Bayesian analysis of mixed stains when there are two related contributors

A. Peñafiel*, J.M. Ruiz de la Cuesta

Laboratorio de Biopatología Médico Legal, Departamento de Toxicología y Legislación Sanitaria, Universidad Complutense de Madrid, Ciudad Universitaria, 28040 Madrid, Spain

Abstract. The analysis of DNA extracted from biological evidence is today one of the most important tools in forensic science. Particularly usual in casework is to find mixed biological stains. Their mathematical study has already been done, including the case of subestructured populations. Here we present a set of formulae valid for the case where we know there are only two contributors and these are genetically related. Furthermore, we extract some interesting links between mixed biological stains belonging to two or more related people and those coming from unrelated people. The analysis rests on probability theory and mathematical genetics well described in scientific literature. © 2003 Elsevier B.V. All rights reserved.

Keywords: Likelihood ratio; Mixed stains; Relatedness; Bayesian evaluation; Forensic casework

1. Introduction

It is usual to find in forensic casework biological stains coming from two or more people [1–4]. If we are facing offences against sexual freedom, as is the case of a violation, in which two non-typed and perhaps genetically related offenders have participated, it is necessary to evaluate the genetic evidence taking this possibility carefully into account. The objective of this work is to obtain a set of expressions allowing us to assign probabilities to the allelic configuration for some given markers studied under the hypothesis that the contributors have a given relatedness between them.

2. Methodology and results

2.1. Theoretical aspects

According to Evett and Weir [1], the joint genotypic probability of the several pairs of genotypes for the different kinds of relatedness is:

$$P(aa, aa) = a^4 + 4\theta_{XY}a^3(1-a) + 2\Delta_{X+Y}a^3(1-a)^2$$
$$P(aa, bb) = (1 - 4\theta_{XY} + 2\Delta_{X+Y})a^2b^2$$
$$P(aa, ab) = 2(1 - 4\theta_{XY} + 2\Delta_{X+Y})a^3b + 4(\theta_{XY} - \Delta_{X+Y})a^2b$$

* Corresponding author. Tel.: +34-91-7738532; fax: +34-91-3051593.
E-mail address: armandomatematicas@hotmail.com (A. Peñafiel).

Table 1
Values of parameters θ_{XY} and Δ_{X+Y}

Relatedness	θ_{XY}	Δ_{X+Y}
Parent–offspring	1/4	0
Full sibs	1/4	1/8
Uncle–nephew	1/8	0
Half sibs	1/8	0
Grandparent–grandchild	1/8	0
First cousins	1/16	0

$$P(aa, bc) = 2(1 - 4\theta_{XY} + 2\Delta_{X+Y})a^2 bc$$
$$P(ab, ab) = 4[(1 - 4\theta_{XY} + 2\Delta_{X+Y})a^2 b^2 + (\theta_{XY} - \Delta_{X+Y})ab(a + b) + \Delta_{X+Y}ab]$$
$$P(ab, ac) = 4[(1 - 4\theta_{XY} + 2\Delta_{X+Y})a^2 bc + (\theta_{XY} - \Delta_{X+Y})abc]$$
$$P(ab, cd) = 4(1 - 4\theta_{XY} + 2\Delta_{X+Y})abcd$$

where the parameters θ_{XY} and Δ_{X+Y} take different values according to the presumed relatedness (Table 1).

With this as a starting point we will study mixed stains in those cases where a relatedness is presumed. Since we have applied the method for only two contributors, there will appear four alleles at most. Let M_α denote the stain which has the alleles $\alpha = a,b,c,d$. If G_{S_1} and G_{S_2} are the contributors' genotypes and H_p represents the hypothesis of relatedness between contributors, we will have:

$$P(M_\alpha | H_p) = \sum_{G_{S_1}, G_{S_2}} P(G_{S_1}, G_{S_2} / \alpha, H_p)$$

where the sum is done over all the genotypes compatible with the alleles which are found in the stain. Here we present the calculated expressions along with those corresponding to the non-relatedness case (Table 2).

