	0.839
AB	O	Mother excluded!	--	--
O	A	AB	$P_{AB}$	0.038
A	A	None	0	0.000
B	A	O, A	$P_O + P_A$	0.839
AB	A	O, A	$P_O + P_A$	0.839
O	B	AB	$P_{AB}$	0.038
A	B	O, B	$P_O + P_B$	0.602
B	B	None	0	0.000
AB	B	O, B	$P_O + P_B$	0.602
O	AB	Mother excluded!	--	--
A	AB	None	0	0.000
B	AB	None	0	0.000
AB	AB	O	$P_O$	0.479

The estimated probability of exclusion for the ABO system in a white population is the average of these estimates. This average is with respect to the probabilities of the various child/mother phenotype combinations. These are given in Table 4. For example, for "child: O; mother: O" the calculation of  $g_0^3$  proceeds as follows: The probability of "mother: O" is  $g_0^2$ ; if the mother is O the child inherits an O allele from her, the probability that the other allele is O is simply  $g_0$ . For the population of white Californians the estimated probability of exclusion on the basis of ABO is 0.181. Salmon and Salmon (1980) suggest that only systems with high exclusion probability be used in paternity cases. There are many known systems of red cell antigens. The first system for exclusion of paternity based on blood group evidence was used more than 40 years ago, although early tests were only used to exclude putative fathers. As the number of red blood groups used in such tests increased it was realized that continual nonexclusion enhances the possibility of paternity. As still other genetic polymorphic (multiple allelic forms) systems, including serum protein groups and white cell antigens such as human leukocyte antigen (HLA), were introduced into this legal enterprise, systematic efforts were made to determine a canonical measure of the "likelihood of paternity" of an alleged father. In many European and U.S. courts it has now become standard practice to accept genetic evidence in terms of a "probability" that an alleged father is indeed the father. This concept is developed in the next section.

Table 4: Proportion of Mother/Child Phenotype Combinations

<u>Child's Phenotype</u>	<u>Mother's Phenotype</u>	<u>Combination Proportion</u>	<u>Estimate for White Californians</u>
O	O	$\epsilon_0^3$	0.331
A	O	$\epsilon_0^2 \epsilon_A$	0.107
B	O	$\epsilon_0^2 \epsilon_B$	0.040
AB	O	0	0
O	A	$\epsilon_0^2 \epsilon_A$	0.107
A	A	$\epsilon_A^3 + 3\epsilon_0 \epsilon_A^2 + \epsilon_0^2 \epsilon_A$	0.223
B	A	$\epsilon_0 \epsilon_A \epsilon_B$	0.013
AB	A	$\epsilon_A^2 \epsilon_B + \epsilon_0 \epsilon_A \epsilon_B$	0.017
O	B	$\epsilon_0^2 \epsilon_B$	0.040
A	B	$\epsilon_0 \epsilon_A \epsilon_B$	0.013
B	B	$\epsilon_B^3 + 3\epsilon_0 \epsilon_B^2 + \epsilon_0^2 \epsilon_B$	0.055
AB	B	$\epsilon_A \epsilon_B^2 + \epsilon_0 \epsilon_A \epsilon_B$	0.015
O	AB	0	0
A	AB	$\epsilon_A^2 \epsilon_B + \epsilon_0 \epsilon_A \epsilon_B$	0.017
B	AB	$\epsilon_A \epsilon_B^2 + \epsilon_0 \epsilon_A \epsilon_B$	0.015
AB	AB	$\epsilon_A^2 \epsilon_B + \epsilon_A \epsilon_B^2$	0.006

### 53. From Evidence to Inference

Either an alleged father is the true father or not, and a court may ultimately be called on to render a verdict. If genetic testing excludes the man then, barring a gene mutation and errors in testing and transcribing, he is not the father. (Presumably a modern court would reach a conclusion different from the one that decided against Charlie Chaplin in a famous case<sup>3</sup> from the 1940's.) If the man is not excluded then the evidence is not decisive--one possibility is to quantify "degree of paternity": How likely is it that the man is the father on the basis of the quantitative evidence? Such questions are statistical in nature and can be addressed by either Bayesian or classical methods.

The Bayesian approach is ideal for this problem in the sense that Bayes's theorem gives the relation between "inverse probabilities": the probability of guilt given evidence and the probability of evidence given guilt. The usual objection to a Bayesian approach is that the decision maker must assess prior information before, or independently from, the evidence at hand. This in turn leads to a subjective interpretation of probability. There is a natural "subject" or "decision maker" when a paternity case is brought to court: the individual juror (or the judge in non-jury trials). The ability (or willingness!) of a juror to make a probability assessment is another matter--this will be discussed in Section 5.

The problem of disputed paternity can also be addressed from

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<sup>3</sup>Berry v. Chaplin, 74 Cal. App. 2d 652, 664-65, 169 P. 2d 442, 450-451 (1946).

a classical hypothesis testing point of view. The null hypothesis is that the alleged father is the father and the alternative is that he is not. A test based on a series of blood group determinations that rejects the null only when the alleged father is excluded has significance level  $\alpha$  but indeterminate power. (Any other nonrandomized test with  $\alpha < 1$  will be arbitrary and difficult to describe. Randomized tests are inappropriate in this and similar contexts and would be disallowed by courts--see Section 5 for related discussion.) There is no natural alternative (unless there are two potential fathers, with the blood types of both available) at which to evaluate power, and averaging power over the population seems reasonable. This of course is a Bayesian notion. To our knowledge classical approaches have not been used in paternity cases. The remainder of this article deals with the Bayesian approach.

#### Bayes's Theorem

Bayes's theorem is an immediate consequence of the definition of conditional probability. Stated simply, it says that the probability of a statement being true given some new evidence  $E$  is proportional to the probability that it was considered true before obtaining  $E$ , times the probability that  $E$  would obtain if the statement were true.

For example, we suspect that a coin that has just been tossed five times yielding five heads is not a fair coin. The probability

that a fair coin would show five heads in any five independent tosses is  $1/32$ . There is a nonsensical tendency among naive users of significance tests to say that  $1/32$  is the probability that the coin is a fair coin. The probability that the coin is fair is of course related to the evidence at hand, but the problem cannot be addressed unless alternative hypotheses are specified. Suppose that prior to tossing a coin we consider it to be fair with probability 0.95 and, say, two-headed with probability 0.05. Given the new evidence, the probability that the coin is fair is proportional to  $(0.95)(1/32)$  and the probability that the coin is two-headed is proportional to  $(0.05)(1)$ ; since the corresponding probabilities must sum to 1 they are approximately 0.37 and 0.63, changed from 0.95 and 0.05. The output of Bayes's theorem is a "posterior" probability assessment of the truth of the opposing statements--posterior to the new evidence.

