Forensic Genetics

Module 15 – Section 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:

$$\begin{aligned}
\Pr(AAAA) &= \Pr(A)\Pr(A|A)\Pr(A|AA)\Pr(A|AAA) \\
\Pr(AA) &= \Pr(A)\Pr(A|A)
\end{aligned}$$

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