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Pedigree Formulas

Let B, C be individuals. Let A be a common ancestor in the ancestor set $\mathcal{A}$. $\mathcal{P}(\cdot)$ is a function that takes as input a common ancestor A and outputs the set of paths $\{p_A\}$ by which B and C are connected through A. $n(\cdot)$ is a counting function that takes as input a path and reports the number of edges (meioses) in the path.

Kinship

- path counting: $\psi(B,C) = \sum_{A\in\mathcal{A}} \sum_{p_A\in\mathcal{P}(A)} (1+f_A)(1/2)^{n(p_A)+1}$
- condensed IBD states: $\psi(B,C) = \Delta_1 + 1/2 \cdot (\Delta_3 + \Delta_5 + \Delta_7) + 1/4 \cdot \Delta_8$
- (non-inbred) gene identity states: $\psi(B,C) = 1/2 \cdot \kappa_2 + 1/4 \cdot \kappa_1 + 0 \cdot \kappa_0 = (2\kappa_2 + \kappa_1)/4$
- parental kinships: $\psi(C,B) = 1/4 \cdot (\psi(M_C, M_B) + \psi(F_C, F_B) + \psi(M_C, F_B) + \psi(F_C, M_B))$

(Non-inbred) Gene Identity States

κ probabilities are for pedigrees without inbreeding.

- $\kappa_2 + \kappa_1 + \kappa_0 = 1$
- parental kinships: $\kappa_2(B,C) = \psi(M_C, M_B)\psi(F_C, F_B) + \psi(M_C, F_B)\psi(F_C, M_B)$
- condensed IBD states: $\kappa_2 = \Delta_7$, $\kappa_1 = \Delta_8$, $\kappa_0 = \Delta_9$

Inbreeding

- kinship: $f(B) = \psi(M_B, F_B)$
- condensed IBD states: $f(B) = \Delta_1 + \Delta_2 + \Delta_3 + \Delta_4$ or $f(C) = \Delta_1 + \Delta_2 + \Delta_5 + \Delta_6$

Conditional Probabilities

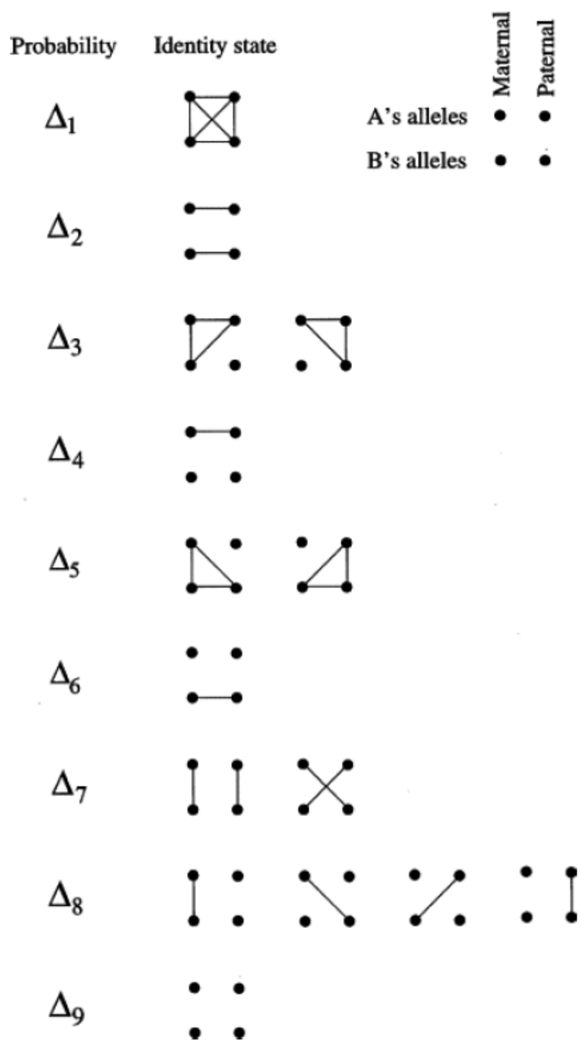
- Conditional probabilities: $P(D|E) = \frac{P(D,E)}{P(E)}$, $P(D|E,F) = \frac{P(D,E|F)}{P(E|F)}$
- Law of total probability: $P(D) = \sum_i P(D, E_i) = \sum_i P(D|E_i)P(E_i)$
- $P(G_C|\text{tree}) = P(G_C|\text{IBD } 0)(1 - f(C)) + P(G_C|\text{IBD } 1)(f(C))$
- $P(G_B, G_C|\text{tree}) = \frac{\sum_{\text{IBD states}} P(G_B, G_C|\text{IBD state})P(\text{IBD state}|\text{tree})}{P(G_C|\text{tree})}$