This example is obviously simplistic. There is seldom a single clear-cut alternative (such as "two-headed coin") to the statement under consideration. It is unlikely in a legal case that all parties would agree that one of two particular people is guilty. Usually there are a large number of alternatives. The probability of obtaining E, the evidence at hand, must be assessed under each alternative. In addition, the probability of each of the possible alternatives must be assessed.

Let  $S_1, S_2, \dots$  stand for the possible true statements and  $\Pr(S_i)$  the prior probability of  $S_i$ . The likelihood of  $S_i$  on the basis of evidence  $E$  is  $\Pr(E|S_i)$ . Then the posterior probability of  $S_i$  is given by Bayes's theorem:

$$\Pr(S_i|E) = \Pr(E|S_i) \cdot \Pr(S_i) / K$$

The constant  $K$  is determined by the requirement that the total probability is 1:

$$K = \Pr(E) = \Pr(E|S_1) \cdot \Pr(S_1) + \Pr(E|S_2) \cdot \Pr(S_2) + \dots$$

#### An Example

To illustrate with a case of disputed paternity and very simple genetic evidence, consider ABO system phenotypes for white Californians discussed by Grunbaum, et al. (1978), with frequencies given in Table 2. The least complicated application of Bayes's theorem is for a set of possible fathers with their phenotypes given (a more realistic assumption will be made in the next section).

Six men (Mr. 1, ..., Mr. 6) are the only possibilities as the father of the child in question and they are deemed equally likely on the basis of other evidence. That is,  $\Pr(S_i) = 1/6$  where  $S_i$  is the statement "Mr.  $i$  is the child's father." Evidence  $E$  consists of the blood type information given in Table 5. The problem is to incorporate this new evidence.

Table 5: Evidence  $E$  -- ABO system

Person	Child	Mother	Mr. 1	Mr. 2	Mr. 3	Mr. 4	Mr. 5	Mr. 6
Phenotype	O	O	O	A	B	AB	O	A

Since parental genotypes AB and OO cannot produce a type O child,  $\Pr(E|S_4) = 0$ . Since type O crossed with type O always gives rise to type O,

$$\Pr(E|S_1) = \Pr(E|S_5) = 1.$$

The other likelihoods are complicated by the fact that the genotypes of types A and B are not known. Consider  $S_2$ , or equivalently,  $S_6$ . If Mr. 2 is the father then he must be genotype AO--this has probability  $2g_0g_A/p_A = 31/36$  given he is type A. Further, the probability of AO (father) and OO (mother) giving rise to type O (child) is 1/2: the child is type O if and only if the father's O allele is passed on. So

$$\Pr(E|S_2) = \frac{1}{2} \cdot \frac{31}{36} = 0.431.$$

Similarly,

$$\Pr(E|S_3) = \frac{1}{2} \cdot \frac{116}{123} = 0.472.$$

The value of K is 0.556. The required likelihoods and posterior probabilities are given in Table 6.

Table 6: From Prior to Posterior via Evidence E

Statement	$S_1$	$S_2$	$S_3$	$S_4$	$S_5$	$S_6$
Probability Prior to E	1/6	1/6	1/6	1/6	1/6	1/6
Likelihood: $\Pr(E S_i)$	1	0.431	0.472	0	1	0.431
Probability Posterior to E	0.300	0.129	0.141	0	0.300	0.129



Among the six candidates, only Mr. 4 is exonerated by the evidence. Every other man with type AB would also be exonerated. On the other hand, men with type O have the highest likelihood.

Now suppose new evidence  $E'$  involving the Gc serum protein system is introduced. For the same population, estimates of the frequencies of alleles 1 and 2 are 0.710 and 0.290 (Grunbaum, et al. 1978). Since these are codominant, the genotypic frequencies are the same as the phenotypic frequencies; these are 0.504, 0.084, 0.412 for genotypes 11, 22, 12 respectively. Evidence  $E'$  is given in Table 7.

Table 7: Evidence  $E'$  -- Gc System

Person	Child	Mother	Mr. 1	Mr. 2	Mr. 3	Mr. 4	Mr. 5	Mr. 6
Phenotype	12	11	22	12	12	12	11	11

Assuming  $E'$  and  $E$  are independent, Bayes's theorem can be applied again. The likelihoods of the  $S_i$  for these new data are given in Table 8 along with the probabilities posterior to both  $E'$  and  $E$ .

Table 8: From Posterior to  $E$  to Posterior to  $E'$

Statement	$S_1$	$S_2$	$S_3$	$S_4$	$S_5$	$S_6$
Probability Prior to $E'$	0.300	0.129	0.141	0	0.300	0.129
Likelihood: $\Pr(E'   S_i)$	1	1/2	1/2	1/2	0	0
Probability Posterior to $E'$	0.690	0.148	0.162	0	0	0

The value of  $K$ , the probability of  $E'$  given  $E$ , is 0.435. Mr. 5 and Mr. 6 are now exonerated and, of course, Mr. 4 is still excluded.

As more genetic systems are included, more potential fathers are eliminated, and the probability of paternity for those not excluded tends to increase.

The way in which Bayes's theorem is applied in many courts is presented in the next section, which can properly be regarded as an extension of this section. This application will be discussed and carefully scrutinized in the next section and, especially, in the subsequent section.

#### §4. Current Use of Blood Testing in Law

The example in the previous section is not very realistic. First, it is unusual for all possible candidates to be known with certainty-- mothers conceal the number of possible fathers in at least 48% of paternity cases (Arthur and Reid 1954). Second, many more genetic polymorphisms than that provided by the ABO and Gc systems are available. We shall reconsider the example of the previous section making the more realistic assumption that one man, say Mr. 1, has been accused of being the child's father. For expository purposes we shall consider only evidence from the ABO system.

