International Congress Series 1288 (2006) 831-833





An unusual case of disputed paternity: When the legitimate children of a deceased alleged father deny DNA

Silvano Presciuttini ^{a,*}, Chiara Toni ^b, Isabella Spinetti ^b, Anna Rocchi ^b, Ranieri Domenici ^b

^a Center of Statistical Genetics, University of Pisa, Scuola Medica, Via Roma 55, 56127 Pisa, Italy ^b Unit of Legal Medicine, School of Medicine, University of Pisa, Italy

Abstract. In a recent disputed paternity case involving a family of six siblings with deceased parents and a woman claiming to be half sib of them, the question asked by the judge concerned the significance of knowing the genotypes of the legitimate children for determining the probability of paternity. We addressed this problem both by an analytical method and by computer simulations. The chance of excluding a false relationship by at least three incompatibilities, if the relationship was false, was rather poor (0.123), whereas the chance of obtaining a high probability of paternity if the relationship was true was higher than 99.9% almost with certainty. © 2005 Published by Elsevier B.V.

Keywords: Deficiency paternity; Paternity exclusion; Multiple siblings; Probability of paternity

1. Introduction

The following case of disputed paternity has come at our attention recently. The claimant pretended to be the natural daughter of a long-deceased man, survived by six legitimate children, who denied their DNA. In Italy, if the alleged father denies DNA, this is considered proof against him. Thus, the judiciary question verged on the importance of knowing the genotypes of the defendants in determining the probability of paternity in this particular case. Nine loci had already been typed in the claimant.

* Corresponding author. Tel.: +39 050 2218500; fax: +39 050 2218513. *E-mail address:* ranieri@biomed.unipi.it (S. Presciuttini).

^{0531-5131/} $\ensuremath{\mathbb{C}}$ 2005 Published by Elsevier B.V. doi:10.1016/j.ics.2005.09.043

2. Methods

We addressed separately the two questions of the probability of excluding a false parenthood due to locus incompatibility and of the probability of paternity given no incompatibilities. For the exclusion probability, we used both an analytical method and a simulation approach; for the probability of paternity, we used a simulation approach followed by a likelihood ratio calculation. We considered that 20 standard forensic loci would have been available for analysis.

2.1. Analytical method

We noted that exclusion of paternity occurs, in the case at hand, only if four different alleles are present among the legitimate siblings, and none of them is present in claimant's genotype. These conditions can be re-formulated in the following three statements, which must be concurrently true.

Both parents of the defending children are heterozygous for different alleles. The probability of this occurrence, i.e., that any two random individuals are different heterozygotes $[Pr(A_iA_j,A_kA_l), i\neq j\neq k\neq 1]$, is equal to the probability of drawing four different alleles from the ordered list of allele frequencies $[p_{(.)}]$, and it is easily found to be $Pr(A_iA_j,A_kA_l)=\sum_{(i< j< k< l)} 24 p_ip_jp_kp_l$.

Any of these four alleles have been transmitted at least once to a legitimate child. In general, the probability of this occurrence is independent of the allele frequencies, and in a sibship of size n is $= 1 - 4(1/2^n - 1/2^{2n})$. In our case this reads Pr(4 alleles present in 6 siblings) $= 1 - 4(1/2^6 - 1/2^{12}) = 0.9385$.

None of the alleles are present in claimant's genotype. The probability that a random individual (the claimant) does not share any of these four alleles $[Pr(A_mA_n|A_iA_j,A_kA_l), m \le n < i < j < k < l$, is $Pr(A_mA_n|A_iA_j,A_kA_l) = [1 - (p_i + p_j + p_k + p_l)]^2$.

These three probabilities were combined to obtain the probability of exclusion of each locus, and the exclusion probabilities were multiplied across loci to obtain the cumulative probability of exclusion.

2.2. Computer simulations

For determining the probability of paternity exclusion, we generated 10,000 families of six full sibs plus a random individual for each of the 20 chosen loci (the nine loci already typed in the claimant were fixed); the distribution of the number of exclusionary loci per family was then established by direct count.

Table 1

Distribution of the number of exclusionary loci in the hypothesis that claimant's contention is false

Number of loci with at least this number of exclusions	Probability (analytical method)	Estimated proportion \pm S.D. (simulation results)
0	0.241	0.235 ± 0.004
1	0.759	0.765 ± 0.004
2	0.376	0.380 ± 0.005
3	0.123	0.125 ± 0.003

Table 2

>99.99%

that chaimant's contention is true		
Probability of paternity	Number of simulated pedigree with probability higher than that	Estimated probability that a pedigree has a paternity probability higher than that
>99%	999/1000	0.999 ± 0.001
>99.9%	997/1000	0.997 ± 0.002

 0.967 ± 0.006

Thresholds of probability of paternity and the number of pedigrees with probability higher that in the hypothesis that claimant's contention is true

For determining the probability of paternity given no exclusions, we generated 1000 families composed by six full sibs and one half sib, and used MENDEL 5.0 (http://www.genetics.ucla.edu/software) to compute the likelihood ratio (LR) that the half sib was truly half sib rather than unrelated. In the LR calculation, only the genotypes of the seven children were used. LRs were converted into probabilities assuming equal priors, and the frequency distribution of these probabilities was calculated.

3. Results

3.1. Exclusion probability

967/1000

Table 1 shows the distribution of the number of loci (out of the 20 chosen for the analysis) that determined an exclusion of paternity if women's claim was false, i.e., if she was unrelated to the six legitimate siblings. Both the exact calculation and the results of the simulations are reported. These latter are not significantly different from the exact probability values. Two relevant cases are the probability of observing zero exclusions, which was 0.241, and the probability of observing at least three exclusions, which was 0.123.

3.2. Probability of paternity

Table 2 shows the results of the simulations performed in the hypothesis that woman's claim was true. For three values of the probability of paternity, the number of pedigrees with probability of paternity higher than that is reported, together with the corresponding estimated probability of this occurrence.

4. Conclusions

In conclusion, whereas the chance of excluding a false relationship is rather poor in this case, as at least three incompatibilities can be obtained in about 12% of cases only, the chance of obtaining a high probability of paternity is higher than 99.9% in almost all cases if the relationship is true.