

# Chapter 6

## Individual Identification

### Introduction

Paternity testing  
Forensic testing

### Baye's Theorem

$$\Pr(C | E) = \frac{\Pr(E | C)\Pr(C)}{\Pr(E | C)\Pr(C) + \Pr(E | \bar{C})\Pr(\bar{C})}$$

$$\frac{\Pr(C | E)}{\Pr(\bar{C} | E)} = \frac{\Pr(E | C)}{\Pr(E | \bar{C})} \times \frac{\Pr(C)}{\Pr(\bar{C})}$$



Paternity index L

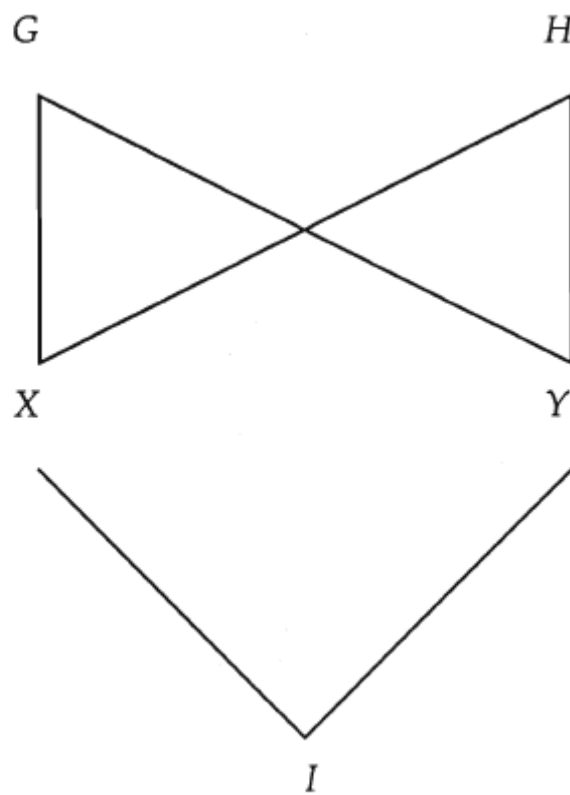
## Inbreed and Relatedness

IBD : identical by descent

F and :

$$F_I = \theta_{XY}$$

$$\theta_{XX} = \frac{1}{2}(1 + F_X)$$



**Figure 6.1** *I* is offspring of full sibs *X*, *Y*

$$\begin{aligned} F_I = \theta_{XY} &= \frac{1}{4}(\theta_{GG} + \theta_{GH} + \theta_{HG} + \theta_{HH}) \\ &= \frac{1}{8}(1 + F_G) + \frac{1}{2}\theta_{GH} + \frac{1}{8}(1 + F_H) \\ &= \frac{1}{4} \end{aligned}$$

$$F_I \; = \; \sum_i \left(\frac{1}{2}\right)^{n_i} \left(1 + F_{A_i}\right)$$

Descent relationship among alleles for two individuals  $X$  and  $Y$  with alleles for two individuals  $a, b$  and  $c, d$ .

Table 6.1.

Alleles ibd*	Probability	
	General	Full sibs <sup>§</sup>
none	$\delta_0$	1/4
$a \equiv b$	$\delta_{ab}$	0
$c \equiv d$	$\delta_{cd}$	0
$a \equiv c$	$\delta_{ac}$	1/4
$a \equiv d$	$\delta_{ad}$	0
$b \equiv c$	$\delta_{bc}$	0
$b \equiv d$	$\delta_{bd}$	1/4
$a \equiv b \equiv c$	$\delta_{abc}$	0
$a \equiv b \equiv d$	$\delta_{abd}$	0
$a \equiv c \equiv d$	$\delta_{acd}$	0
$b \equiv c \equiv d$	$\delta_{bcd}$	0
$a \equiv b, c \equiv d$	$\delta_{ab.cd}$	0
$a \equiv c, b \equiv d$	$\delta_{ac.bd}$	1/4
$a \equiv d, b \equiv c$	$\delta_{ad.bc}$	0
$a \equiv b \equiv c \equiv d$	$\delta_{abcd}$	0

\* Alleles not specified are not ibd.