#### Likelihood Ratios and Prior Odds

Let  $S_i$  continue to stand for "Mr.  $i$  is the father" and let  $S_1^c$  be the complement of  $S_1$ . Mr. 1 plays a central role in the current discussion so it is convenient to write, for  $i \geq 2$ ,

$$\Pr(S_i) = \Pr(S_i | S_1^c) \Pr(S_1^c).$$

Rewriting Bayes's theorem,

$$\Pr(S_1|E) = \left[ 1 + \frac{\Pr(S_1^c)}{\Pr(S_1)} \cdot \frac{\Pr(E|S_1^c)}{\Pr(E|S_1)} \right]^{-1}$$

where the "likelihood" of  $S_1^c$  is

$$\Pr(E|S_1^c) = \sum_{i \geq 2} \Pr(E|S_i) \Pr(S_i|S_1^c),$$

really an average or integrated likelihood. Expressed equivalently,

$$\frac{\Pr(S_1^c|E)}{\Pr(S_1|E)} = \frac{\Pr(S_1^c)}{\Pr(S_1)} \cdot \frac{\Pr(E|S_1^c)}{\Pr(E|S_1)};$$

the posterior odds ratio is the product of the prior odds ratio and the likelihood ratio.

Evidence E is the child's, mother's, and alleged father's ABO system blood types. The likelihood of  $S_i$  depends on Mr. i's blood type and is given as in the previous section. It may be appropriate to restrict laboratories from supplying any more than the various  $\Pr(E|S_i)$  to court. But the current practice of many laboratories goes further, and their calculations are usually admitted--if not well understood!

First, it is assumed that the true father is a randomly selected man from some population (we'll return to this in Chapter 5) if the alleged father is not the father. Since all men with the same genotype have the same likelihood and conditional (on  $S_1^c$ ) prior probability, they can be grouped together. The conditional prior probability of each group is the proportion of the corresponding genotype in the population. So

$$\begin{aligned} \Pr(E|S_1^c) = & g_0^2 \Pr(E|\text{Father is OO}) + 2g_0g_A \Pr(E|\text{Father is AO}) \\ & + g_A^2 \Pr(E|\text{Father is AA}) + 2g_0g_B \Pr(E|\text{Father is BO}) \\ & + g_B^2 \Pr(E|\text{Father is BB}) + 2g_Ag_B \Pr(E|\text{Father is AB}) \end{aligned}$$

In the example of the previous section in which both mother and child are type O, there is a much easier route. Namely, this evidence will result if and only if an O allele is selected (randomly) from the population; so  $P(E|S_1^c) = g_0$ , or about 0.692 in the example.

The above assumption is innocuous when compared to the next one! Suppose that Mr. 1 and one other man of unknown blood type are equally likely to be the father; that is,  $\Pr(S_1) = \Pr(S_1^c) = 0.5$ . Then the posterior odds equal the likelihood ratio. If, as in Section 3, Mr. 1 is type O then

$$\Pr(S_1|E) = \frac{1}{1 + 0.692} = 0.591,$$

increased somewhat from the prior probability since Mr. 1 is in a group of men--those with type O--who have the highest likelihood.

The inverse of the above likelihood ratio,  $\Pr(E|S_1)/\Pr(E|S_1^c)$ , plays an important role in some courtroom presentations. It is called the paternity index, or P.I., by Salmon and Salmon (1980). The higher the paternity index the greater the relative likelihood of  $S_1$ .

One problem with converting a paternity index into a probability via Bayes's theorem is assessing the prior probability  $\Pr(S_1)$ . It is artificial to suppose, as we essentially did above, that exactly two men, including the alleged father, had intercourse with the mother near the time of conception, each the same number of times. The number of men who could be the father is usually a point of contention between the two sides. Another point of contention may be whether intercourse with the alleged father ever took place, or if it did, the timing of such intercourse relative to the child's birthdate.

The other problem with this paternity index is that it assumes a "random man" is the alternative to the alleged father. The question is, random from what population? Averaging with respect to different relative frequencies to obtain the likelihood  $\Pr(E|S_1^c)$  can substantially affect it.

These two issues are related; both will be returned to in Section 5.

#### Likelihoods for Multiple Gene Systems: Independence

In the above example, suppose the Gc phenotypes of child, mother, and Mr. 1 are known to be 12, 11, and 22, respectively; call this evidence  $E'$ . Now the likelihood of  $S_1$  for  $E'$  is

$$\Pr(E' | S_1) = 1.$$

Assuming that the true father is selected randomly from the hypothesized population when  $S_1$  is false implies that the (average) likelihood of  $S_1^c$  for  $E'$  is

$$\begin{aligned} \Pr(E' | S_1^c) &= g_1^2 \Pr(E' | \text{Father is 11}) + 2g_1g_2 \Pr(E' | \text{Father is 12}) \\ &+ g_2^2 \Pr(E' | \text{Father is 22}). \end{aligned}$$

Again the analysis is simpler. Evidence  $E'$  obtains when a 2 allele is selected randomly from the population; this has probability  $g_2 \approx 0.290$ . Hence, using probabilities posterior to  $E$  as prior to  $E'$ ,

$$\Pr(S_1 | E, E') = \frac{1}{1 + \frac{0.290 \cdot 0.409}{1 \cdot 0.591}} = 0.833.$$

This still assumes that the probability of  $S_1$  apart from blood data is 0.5.

Bringing more and more evidence to bear in this way will tend to increase the posterior probability of Mr. 1 if he (or his identical twin!) is the father and tend to decrease it--perhaps make it 0--if he is not. This repeated application of Bayes's theorem is not only appropriate for blood test data or other genetic polymorphisms but applies whenever information can be quantified using probabilities. But there is a proviso. It would be incorrect to use the same data twice. More generally, the individual pieces of information should be independent.

Instead of applying equation (1) for E and then for E', a more direct path, in terms of odds, is as follows:

$$\frac{\Pr(S_1^c | E, E')}{\Pr(S_1 | E, E')} = \frac{\Pr(E | S_1^c)}{\Pr(E | S_1)} \cdot \frac{\Pr(E' | S_1^c)}{\Pr(E' | S_1)} \cdot \frac{\Pr(S_1^c)}{P(S_1)}$$

The "paternity index" is now

$$\frac{\Pr(E | S_1)}{\Pr(E | S_1^c)} \cdot \frac{\Pr(E' | S_1)}{\Pr(E' | S_1^c)}$$

the product of individual likelihood ratios. Multiplication of probabilities is appropriate only if E and E' are statistically independent. For example, it must be that frequencies 0.504, 0.084, 0.412 for Gc phenotypes 11, 22, 12 hold for each blood type. If almost every type 0 has Gc-22, say, then the above calculations are inappropriate. Bayes's theorem would still apply, but the ABO and Gc systems would have to be considered jointly, with frequencies given for the various combinations of ABO and Gc phenotypes.

