

# Forensic Genetics

Module 19 – Topic 5 Answers

# Balding Sampling Formula

For a case, suppose  $n$  alleles have been seen among the known and typed contributors, and  $n_A$  of these are of type  $A$ .

If allele  $A$  in the evidence profile must be contributed by an unknown contributor under some hypothesis, the probability of that allele is

$$\Pr(A|n_A \text{ of } n) = \frac{n_A\theta + (1 - \theta)p_A}{1 + (n - 1)\theta}$$

The  $A$  allele is then added to the  $n$  known alleles, and the probability of the next required allele, say  $B$ , is then calculated (if there were  $n_B$  among the original  $n$  alleles):

$$\Pr(B|n_B \text{ of } n + 1) = \frac{n_B\theta + (1 - \theta)p_B}{1 + (n + 1 - 1)\theta}$$

# Population Structure Exercise

Use the Balding sampling formula to find a formula for:

		$\theta \neq 0$	$\theta = 0$
$n = 0, n_A = 0$	$\Pr(A)$	$p_A$	$p_A$
$n = 1, n_A = 1$	$\Pr(A A)$	$\theta + (1 - \theta)p_A$	$p_A$
	$\Pr(B A)$	$(1 - \theta)p_B$	$p_B$
$n = 2, n_A = 2$	$\Pr(A AA)$	$\frac{2\theta + (1 - \theta)p_A}{1 + \theta}$	$p_A$
	$\Pr(B AA)$	$\frac{(1 - \theta)p_B}{1 + \theta}$	$p_B$
$n = 2, n_A = n_B = 1$	$\Pr(A AB)$	$\frac{\theta + (1 - \theta)p_A}{1 + \theta}$	$p_A$
	$\Pr(B AB)$	$\frac{\theta + (1 - \theta)p_B}{1 + \theta}$	$p_B$
	$\Pr(C AB)$	$\frac{(1 - \theta)p_C}{1 + \theta}$	$p_C$

# Population Structure Exercise

Use the Balding sampling formula to evaluate:

		$p = 0.10$		$p = 0.01$	
		$\theta = 0$	$\theta = 0.01$	$\theta = 0$	$\theta = 0.01$
$n = 0, n_A = 0$	$\Pr(A)$	0.10	0.10	0.01	0.01
$n = 1, n_A = 1$	$\Pr(A A)$	0.10	0.109	0.01	0.0199
	$\Pr(B A)$	0.10	0.099	0.01	0.0099
$n = 2, n_A = 2$	$\Pr(A AA)$	0.10	0.118	0.01	0.030
	$\Pr(B AA)$	0.10	0.098	0.01	0.0098
$n = 2, n_A = n_B = 1$	$\Pr(A AB)$	0.10	0.108	0.01	0.0197
	$\Pr(B AB)$	0.10	0.108	0.01	0.0197
	$\Pr(C AB)$	0.10	0.098	0.01	0.0098

# Joint Probabilities

The joint probability of a set of alleles is the probability of the first, times the probability of the second given the first, times the probability of the third given the first two, etc:

$$\Pr(AA) = \Pr(A) \Pr(A|A)$$

$$\Pr(AB) = \Pr(A) \Pr(B|A)$$

$$\Pr(AAA) = \Pr(A) \Pr(A|A) \Pr(A|AA)$$

$$\Pr(AAB) = \Pr(A) \Pr(A|A) \Pr(B|AA)$$

$$\Pr(ABC) = \Pr(A) \Pr(B|A) \Pr(C|AB)$$

The events can be considered in any order.

# Balding-Nichols Formula

We can then derive the Balding-Nichols match probabilities. For homozygotes:

$$\Pr(AAAA) = \Pr(A) \Pr(A|A) \Pr(A|AA) \Pr(A|AAA)$$

$$\Pr(AA) = \Pr(A) \Pr(A|A)$$

$$\begin{aligned} \Pr(AA|AA) &= \frac{\Pr(AAAA)}{\Pr(AA)} \\ &= \Pr(A|AA) \Pr(A|AAA) \\ &= \frac{2\theta + (1 - \theta)p_A}{1 + \theta} \times \frac{3\theta + (1 - \theta)p_A}{1 + 2\theta} \end{aligned}$$