§  $a, c$  from mother:  $b, d$  from father

**Table 6.2** Joint genotypic probabilities for two individuals  $X(a, b)$  and  $Y(c, d)$ .

$X$	$Y$	Probability
$AA$	$AA$	$\delta_{abcd}p_A + (\delta_{abc} + \delta_{abd} + \delta_{acd} + \delta_{bcd})p_A^2$ $+ (\delta_{ab.cd} + \delta_{ac.bd} + \delta_{ad.bc})p_A^2 + (\delta_{ab} + \delta_{ac} + \delta_{ad} + \delta_{bc} + \delta_{bd} + \delta_{cd})p_A^3 + \delta_0 p_A^4$ $\frac{1}{4}p_A^2 + p_A^3 + \frac{1}{4}p_A^4 = \frac{1}{4}p_A^2(1 + 2p_A + p_A^2)$
$AA$	$Aa$	$(\delta_{abc} + \delta_{abd})p_A p_a + (2\delta_{ab} + \delta_{ac} + \delta_{bc} + \delta_{ad} + \delta_{bd})p_A^2 p_a + 2\delta_0 p_A^3 p_a$
$Aa$	$AA$	$(\delta_{acd} + \delta_{bcd})p_A p_a + (2\delta_{cd} + \delta_{ac} + \delta_{bc} + \delta_{ad} + \delta_{bd})p_A^2 p_a + 2\delta_0 p_A^3 p_a$
$AA$	$aa$	$\delta_{ab.cd}p_A p_a + \delta_{ab}p_A p_a^2 + \delta_{cd}p_A^2 p_a + \delta_0 p_A^2 p_a^2$
$aa$	$AA$	$\delta_{ab.cd}p_A p_a + \delta_{cd}p_A p_a^2 + \delta_{ab}p_A^2 p_a + \delta_0 p_A^2 p_a^2$
$Aa$	$Aa$	$2(\delta_{ac.bd} + \delta_{ad.bc})p_A p_a + (\delta_{ac} + \delta_{ad} + \delta_{bc} + \delta_{bd})(p_A p_a^2 + p_A^2 p_a) + 4\delta_0 p_A^2 p_a^2$
$aa$	$Aa$	$(\delta_{abc} + \delta_{abd})p_A p_a + (2\delta_{ab} + \delta_{ac} + \delta_{bc} + \delta_{ad} + \delta_{bd})p_a^2 p_A + 2\delta_0 p_a^3 p_A$
$Aa$	$aa$	$(\delta_{acd} + \delta_{bcd})p_A p_a + (2\delta_{cd} + \delta_{ac} + \delta_{bc} + \delta_{ad} + \delta_{bd})p_a^2 p_A + 2\delta_0 p_a^3 p_A$
$aa$	$aa$	$\delta_{abcd}p_a + (\delta_{abc} + \delta_{abd} + \delta_{acd} + \delta_{bcd})p_a^2$ $+ (\delta_{ab.cd} + \delta_{ac.bd} + \delta_{ad.bc})p_a^2 + (\delta_{ab} + \delta_{ac} + \delta_{ad} + \delta_{bc} + \delta_{bd} + \delta_{cd})p_a^3 + \delta_0 p_a^4$

An example:  $X$  and  $Y$  are sibs.

## Conditional genotypic frequencies

$$\Pr(X = A_i A_j | Y = A_i A_j) = \frac{\Pr(X = A_i A_j, Y = A_i A_j)}{\Pr(Y = A_i A_j)}$$

$\theta, \gamma, \delta$  and  $\Delta$ .

$$\delta_0 = 1 - 6\theta + 8\gamma + 3\Delta - 6\delta$$

$$\delta_{ab} = \dots = \delta_{bd} = \delta_{cd} = \theta - 2\gamma - \Delta + 2\delta$$

$$\delta_{abc} = \dots = \delta_{acd} = \delta_{bcd} = \gamma - \delta$$

$$\delta_{ab.cd} = \dots = \delta_{ab.cd} = \Delta - \delta$$

$$\delta_{abcd} = \delta$$

## Paternity Testing

In Paternity disputes it is necessary to determine whether a particular man is the father of a particular child. Classical considerations of such questions were limited to excluding a man from paternity of a child if the man did not have the child's paternal allele at some loci, or, if the paternal allele can not be determined, if the man has neither of the child's alleles.