In paternity cases, calculations of an index and the posterior probability of paternity assume independence of the blood factors tested. There is some justification for this assumption.<sup>4</sup> Grunbaum, et al. (1978) present data for over 10,000 individuals to show that 12 factors--including ABO and Gc--are either pairwise independent or, perhaps, negligibly dependent. While pairwise independence is weaker than independence, this result does lend credence to a calculation in which likelihoods for these 12 factors are multiplied.

A hypothetical example is shown in Table 9. None of the tests excludes the alleged father. Estimates of genetic frequencies for a white and a black California population were taken from Grunbaum, et al. (1978). (The appropriate "population" to be used is that of the true father, not that of the alleged father, as used by all laboratories we know about--see Section 5.) For certain factors these frequencies vary considerably by race. In this case the paternity index is 65 times larger in the black population than in the white one! Obviously, some of these blood factors are racially dependent.

Table 9: Gene System Likelihood Ratios; White vs. Black

Gene System	Child	Mother	Mr. 1	Likelihood Ratio (White)	Likelihood Ratio (Black)
ABO	O	O	O	0.692	0.690
Rhesus	-	-	-	0.208	0.074
PGM	11	11	12	1.534	1.619
AK	12	11	22	0.037	0.008
ADA	11	11	12	1.898	1.796
EAP	AA	CA	BA	0.670	0.428
EsD	12	12	12	1.807	1.840
G-6-PD	B	B	B	0.995	0.730
Hb	A	A	A	0.999	0.956
Hp	12	12	12	1.507	1.517
Gc	12	11	22	0.290	0.129
PGD	A	A	A	0.981	0.964
Product				$8.00 \times 10^{-3}$	$1.23 \times 10^{-4}$
Paternity Index (=1/Product)				125	8100
"Plausibility of Paternity" (=1/(1 + Product))				99.2%	99.99%

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<sup>4</sup>We have heard indirectly from several geneticists and pathologists who claim that this is not an assumption, but a fact. Indeed, it is standard practice to assume independence of genetic polymorphisms in legal settings without stating the assumption. For example, the two volume work by Schatkin (1984) never mentions it as an assumption though hundreds of genetics articles are referenced. Also, Schatkin (1984, Chs. 5 to 9) cites many experts who casually multiply probabilities for up to 100 genetic polymorphisms (not all of which are specified), assuming independence without saying so. We are not convinced!



"Plausibility of paternity" is also given in the table. Although the black paternity index is 65 times that of the white, both plausibilities are close to 1. (Schatkin (1984, p. 8-37)) cites a case in which the "plausibility of paternity" is 98.5% when the reference population is white and 54% when it is black.) This term is used by some to mean the posterior probability of paternity assuming the prior probability,  $PR(S_1)$ , is 0.5 and the true father is selected randomly from the population if  $S_1$  is false. While the term is misleading, it is better than the alternatives which are also used: "probability of paternity" and "likelihood of paternity."

Some laboratories take the liberty of transforming this "plausibility of paternity" into an assessment of the truth of the ultimate question by providing Hummel's<sup>6</sup> Likelihood of Paternity given in Table 10.

Table 10: Hummel's Likelihood of Paternity

<u>Plausibility of Paternity</u>	<u>Likelihood of Paternity</u>
0.9980 - 0.9990	practically proved
0.9910 - 0.9979	extremely likely
0.9500 - 0.9909	very likely
0.9000 - 0.9499	likely
0.8000 - 0.8999	undecided
less than 0.8000	not useful

In practice, factors are tested that are not among the 12 given in Table 9. But if additional factors cannot be shown to be independent of all other factors tested, their use in calculating these indices should be criticized and should be disallowed in court (unless they serve to exclude a putative father).

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<sup>6</sup>Family Law Quarterly 10:262 (1976).

Some of the factors given in Table 9 are not tested in practice. The reason is clear: several of them (e.g., AK, G-6-PD, Hb, PGD) are poor discriminators and a greater number of tests allows more room for misclassification errors (Chakraborty, et al. 1974). These tests have low (average) probabilities of exclusion, but there are measures for use in selecting tests that are somewhat more appropriate for the analysis we have described. For a man selected randomly from the population, it is a trivial calculation to show that the expected value of his paternity index is 1 regardless of the number of gene systems involved. This is true whether or not the child/mother phenotypic combination is given. (A corollary is that the expected posterior odds of paternity is the prior odds.) Systems should be chosen if for that system the paternity index of a randomly selected man has substantial variability--measured, say, by its standard deviation.

The standard deviation of the system P.I. depends on the child's and mother's phenotypes. Table 11 provides an example using the ABO system and the white California population discussed in Section 2. Since these phenotypes are, of course, not available before deciding which systems to test, the unconditional standard deviation is required. The variance of P.I. can be calculated by averaging the conditional variances over the distribution of the various child/mother combinations since the mean P.I. is constant. For the example of Table 11 the requisite distribution is given in Table 4; the (average) variance is 0.700 and standard deviation is 0.837. Only systems with sufficiently large standard deviations of their P.I., say at least 0.5, should be tested. If, say, 12 blood group systems are used and all have standard deviation 0.5, then the standard deviation of the P.I. is only  $\sqrt{(1 + (0.5)^2)^{12} - 1} = 3.7$ . On the other hand, if the individual standard deviations are 1 then the overall standard deviation is 64. The latter case not only provides a much better chance for exclusion, but also will yield a much larger probability of paternity for a man who is not excluded.

Table 11: Estimates of Standard Deviation of Random Paternity Index (White California Population)

<u>Child/Mother Phenotypes</u>	<u>Standard Deviation of Random P.I.</u>	<u>Child/Mother Phenotypes</u>	<u>Standard Deviation of Random P.I.</u>
O/O	0.443	O/B	0.445
A/O	1.230	A/B	1.229
B/O	2.282	B/B	0.410
AB/O	--	AB/B	1.235
O/A	0.445	O/AB	--
A/A	0.283	A/AB	0.209
B/A	2.297	B/AB	0.356
AB/A	2.277	AB/AB	0.997

## Probability of Paternity Based on Nonexclusion

An analysis using Bayes's theorem applied to exclusion/nonexclusion has been suggested by many authors; e.g., Lee (1975), Wiener (1976). For example, given that a man has not been excluded but 90% of all men would be excluded, the posterior odds of paternity are nine times the prior odds. Such inferences are weaker than the approach described earlier because they are based on a reduction of the data.