Relationships Table

Note that the notation ϕ is ψ and indices i, j are individuals B, C from the previous page.

RELATIONSHIP	Δ_1	Δ_2	Δ_3	Δ_4	Δ_5	Δ_6	Δ_7	Δ_8	Δ_9	ϕ_{ij}
Self	0	0	0	0	0	0	1	0	0	$\frac{1}{2}$
Parent-offspring	0	0	0	0	0	0	0	1	0	$\frac{1}{4}$
Half sibs	0	0	0	0	0	0	0	$\frac{1}{2}$	$\frac{1}{2}$	$\frac{1}{8}$
Full sibs/dizygotic twins	0	0	0	0	0	0	$\frac{1}{4}$	$\frac{1}{2}$	$\frac{1}{4}$	$\frac{1}{4}$
Monozygotic twins	0	0	0	0	0	0	1	0	0	$\frac{1}{2}$
First cousins	0	0	0	0	0	0	0	$\frac{1}{4}$	$\frac{3}{4}$	$\frac{1}{16}$
Double first cousins	0	0	0	0	0	0	$\frac{1}{16}$	$\frac{6}{16}$	$\frac{9}{16}$	$\frac{1}{8}$
Second cousins	0	0	0	0	0	0	0	$\frac{1}{16}$	$\frac{15}{16}$	$\frac{1}{64}$
Uncle-nephew	0	0	0	0	0	0	0	$\frac{1}{2}$	$\frac{1}{2}$	$\frac{1}{8}$
Offspring of sib-matings	$\frac{1}{16}$	$\frac{1}{32}$	$\frac{1}{8}$	$\frac{1}{32}$	$\frac{1}{8}$	$\frac{1}{32}$	$\frac{7}{32}$	$\frac{5}{16}$	$\frac{1}{16}$	$\frac{3}{8}$

IBD States


Probability
Identity state
Maternal
Paternal
A's alleles
B's alleles
Δ1
Δ2
Δ3
Δ4
Δ5
Δ6
Δ7
Δ8
Δ9

IBD Labels

Note that these IBD labels correspond to the IBD states on the preceding page. Jacquard's IBD states are for pairwise comparisons. IBD labeling extends to multi-individual comparisons, with the accompanying visual diagrams being IBD graphs.

ibd pattern B_1 p m	B_2 p m	*ibd* label	*ibd* group	state description: individuals autozygous	genes shared
• •	• •	1 1 1 1	1 1 1 1	B_1, B_2	4 genes *ibd*
• •	• ∘	1 1 1 2	1 1 1 2	B_1	3 genes *ibd*
• •	∘ •	1 1 2 1			
• ∘	• •	1 2 1 1	1 2 1 1	B_2	3 genes *ibd*
• ∘	∘ ∘	1 2 2 2			
• •	∘ ∘	1 1 2 2	1 1 2 2	B_1, B_2	none
• •	∘ †	1 1 2 3	1 1 2 3	B_1	none
• ∘	† †	1 2 3 3	1 2 3 3	B_2	none
• ∘	• ∘	1 2 1 2	1 2 1 2	none	2 genes
• ∘	∘ •	1 2 2 1			shared
• ∘	• †	1 2 1 3	1 2 1 3	none	1 gene
• ∘	† •	1 2 3 1			shared
• ∘	∘ †	1 2 2 3			
• ∘	† ∘	1 2 3 2			
• ∘	† ⋆	1 2 3 4	1 2 3 4	none	none

TABLE 3.1. *States of gene ibd among the four genes of two individuals*

References

- Thompson, E. A. (2000). Statistical inference from genetic data on pedigrees. IMS.
- https://brainder.org/tag/jacquard-coefficient/