

YHRD kinship formula

L : set of loci with typed results for ancestor and offspring

s_l : allelic difference between ancestor and offspring at locus l

$\mu_{l,i}$: i -step mutation rate at loci l

$\frac{\mu_{l,i}}{2}$: probability of exactly one i -step mutation at locus l

$\left(\frac{\mu_{l,i}}{2}\right)^{s_l}$: probability of exactly one s_l i -step mutations at locus l

$1 - \mu_l$: probability of one non-mutation at locus l

$(1 - \mu_l)^{s_l}$: probability of s_l non-mutations at locus l

H_0 : hypothesis that ancestor and offspring are related

H_1 : hypothesis that ancestor and offspring are unrelated

m : number of transmission events under H_0 and H_1

$\binom{m}{s_l}$: binomial coefficient ‘ m choose s_l ’

$$LR = \frac{P(E|H_0)}{P(E|H_1)}$$

$$LR_{\text{only 1-steps}} = \frac{\prod_{l \in L} \left[\binom{m}{s_l} \left(\left(\frac{\mu_l}{2} \right)^{s_l} (1 - \mu_l)^{m - s_l} \right) \right]}{f(\text{offspring})} \quad (2)$$

Example

ancestor haplotype : $\{L_1 : \text{"10"}, L_2 : \text{"11"}\}$

offspring haplotype : $\{L_1 : \text{"10"}, L_2 : \text{"12"}\}$

$f(\text{offspring}) : 1/100$; frequency of offspring's haplotype

$m : 1$

$\mu : \{L_1 : \{1 : 0.1, 2 : 0.05, 3 : 0.01\}, L_2 : \{1 : 0.1, 2 : 0.05, 3 : 0.01\}\}$

$(1 - \mu) : \{L_1 : 0.84, L_2 : 0.84\}$

$L : \{L_1, L_2\}$

$s : \{L_1 : 0, L_2 : 1\}$

$$P(E|H_0) = \prod_{l \in L} \sum_{S_{l,i} \in p(s_l)} \left[\binom{m}{\text{rank}(S_{l,i})} \left(\prod_{s_{l,i,j} \in S_{l,i}} \left(\frac{\mu_{l,s_{l,i,j}}}{2} \right) \times (1 - \mu_l)^{m - \text{rank}(S_{l,i})} \right) \right] \quad \Big| \quad L = \{L_1, L_2\}$$

$$P(E|H_0) = \sum_{S_{L_1,i} \in p(s_{L_1})} [\dots] \times \sum_{S_{L_2,i} \in p(s_{L_2})} [\dots] \quad \Big| \quad p(s_{L_1}) = \{\emptyset\} \text{ and } p(s_{L_2}) = \{\{1\}\}$$

$$\sum_{S_{L_1,i} \in p(s_{L_1})} [\dots] = \left[\binom{m}{\text{rank}(\emptyset)} \left(\prod_{s_{L_1,0,j} \in \emptyset} \left(\frac{\mu_{L_1, s_{L_1,0,j}}}{2} \right) \times (1 - \mu_{L_1})^{m - \text{rank}(\emptyset)} \right) \right]_{S_{L_1,0} = \emptyset}$$

$$\sum_{S_{L_1,i} \in p(s_{L_1})} [\dots] = \left[\binom{1}{0} (1 \times (1 - \mu_{L_1})^1) \right]_{S_{L_1,0} = \emptyset} = 1 - \mu_{L_1}$$

$$\sum_{S_{L_2,i} \in p(s_{L_2})} [\dots] = \left[\binom{m}{\text{rank}(\{1\})} \left(\prod_{s_{L_2,0,j} \in \{1\}} \left(\frac{\mu_{L_2, s_{L_2,0,j}}}{2} \right) \times (1 - \mu_{L_2})^{m - \text{rank}(\{1\})} \right) \right]_{S_{L_2,0} = \{1\}}$$

$$\prod_{s_{L_2,0,j} \in \{1\}} \left(\frac{\mu_{L_2, s_{L_2,0,j}}}{2} \right) = \left(\frac{\mu_{L_2, s_{L_2,0,0}}}{2} \right)_{s_{L_2,0,0} = 1} = \left(\frac{\mu_{L_2,1}}{2} \right)$$

$$\sum_{S_{L_2,i} \in p(s_{L_2})} [\dots] = \left[\binom{1}{1} \left(\left(\frac{\mu_{L_2,1}}{2} \right) \times (1 - \mu_{L_2})^0 \right) \right] = \frac{\mu_{L_2,1}}{2}$$

$$P(E|H_0) = \sum_{S_{L_1,i} \in p(s_{L_1})} [\dots] \times \sum_{S_{L_2,i} \in p(s_{L_2})} [\dots] = (1 - \mu_{L_1}) \times \frac{\mu_{L_2,1}}{2} = 0.84 \times 0.05 = 0.042$$

$$P(E|H_1) = f(\text{offspring}) = 0.01$$

$$LR = \frac{P(E|H_0)}{P(E|H_1)} = \frac{0.042}{0.01} = 4.2$$