Sometimes this reduction is substantial. Suppose mother and child have type A blood. Then there is no information in the fact that an alleged father is not excluded: the prior odds of paternity would be unchanged because no men are excluded. However, the P.I. varies in white Californians from 0.44 (odds of paternity decreased) for type B men to 1.32 (odds increased) for type A men. For a nonexcluded man the odds of paternity would usually increase if full information is used; it would be a mistake for the counsel of the alleged father to object to this approach in favor of the one described earlier!

## §5. Discussion and Recommendations

There are a number of important issues that we have not resolved. How accurate are blood tests? What effect do inaccuracies have? How are prior probabilities assessed? Who does the assessing? How does one combine genetic and other evidence? These are among the questions considered in this section. In addition, questions for expert witnesses and effective communication of these issues to a jury are discussed.

### The Role of Probabilities in Law

Should courts be guided to a posterior probability of paternity, or probability of guilt in criminal cases? Fairley (1973) presents arguments for both sides. He also describes a study that shows unaided intuition to be inept in learning from probabilistic evidence. While the case for formal

analysis, administered with appropriate qualifiers, seems strong, it assumes that probability does have a role in law.

Ellman and Kaye (1979) argue for the appropriate use of probabilistic evidence in legal cases. They indicate that many people view such evidence as being comparable to Rabelais's Judge Bridlegoose who rolled dice to decide cases, the higher roll winning. We agree with them that a decision based on an assessed probability is not a randomized decision. The distinction is between constructing dice to specifications and actually rolling them.

Courts frequently decide cases in which they are uncertain about the correct disposition. In giving his reasons for rolling dice to his peers, Judge Bridlegoose repeatedly says that he throws dice "just like you other gentlemen." This repetition--the phrase occurs in practically every sentence--suggests that Rabelais was convinced that an element of randomness is present in all court decisions. Judge Bridlegoose had the advantage over his peers in knowing which dice he used in each case! "It took the testimony of a sage, an oracle, a drunken party goer, a messenger, a shepherd and his own wife before Oedipus could figure out who his father was." (New York Times, June 1981, as quoted by Schatkin (1984, p. 8-29)). Modern paternity cases offer little good evidence of a nature other than probabilistic.

#### Blood Tests and Other Evidence: Assessing Priors

Defining a probability of paternity on the basis of genetic testing alone is like assigning a probability to the proposition that a coin is fair using the results of several tosses of the coin. There is no logical foundation for an assignment based solely on the data, but

people try to do it nonetheless.<sup>5</sup>

A mistake made in the literature and practice of genetic testing for paternity is easy to identify. The entire discipline recognizes that Bayes's theorem must be applied, but some paternity testers

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<sup>5</sup>The following quote from Schatkin (1984, preface pp. 809) is given without comment.

As a rule, one hundred blood tests will result in some fifteen exclusions. Multiplying 15 by 2 (because the blood test potential for exonerating an innocent man is about 55 per cent.) in that series of one hundred blood tests carried out, 30 per cent. are actually not the fathers. Falling back on the analogy of a woman putting her hand into an urn and making her selection of whom to accuse, the thirty innocent men in that series represent a wrong guess on her part and a gamble on her part that failed. Blood test exclusions, therefore, demonstrate those cases where the woman "guessed wrong."

As stated, 15 exclusions result from 100 blood tests carried out, and of those 100 men, 30 are actually not the fathers. And, 85 are not excluded. And of those 85 not excluded, we know that 15 are not the father. So that, what are the probabilities of one of those 85 actually being the father? We divide 85 by 15. Therefore the chances of one of those men not excluded being the actual father, is in the proportion of 5½ to 1. We conclude, therefore, that if a man is not excluded, the chances are 5 to 1 that he is the father of the child.

(apparently dating to Essen-Möller (1938)) want to use the same prior probability in every case! Namely, they assume that, aside from the genetic data, the alleged father is the true father with probability 1/2. From our personal experience we know of one pathologist who testified in court that the probability, or "plausibility", of paternity does not depend on the number of men who had sexual intercourse with the mother near the time of conception. In fact, a candidate for the prior probability for an alleged father is the number of times he had intercourse with the mother near the time of conception divided by the total number of times she had intercourse in that period. (This could be refined to take into account more likely times for conception, viability of the man's sperm, etc.) But this information is known only to the mother and even she may have forgotten. In assessing a prior probability a juror must digest a variety of conflicting testimony concerning this and other issues.

There may be cases in which some jurors actually have a prior probability of 1/2. But introducing it in court under the guise of blood typing is grossly misleading unless the implications are made clear. Ideally, each juror should appraise the information, other than the blood typing data, in the case at hand and assess a prior probability on that basis. They can then be told how to transform it into posterior probabilities and in turn use it to reach a verdict.

The posterior probability of paternity, say  $\pi'$ , is a function of the prior, say  $\pi$ : namely,

$$\pi' = \left[ 1 + \frac{1}{\text{P.I.}} \frac{1-\pi}{\pi} \right]^{-1} ,$$

where P.I. = paternity index. Of course, judges and juries will have trouble with such a formula. But it can be tabled, and the table can include the traditional  $\pi = 1/2$ . Table 12 provides an example using Mr. 1's P.I. = 125 found in Table 9. It also gives the prior and posterior odds against paternity since some people think in those terms rather than in probabilities.

Table 12: Prior to Posterior Probabilities of Paternity for P.I. = 125

Prior Probability $\pi$	0	$\frac{1}{1,000,000}$	$\frac{1}{1,000}$	$\frac{1}{100}$	$\frac{1}{10}$	$\frac{1}{2}$	$\frac{9}{10}$	1
Posterior Probability $\pi'$	0	0.00012	0.111	0.558	0.933	0.992	0.999	1
Prior odds against	$\infty$	999,999:1	999:1	99:1	9:1	1:1	1:9	0
Posterior odds against	$\infty$	8000:1	8:1	4:5	1:14	1:124	1:999	0

There are several substantive problems with this "ideal". One is that the juror may refuse to quantify nongenetic information in terms of a probability, or even a range of probabilities. The juror can be asked to interpret probabilities in terms of small-stake bets, or betting odds. And many jurors will go along willingly. But some may have an aversion, moral or otherwise, to betting. It may help to indicate that these are only thought bets or preferences designed to quantify strength of belief.

(For related ideas see DeGroot (1970, Chapter 6).)