2.2. Example

Let us suppose we are facing a crime-scene piece of evidence having one, two or three alleles. This makes us formulate the hypothesis of two contributors related as parent and offspring. In order to calculate the L.R. (likelihood ratio) properly [5,6], we should use the rows 2 and 3 of the previous table:

$$\text{L.R.}_a = \frac{1}{a} \Rightarrow \text{if we take } a = 0.25 \Rightarrow \text{L.R.} = 4$$

Table 2
Calculated expressions for $P(M_\alpha | H_p)$

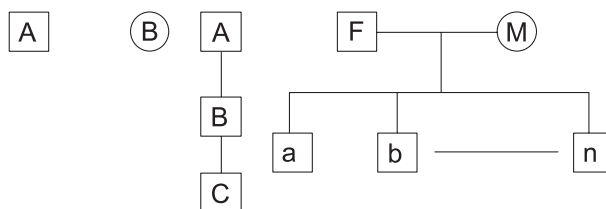|	$P(M_a)$	$P(M_{ab})$	$P(M_{abc})$	$P(M_{abcd})$
Genetically unrelated	a^4	$2ab[2(a+b)^2 - ab]$	$12abc(a+b+c)$	$24abcd$
Parent–offspring	a^3	$3ab(a+b)$	$6abc$	–
Full sibs	$(1/4)a^2(1+a)^2$	$(1/2)ab[2(a+b)^2 + 3(a+b) - ab + 1]$	$3abc(a+b+c+1)$	$6abcd$
Uncle–nephew, half sibs, grandparent–grandchild	$(1/2)a^3(1+a)$	$(1/2)ab[4(a+b)^2 - 2ab + 3(a+b)]$	$3abc[2(a+b+c)+1]$	$12abcd$
First cousins	$(1/4)a^3(1+3a)$	$(3/4)ab[4(a+b)^2 - 2ab + a + b]$	$(3/2)abc[6(a+b+c)+1]$	$18abcd$

Fig. 1. Examples of equivalent genetic configurations.

$$\text{L.R.}_{a,b} = \frac{3(a+b)}{2[2(a+b)^2 - ab]} \Rightarrow \text{if we take } a = 0.3, b = 0.4 \Rightarrow \text{L.R.} = 1.22$$

$$\text{L.R.}_{a,b,c} = \frac{1}{2(a+b+c)} \Rightarrow \text{if we take } a = 0.10, b = 0.15, c = 0.20 \Rightarrow \text{L.R.} = 5.42$$

With these values, the total likelihood ratio is, assuming independence, 26.45.

3. Discussion

The deduced expressions are useful to analyse forensic casework in which it is presumed that contributors are related. We still have to determine the minimum threshold for the L.R. suitable to accept the hypothesis H_p, a problem which is still open in the bayesian evaluation of results from forensic genetic analyses. There is, however, one relatively important restriction when the circumstances of the case are unknown. The following configurations would give identical results, preventing their distinction: two non-related people; parents and offspring; grandparent, parent and offspring... Generally speaking, all those cases having the same number of independent alleles—i.e. non inherited by a contributor from another—cannot be discriminated (Fig. 1).

Acknowledgements

References

[1] I.W. Evett, B.S. Weir, Interpreting DNA Evidence: Statistical Genetics for Forensic Science, Sinauer, Saunderland, MA, 1998.
[2] B.S. Weir, C.M. Triggs, L. Starling, L.I. Stowell, K.A.J. Walsh, J.S. Buckleton, Interpreting DNA mixtures, J. Forensic Sci. 42 (1997) 113–122.
[3] B.S. Weir, Genetic Data Analysis II, Sinauer, Sunderland, MA, 1996.
[4] B.S. Weir, Forensics, in: D.J. Balding, M. Bishop, C. Cannings (Eds.), Handbook of Statistical Genetics, Wiley, England, 2001, pp. 721–740.
[5] C.G.G. Aitken, Statistics and the Evaluation of Evidence for Forensic Scientists, Wiley, New York, 1995.
[6] B. Robertson, G.A. Vignaux, Interpreting Evidence: Evaluating Forensic Science in the Courtroom, Wiley, Chichester, 1995.