## Paternity exclusion probability with codominant loci:

Exclusion Probabilities of possible fathers for all possible mother-child combinations are shown in Table 6.3.

Mother		Child		Excluded Man	
Type	Prob. <sup>1</sup>	Type	Probability. <sup>2</sup>	Genotype	Probability <sup>3</sup>
$A_u A_u$	$P_u^2$	$A_u A_u$	$P_u$	$A_w A_x, w, x \neq u$	$(1 - P_u)^2$
		$A_u A_v$	$P_v$	$A_w A_x, w, x \neq v$	$(1 - P_v)^2$
$A_u A_v$ $u \neq v$	$2P_u P_v$	$A_u A_u$	$P_u / 2$	$A_w A_x, w, x \neq u$	$(1 - P_u)^2$
		$A_v A_v$	$P_v / 2$	$A_w A_x, w, x \neq v$	$(1 - P_v)^2$
		$A_u A_v$	$(P_u + P_v) / 2$	$A_w A_x, w, x \neq u, v$	$(1 - P_u - P_v)^2$
		$A_u A_y^*$	$P_y / 2$	$A_w A_x, w, x \neq y$	$(1 - P_y)^2$
		$A_v A_y^*$	$P_y / 2$	$A_w A_x, w, x \neq y$	$(1 - P_y)^2$

<sup>1</sup> Probability of mother's genotype

<sup>2</sup> Probability of child's genotype given mother's genotype

<sup>3</sup> Probability of excluded's genotype

\*  $y \neq u, v$

An assessment of how good this locus is for being able to exclude a random man from paternity, assuming that he is not the father of the child, is given by summing the joint probabilities of all the mother-child-excluded man combinations shown in the Table. For one locus, the exclusion probability

$$Q_l = \sum_u p_u (1 - p_u)^2 - \frac{1}{2} \sum_u \sum_{v \neq u} p_u^2 p_v^2 (4 - 3p_u - 3p_v)$$

and will be maximized when all  $V$  alleles at the locus have frequency  $1/V$ .

Overall probability for  $l$  loci,

$$Q = 1 - \prod_l (1 - Q_l)$$

# **Paternity exclusion probabilities with dominant loci:**

For ABO system,

$$Q = pQ(1-p)^4 + q(1-p)^4 + pqr^2(3-r)$$

**Table 6.4** Paternity exclusion arrangements for the three-allele *ABO* system.

		Mother			
Excluded Men		<i>A</i>	<i>B</i>	<i>AB</i>	<i>O</i>
Child	<i>A</i>	None	<i>B, O</i>	None	<i>B</i>
	<i>B</i>	<i>A, O</i>	None	None	<i>A</i>
	<i>AB</i>	<i>A, O</i>	<i>B, O</i>	<i>O</i>	Impossible
	<i>O</i>	<i>AB</i>	<i>AB</i>	Impossible	<i>AB</i>



**Paternity index:**

Unlike the exclusion probability, the paternal index is based on the actual genotypes of the mother, child and alleged father. Bayes' theorem provides

$$\frac{\Pr(\text{man is father} | E)}{\Pr(\text{man is not father} | E)} = \frac{\Pr(E | \text{man is father})}{\Pr(E | \text{man is not father})} \times \frac{\Pr(\text{man is father})}{\Pr(\text{man is not father})}$$

$$\begin{aligned} \frac{\Pr(C | E)}{\Pr(\bar{C} | E)} &= L \times \frac{\Pr(C)}{\Pr(\bar{C})} \\ \Pr(C | E) &= \frac{L \times \Pr(C)}{L \Pr(C) + (1 - \Pr(C))} \end{aligned}$$

Discussion of paternity indices is quite valid, but **debate begins when particular values are given to the prior probabilities.**