Some care in advising a jury in the matter of assigning a prior probability is necessary because the best mode of elicitation--dialogue--is not available. On the other hand, a high degree of precision is not necessary;



only a gross assessment of magnitude is needed. Bets or lotteries can be described which will help a juror decide on a range of prior probabilities. For example, the jury can be told: "If you would prefer being paid \$1 if this man is not the father to \$1 if he is, then your prior probability of paternity is less than 1/2. If, in addition, you would prefer \$1 if this man is the father to \$1 if a "1" is rolled on a fair die then it is greater than 1/6. A range of prior probabilities corresponds to a range of posterior probabilities as exemplified in Table 12. These bounds on the posterior probability may be sufficient to determine the juror's vote--in any event it will help. This suggests a method for setting up an analog of Table 12. The prior probabilities tabled can be calculated to correspond to some interesting posteriors probabilities; e.g., 0.99, 0.95, 0.90, 0.50.

The above discussion assumes that jurors are willing to assess prior probability distributions. While little can be done if they have no feeling for randomness, there are various devices that may help. One is as follows. They can be asked to suppose that the proportion of the time the mother had sexual intercourse with the alleged father, as opposed to other men, during the time in which conception was possible were known. Then this could serve as a prior probability. Since it is not known, relevant testimony can be weighed. If a juror can be made to assess a probability distribution on this proportion ("How likely do you feel this proportion is less than 10%?" Etc.) then Table 12 can be used with  $\pi$  equal to the mean of this distribution. The values  $\pi=0$  and  $\pi=1$  are important in this regard for they correspond to frequently heard testimony: namely, "never had intercourse with her" and "no other man". For example, if these latter are the only two possibilities, and are given equal weight by the juror, then the column  $\pi=1/2$  in the analog of Table 12 is appropriate.

The most serious difficulty in assessing a prior probability of paternity is setting the genetic information aside to ensure that it is, in effect, not used twice in evaluating a posterior probability. It would help if blood group and other genetic data were presented subsequent to all other evidence, but it can never be kept completely separate! That a case comes to trial almost always implies that the alleged father was not excluded, so "nonexclusion" is, in the broad sense, used twice. The double usage of this evidence can have an enormous impact on a jury. (Our experience is that cases that go to court are almost always decided for the plaintiff when the alleged father has a high paternity index and acknowledges intercourse with the mother at some time, though not necessarily during the time that conception was possible--Schatkin (1984) gives many case histories.)

One remedy is to calculate a paternity index conditionally on the fact that the man was not excluded. This would be easy to do and would result in substantially lower P.I.s for nonexcluded men. Such an adjustment is appropriate and seems essential if justice is to be served.

The following recommendation by Wiener (1976) is related to this double usage: "The value of [the a priori probability of paternity] depends on the experience of the courts--e.g., if 75 percent of the defendants have been found innocent of the charge then the a priori probability of paternity ... is 0.75 (sic; he meant 0.25)." There are a number of serious objections to this proposal. One is that the nonexclusion of the alleged father is used twice in an obviously formal way (while true for the use of Bayes's theorem with a paternity index, this is especially clear in Wiener's context because he goes on to use Bayes's theorem conditioning on nonexclusion). As stated previously, prior probabilities should depend only on the particulars of the case at hand but not on any blood typing data that will be presented in evidence.

### What Reference Population?

The paternity index depends heavily on the reference population used. A common practice of laboratories is to use the genetic frequencies in the race of the alleged father to calculate the paternity index. The logic for this is difficult to comprehend; the calculation is appropriate only if the true father is of the same race. As indicated in the example of the previous section, the resulting bias can be substantial. While not perfect, it would be much more appropriate to use the race of the child. If the race of the true father is an issue in a particular case, then the P.I. should be calculated by averaging over the local population. Alternatively, if the alleged father is white, say, and the defense claims the true father is black, then different sets of calculations should be made. The jury can decide which to believe, or how to weight them.

An obvious difficulty in making calculations for "the correct" reference population is the lack of appropriate data. Suppose a woman becomes pregnant in a small secluded town in which there are few families, some inbreeding, and little genetic variation. Using the population of the entire country as the reference set is obviously inappropriate. In particular, the proportion of the town's population excluded by blood tests would likely be much smaller than the proportion of the larger population that would be excluded.

Blood group data in a case alleging incest need not be handled differently from any other paternity case. Calculating the likelihood of the alleged father depends only on the three blood samples, and the fact that the mother's and alleged father's genetic structures are similar is of no additional consequence. But the likelihood of the "random man" is greatly affected if a suspected alternative to the alleged father is related to the mother or the alleged father, whether or not the latter two are themselves related. This information would be easy to incorporate if the blood groups of any such alternatives are known, and extremely difficult if not. Men whose identities are known can be regarded as "random", but not if they are related to the mother or alleged father.

Errors in Testing: Other Realities

The analysis of the preceding section assumed that tests for genetic factors are error-free. Chakraborty, et al. (1974) cite studies showing that "misclassifications insofar as the blood groups are concerned are more common than generally acknowledged and even in highly reputable laboratories may involve 2% - 3% of all determinations." This has an effect on the paternity index and in turn on the probability of paternity. While not done in practice, error rates could easily be incorporated: the genetic evidence can be given in terms of a probability distribution which incorporates the possibility of error, and likelihoods can be calculated on this basis. Not explicitly considering misclassification errors encourages a court to lend more credence to the report from a laboratory than is justified.

The possibility of misclassification errors means, of course, that no man is excluded with certainty--only with high probability. For example, assuming a 2% error rate in ABO classification, Mr. 4 would not have been excluded in Section 3. Table 13 revises Table 6 assuming a 2% error rate in the blood tests of the possible fathers (mother and child are both assumed to be type 0--the error possibilities in their tests would make for still less change in the probabilities from prior to posterior).

Table 13: Table 6 Modified by 2% Classification Error Rate

Statement	S <sub>1</sub>	S <sub>2</sub>	S <sub>3</sub>	S <sub>4</sub>	S <sub>5</sub>	S <sub>6</sub>
Probability Prior to E	1/6	1/6	1/6	1/6	1/6	1/6
Likelihood Pr(E S <sub>i</sub> )	0.988	0.439	0.476	0.014	0.988	0.439
Probability Posterior to E	0.295	0.131	0.142	0.004	0.295	0.131

In general, the greater the possibility of classification error, the closer a paternity index is to 1. This means that men who are not excluded as father will tend to have smaller probabilities of paternity, and men who are "excluded" will tend to have greater probabilities.

In the words of Wiener (1976), ". . . researchers have had occasion to retest and have detected errors in more than a score of cases . . . . If the newer tests [new blood groups] are included, the possibility of error will, of course, be multiplied, especially because many of the newer tests are not perfected." And, ". . . as the number of tests increase . . . a point will be reached at which the chances of exclusion increase more slowly even than the chances of error and where further testing is extravagantly costly . . . ."

The better laboratories recognize the possibility of error and do all tests in duplicate and some in triplicate, (Polesky 1975, p. 89). Assuming the individual tests are independent--even though technicians are blinded, this assumption may not be entirely valid--the error rate is substantially reduced. Presumably they report the mode in case of disagreement. The actual policy in these matters should be made public. But more importantly, all results (three, if triplicate) should be presented in court. Requesting all the data would be an appropriate tool for a lawyer whose case is suffering. While this information may tend to obscure matters for a judge or jury, it is necessary for completeness. Withholding such information creates an illusion of precision that may be unwarranted.

(The situation is analogous to that of an experimenter who makes observations in triplicate and uses only their means or medians in a regression problem, an all too common practice-- $R^2$  will be artificially inflated.)

(Blood tests are highly respected as evidence by the legal profession. For example, in a chapter entitled The Unerring Accuracy of Blood Tests, Schatkin (1984, p. 11-1) displays a rather curious logic to come to the following conclusion.

Verification of the accuracy of blood tests came not long after their inception. During the ten-year period March 22, 1935 to March 22, 1945, 656 blood tests carried out in affiliation cases by order of the Court of Special Sessions in New York City resulted in 65 exclusions. The question naturally arises, Were those exclusions accurate? The answer is Yes, because each and every one of those 65 exclusions was followed by the mother's subsequent confession, for the first time, of sexual relations with another man about the time she became pregnant.

Actually, the information given is also consistent with every blood test being wrong!)

There are other errors which enter into the calculation of a paternity index that should be mentioned. One form of error is statistical. The phenotypic frequencies used are based on samples and not complete population counts. Some laboratories use published frequencies while some others, notably blood banks, keep records of previous blood samples tested. For example, the estimates used in Table 9 were reported by Grunbaum, et al. (1978) and were based on blood samples of 6004 white and 1024 black Californians that were collected from blood banks around the state. Assuming these are representative of Californians (a dubious assumption!), the standard errors in estimating the percentage of O alleles in the white and black population to be 0.692 and 0.690 are 0.6% and 1.4%. So a 95% confidence interval for the proportion of O all among black Californians is 66.2 to 71.8%. Of course, the smaller the sample size, the larger the standard error of the estimate.

The next point seems minor and it seems difficult to deal with in the courtroom. More than one laboratory in the United States carries out calculations of likelihood ratios, and presents them in court, using six-digit accuracy; for example, a paternity index of 51.3204 (translating to a plausibility of paternity of 98.089%). In view of the presence of the above errors, calculations reported to more than 2 digits are suspect. They do not deceive educated observers but, again, they can create an illusion of great accuracy in court. A lawyer may question the accuracy of the numbers, but even if the paternity tester recants and, in the above example, says P.I. = 51, the plausibility of paternity is essentially unchanged. The endeavor can be perceived as inconsequential and unnecessary carping to a jury.

Even the three-digit accuracy reported in Table 9 is misleading. (We note that the frequency of AK in blacks was reported to one-digit accuracy. This alone makes the reported paternity index of 8100 subject to an error of up to 500.) Suppose the relative error in each of the 12 gene system likelihood ratios is 2%. Then the relative error in the product of the 12 is

$$\sqrt{(1 + 0.02^2)^{12} - 1} = 6.9\%$$

or about  $\sqrt{12}$  as large, so an error of 15% is quite possible.

#### Additional Genetic Evidence

A posterior probability of paternity can change in two ways: through the prior and through the paternity index. We have discussed the prior and the denominator of the paternity index (which is affected by the choice of reference population) previously. The P.I. also depends on the probability of the child's phenotypic structure given the mother's and assuming the alleged father is the father. This likelihood can be changed--perhaps to exclude a previously nonexcluded man--by gathering more data.



There is no universal standard that indicates which blood group systems to use in calculating a particular paternity index. Some limit on the number used is necessary to help ensure independence, minimize errors, and keep costs down. But clearly an alleged father who knows he cannot be the father should ask for replicate testing and testing on further systems until he is excluded. On the other hand, a mother who is certain that the father is a man who has been excluded should ask for retesting because either a mistake has been made or a "silent" gene is involved. In certain codominant systems, when it happens that the mother and child are both homozygous for the same allele and apparently the father lacks that allele and so is judged to be homozygous for the second dominant allele, he is excluded. In this case the man may not be homozygous but actually have a "silent" recessive gene which is not detectable by the standard test so that the exclusion is false. The frequency of such a gene in the serum protein haptoglobin system is reported by Cook, et al. (1969). Competent facilities take this into account in their reports (Dodd and Lincoln 1981).

Another way of gathering additional relevant data without testing more factors is to test relatives of the mother and alleged father. For example, in Section 3, Mr. 2 could father a type O child with a type O mother only if his genotype is AO. So if it were determined that his true parents were both AB then he would be exonerated. Still relevant, though not conclusive, would be evidence that his other  $n$  children with a type O mother were type A. The likelihood that he is AA for this latter set of data is 1 and the likelihood of AO is only  $(1/2)^n$ . On the other hand, if one of his parents or one of his other were type O then his genotype is in fact AO and his P.I. would increase somewhat.

Other kinds of genetic evidence that have been used in paternity cases are less accurate measures of heredity but are much better understood by lay people: hair color, eye color, "family resemblance", etc. Incorporating

this information into the P.I. is difficult but incorporating it into the prior is possible. For example, if both the mother and the alleged father are blue-eyed and devoid of any brown pigment in their irises but the child is definitely brown-eyed, the alleged father is excludable. On the other hand, a devastating impact can be made on a jury when a mother presents her red-headed child in court and the nonexcluded alleged father also has red hair. Still, cautious lawyers will often balk at exhibiting a child in court because of the poor understanding people have of these easily observed hereditary traits.

#### Questions for a Geneticist or Pathologist in Court

A number of substantive issues have been raised in this paper concerning the way in which genetic information is used in paternity cases. Many of these issues should be exposed in court. We present a few sample questions for a geneticist here. These may aid an attorney in preparing a case or a geneticist in critically rethinking an analysis; they also serve as a review for this paper.