Example :  $M=A_1A_3$ ,  $O=A_1A_2$ ,  $AF=A_2A_4$

$$\begin{aligned} \Pr(E | C) &= \Pr(O, M, AF | C) = \Pr(O | M, AF, C) \Pr(M, AF | C) \\ &= \frac{1}{2} \times \frac{1}{2} \times \Pr(M, AF) \end{aligned}$$

$$\begin{aligned} \Pr(E | \bar{C}) &= \Pr(O, M, AF | \bar{C}) = \Pr(O | M, AF, \bar{C}) \Pr(M, AF | \bar{C}) \\ &= \Pr(O | M) \Pr(M, AF) = \frac{1}{2} \times p_2 \times \Pr(M, AF) \end{aligned}$$

$$\begin{aligned} \Pr(E | \bar{C}) &= \Pr(O, M, AF | \bar{C}) = \Pr(O | M, AF, \bar{C}) \Pr(M, AF | \bar{C}) \\ &= \Pr(O | M) \Pr(M, AF) = \frac{1}{2} \times p_2 \times \Pr(M, AF) \end{aligned}$$

$$L = \frac{1}{2p_2}$$

Table 6.5 : Paternity index calculations for a two-allele locus (Elston 1986).

<i>E</i>			$\Pr(E C)$	$\Pr(E \bar{C})$	<i>L</i>	
Mother	Child	Alleged Father				
AA	AA	AA	$p_A^4$	$p_A^5$	$1/p_A$	$\rightarrow p_A \cdot (p_A^2 \times p_A)$
		Aa	$p_A^3 p_a$	$2p_A^4 p_a$	$1/2p_A$	
		aa	0	$p_A^3 p_a^2$	0	
	Aa	AA	0	$p_A^4 p_a$	0	$\rightarrow \frac{1}{2}(p_A^2) > p_A p_a$ by pg 243
		Aa	$p_A^3 p_a$	$2p_A^3 p_a^2$	$1/2p_a$	
		aa	$p_A^2 p_a^2$	$p_A^2 p_a^3$	$1/p_a$	
Aa	AA	AA	$p_A^3 p_a$	$p_A^4 p_a$	$1/p_A$	
		Aa	$p_A^2 p_a^2$	$2p_A^3 p_a^2$	$1/2p_A$	
		aa	0	$p_A^2 p_a^3$	0	
	Aa	AA	$p_A^3 p_a$	$p_A^3 p_a$	1	
		Aa	$2p_A^2 p_a^2$	$2p_A^2 p_a^2$	1	
		aa	$p_A p_a^3$	$p_A p_a^3$	1	
	aa	AA	0	$p_A^3 p_a^2$	0	
		Aa	$p_A^2 p_a^2$	$2p_A^2 p_a^3$	$1/2p_a$	
		aa	$p_A p_a^3$	$p_A p_a^4$	$1/p_a$	
aa	Aa	AA	$p_A^2 p_a^2$	$p_A^3 p_a^2$	$1/p_A$	
		Aa	$p_A p_a^3$	$2p_A^2 p_a^3$	$1/2p_A$	
		aa	0	$p_A p_a^4$	0	
	aa	AA	0	$p_A^2 p_a^3$	0	
		Aa	$p_A p_a^3$	$2p_A p_a^4$	$1/2p_a$	
		aa	$p_a^4$	$p_a^5$	$1/p_a$	

## Paternity index for relatives:

C : the alleged father (AF) with genotype  $A_i A_j$  is the true father.

$\bar{C}$  : his brother (B) is the true father.

$$\begin{aligned}\Pr(E | C) &\propto \Pr(AF \rightarrow A_i | AF = A_i A_j) \\ &= \frac{1}{2}\end{aligned}$$

$$\begin{aligned}\Pr(E | \bar{C}) &\propto \Pr(B \rightarrow A_i | AF = A_i A_j) \\ &= \Pr(B \rightarrow A_i, AF = A_i A_j) \Pr(AF = A_i A_j) \\ &= \left[ \Pr(B = A_i A_i, AF = A_i A_j) + \frac{1}{2} \Pr(B = A_i A_i, AF = A_i A_j) \right. \\ &\quad \left. + \frac{1}{2} \sum_{k \neq i, j} \Pr(B = A_i A_k, AF = A_i A_j) \right] / \Pr(AF = A_i A_j) \\ &= \frac{1}{4} p_i (1 + p_i) + \frac{1}{8} (1 + p_i + p_j + 2 p_i p_j) \\ &\quad + \frac{1}{8} (1 - p_i - p_j) (1 + 2 p_i) \\ &= \frac{1 + 2 p_i}{4}\end{aligned}$$