Question: How were the blood factors you analyzed chosen?

Discussion: Presumably, factors were chosen if their phenotypes could be classified reliably and if they were discriminatory (having a high exclusion frequency). Also, tests are time-consuming so some limit is necessary.

Question: Do you always analyze these same factors?

Discussion: A negative answer can be embarrassing, especially if some factors were not tested in the present case that had been in others. For then the obvious question is,

Question: Could the alleged father have been excluded had you tested these other factors?

Answer: Yes.

Question: Are there still other factors that you have never used that might have excluded him?

Answer: Yes.

Question: Do the calculations you made assume that the various blood factors are independent?

Answer: Yes.

Question: Are they independent?

Discussion: There is no way for someone to know the answer a priori. If the answer is "yes" documentation should be requested. The paper by Grunbaum, et al. (1978) mentioned previously provides limited documentation on the 12 factors given in Table 9--"limited" to pairwise independence and Californians.

Question: What is the rate of classification error in your methods?

Discussion: Possible answer: Less than 3% for an individual test, but we do all tests in triplicate with three different analysts so the chance for error is negligible. (Some geneticists claim a very small error rate; documentation should be requested.)

Question: What do you do when the analysts disagree?

Discussion: If the answer is "we take the mode" then there is still a substantial chance for error. Possible answer: We retest until we are virtually certain of the result.

Question: What effect does the possibility of error have on the paternity index?

Discussion: The issue is very complicated. If substantial retesting is done to eliminate errors then the effect may be negligible.

Question: Were there any disagreements in any of the tests in the current case?

Discussion: Probable answer: I don't know.

Question: How accurate are the estimates of the phenotypic frequencies of the factors you used?

Answer: They are based on thousands of samples and so are accurate to within 2%.

Question: What effect can these errors have on the paternity index?

Answer: It could change it by 10-20% but it could not have excluded an alleged father whose P.I. is positive.

Question: How did you calculate the probability (or plausibility) of paternity in this case?

Discussion: The answer is bound to be long, involve references to formulas and computers and include phrases like "standard practice in the field". It is quite unlikely that any juror will understand the answer. The answer may be couched in terms of frequency-based probabilities ("Take the ratio of 125, the P.I., to 125 plus 1 for a random man and express it as a percent.") which will also suggest that the respondent does not really understand the meaning of a probability of paternity.

Question: Does this probability depend on whether or not the mother and alleged father had sexual intercourse proximate to the time of conception?

Discussion: Seemingly a silly question, but it is difficult to give a correct, extended answer. Obviously, it is not possible for the alleged father to be the father unless intercourse occurred near the time of conception. But a juror's prior probability should weigh the various possibilities in this regard. Then the posterior probability also weighs these possibilities--as they should. Put another way, a juror's probability of paternity is an average of two conditional probabilities, one assumes intercourse and the other does not (obviously, the latter is zero).

Question: Would this probability change if it were known that the alleged father and a number of other men had intercourse with the mother near the time of conception?

Answer: Yes. If the proportion of the time intercourse involved the alleged father is  $1/n$  then on the basis of this information the odds against the alleged father are increased by the factor  $n-1$  and there is a corresponding decrease in the probability of paternity.

Question: Under these conditions, what value of  $n$  would make the alleged father an even bet to be the father?

Answer:  $n = P.I.$ , the paternity index.

Question: What would be the effect on the posterior probability if it were known that one of the other men who had intercourse with the mother was a relative of the alleged father?

Answer: Then the calculation of the posterior probability is wrong. If, for example, the relative was his identical twin then both would have the same P.I. and, assuming equal frequency of intercourse, the same posterior probability which means it can be no greater than 50%.

#### 56. Other Available Tests and the Horizon

One of the great advantages of genetic testing for paternity is the potential savings in time, effort and money--especially in regard to litigation. Excluded fathers are rarely if ever these days, brought to trial by a plaintiff, and on the other hand when the true father is confronted with overwhelming genetical evidence as to his paternity, he might well accept the responsibility. Hence as a practical matter genetic testing becomes a reasonable and relatively inexpensive way to clear court dockets. It also serves to reduce welfare costs for the increasing number of children born out of wedlock since the father can be compelled to pay

child support.

Few courts have been hesitant to accept red blood cell antigen testing (Schatkin 1984). When this series fails to exclude, a resolution may be attempted using red blood cell and serum proteins or HLA typing; which one is used depends on state laws and the usual procedure of the laboratory concerned.

In discussing the admissibility of HLA testing, the Kansas Court of Appeals<sup>6</sup> notes that

"Since HLA testing is a relatively new test insofar as its use in the courtroom is concerned, it has been dealt with by only a few appellate courts. Several courts have refused to admit the test to show probability of paternity. In so doing, however these courts have in general acknowledged the test as reliable but nevertheless rejected the evidence under specific statutes which limit admissibility of blood test results to those which exclude the alleged father.

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<sup>6</sup>Tice v. Richardson, 8 F.L.R. 1113. Quoted from (Schatkin 1984, p. 3-28).

The value of an HLA test, though considerably more expensive to administer, lies in the large number of alleles, resulting in a much higher exclusion probability. It is also claimed (Perdue, et al. 1977) that the error rate of HLA typing classifications in pairs of replicate typing tests is less than 0.35% when performed under very carefully controlled conditions. The probability of exclusion for the HLA system alone is claimed to be 0.95 (Sussman and Gilja 1981). They also report that a total probability of exclusion of 0.9995 is available when red blood cell antigens and enzymes and serum proteins are used in conjunction with HLA.

A new potentially rich source of polymorphisms based on recombinant DNA technology is described by Botstein, et al. (1980). Suggestions are made that could eventually lead to a human genetic linkage map which would considerably elucidate modes of inheritance. The new markers are called restriction fragment length polymorphisms (RFLP) and can be assayed from small amounts of peripheral blood and, according to some, appear to be inherited as simple Mendelian codominant alleles. We may expect that the development, perfection, and use of this rich new source of genotypic differences could eventually lead to a probability of excluding an innocent man that, for all intents and purposes, is one. Of course it is not clear when such a goal will become a reality. In the meantime, since juridicial decisions cannot be put off, forensic statisticians can be of much service to litigants and the court by constantly probing assumptions, scrutinizing techniques, assessing the accuracy of results, examining the data closely, and providing a coherent framework for decision making.

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