The paternity index is

$$L = \frac{(1/2)}{(1 + 2 p_i)/4} = \frac{2}{1 + 2 p_i}$$

# Forensic Testing

C : the suspect provided the crime scene material

$\bar{C}$  : someone else provided the material

P : perpetrator

S : suspect

E : S and P are the same person

$$L = \frac{\Pr(S = A, P = A | C)}{\Pr(S = A, P = A | \bar{C})}$$

Suspect-anchored

$$\begin{aligned} L &= \frac{\Pr(P = A | C = A, C) \Pr(S = A | C)}{\Pr(P = A | S = A, \bar{C}) \Pr(S = A | \bar{C})} \\ &= \frac{\Pr(P = A | S = A, C)}{\Pr(P = A | S = A, \bar{C})} \end{aligned}$$

If prior probabilities are introduced,

$$\frac{\Pr(C | E)}{\Pr(\bar{C} | E)} = L \times \frac{\Pr(C)}{\Pr(\bar{C})}$$

If experimental error is ignored,

$$L = \frac{1}{\Pr(P = A | S = A, \bar{C})}$$

The simplest thing is to assume the probabilities of  $S$  and  $P$  and S having profile A are independent when they are different people. Then

$$L = \frac{1}{\Pr(P = A)}$$

**Estimation of profile frequency:**

$$\tilde{P}_l = \begin{cases} \tilde{P}_{l_1}^2, & l_1 = l_2 \\ 2\tilde{P}_{l_1} \tilde{P}_{l_2}, & l_1 \neq l_2 \end{cases}$$

$$\tilde{P} = \prod_l \tilde{P}_l$$

$$Var(\tilde{P}) = \left( \prod_l P_l \right)^2 \left[ \prod_l \left( 1 + \frac{Var(\tilde{P}_l)}{P_l^2} \right) - 1 \right]$$

**Likelihood ratio for members of the same Population:**

$$\Pr(S = A_i A_i \mid P = A_i A_i) = \frac{[p_i + \theta(2 - p_i)][p_i + \theta(3 - p_i)]}{[1 + \theta][1 + 2\theta]}$$

$$\Pr(S = A_i A_j \mid P = A_i A_j) = \frac{2[p_i + \theta(1 - p_i)][p_j + \theta(1 - p_j)]}{(1 + \theta)(1 + 2\theta)}$$

The above expressions are exact for populations at equilibrium under evolutionary forces, and as approximations they often perform well before equilibrium.

Table 6.6

## Likelihood ratio for relatives:

Unrelated individuals have a very low probability of sharing the same genetic profile, but the probability increases for relatives.

**Table 6.7** Probability that the perpetrator  $P$  has a genotype given that a suspect  $S$  has that genotype, when they are related. Likelihood ratio ( $L$ ) assumes allele frequencies of 0.1.

Genotype A	Relationship	$\Pr(P = A   S = A)$	$L$
$A_i A_j$	Full brothers	$(1 + p_i + p_j + 2p_i p_j)/4$	3.3
	Father and son	$(p_i + p_j)/2$	10.0
	Half brothers	$(p_i + p_j + 4p_i p_j)/4$	16.7
	Uncle and nephew	$(p_i + p_j + 4p_i p_j)/4$	16.7
	First cousins	$(p_i + p_j + 12p_i p_j)/8$	25.0
	Unrelated	$2p_i p_j$	50.0
$A_i A_i$	Full brothers	$(1 + p_i)^2/4$	3.3
	Father and son	$p_i$	10.0
	Half brothers	$p_i(1 + p_i)/2$	18.2
	Uncle and nephew	$p_i(1 + p_i)/2$	18.2
	First cousins	$p_i(1 + 3p_i)/4$	30.8
	Unrelated	$p_i^2$	100.0

→ different from (6.3)

# Mixed Samples

## General situation:

Evidentiary samples sometimes contain DNA from more than one person.

$\{ e \}$ : a set of alleles in the sample.

$\{ k \}$ : a set of alleles from known contributors.

$\{ u \}$ : a set of alleles containing unknown contributors.

$P_{\Phi}$  : probability of no unknown contributor.

## Calculating probabilities:

### Profiles with one allele:

$$P_1(a|a) = P_a^2. \quad P_x(a|a) = P_a^{2x}$$

$$P_x(\Phi|a) = P_a^{2x}$$

**Profiles with two alleles:** The probability that one unknown contributor has allele  $a$ , and no allele other than  $a$  or  $b$ , is

$$P_1(a|ab) = P_a^2 + 2P_aP_b = (P_a + P_b)^2 - P_b^2$$

$$P_x(a|ab) = (P_a + P_b)^{2x} - P_b^{2x}$$

$$P_x(\Phi|ab) = (P_a + P_b)^{2x}$$

If there are no known contributors,

$$P_x(ab|ab) = (P_a + P_b)^{2x} - P_a^{2x} - P_b^{2x}$$

### Profiles with three alleles and four alleles (pg 223):

**Example :**

$\{e\} = abcd$  and  $V = ab$ ,  $S = cd$

Under  $C$ ,  $P_\Phi = 1$  or  $P_1(cd | abcd)$

Under  $\bar{C}$

$$(1) P_1(cd | abcd) = 2P_c P_d ,$$

$$L = \frac{P_\Phi}{P_1(cd | abcd)} = \frac{1}{2P_c P_d}$$

$$(2) P_2(abcd | abcd) = 24P_a P_b P_c P_d$$

$$L = \frac{P_\Phi}{P_2(abcd | abcd)} = \frac{1}{24P_a P_b P_c P_d}$$

$$(3) \text{ For } C = P_1(cd | abcd) ,$$

$$P_2(abcd | abcd) = 24P_a P_b P_c P_d$$

$$L = \frac{P_1(cd | abcd)}{P_2(abcd | abcd)} = \frac{2P_c P_d}{24P_a P_b P_c P_d} = \frac{1}{12P_a P_b}$$

**A more complex example:** Sample  $E$  has only three alleles  $abc$  and includes contributions from a victim and a perpetrator. If a victim has genotype  $ab$  and a suspect has profile  $aa$ , then the explanation  $C$  including the suspect still requires another contributor for allele  $c$ .  $\bar{C}$  may still include the victim but exclude the suspect, and includes some unknown contributors. For example, if  $\bar{C}$  supposes two known contributors

$$\begin{aligned} L &= \frac{P_1(c | abc)}{P_2(c | abc)} \\ &= \frac{1}{(p_a + p_b + p_c)^2 - (p_a + p_b)^2} \end{aligned}$$



# Error Rates

**Homework:** (1) Derive  $Q$  for ABO blood-type system on page 211 using Table 6.4. (2) Determine the coefficients for the uncle and niece, parent and offspring, grandparent and grand-offspring, and first cousins. Assume all individuals are noninbred. (3) Calculate  $F_X$

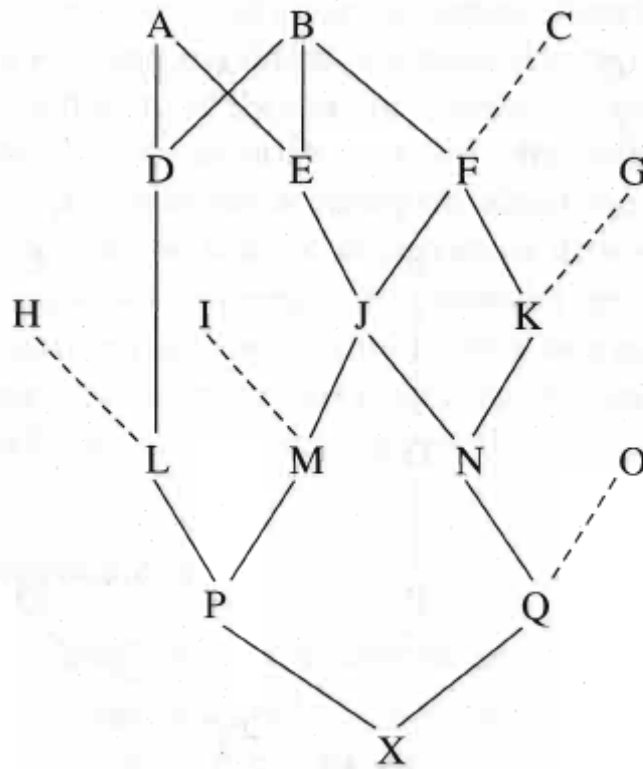


Fig. 5.3