Chapter 4: Genetic Identity Coefficients

§4.1. Kinship and inbreeding coefficients

- **Definition**

  **Identity by state (ibs)**: Two alleles are ibs if they are functionally the same.

  **Identity by descent (ibd)**: Two alleles are ibd if one is a physical copy of the other, or if they are both physical copies of the same ancestral allele.

  **Kinship coefficient** $\Phi_{ij}$: The kinship coefficient $\Phi_{ij}$ between two individuals $i$ and $j$ is the probability that an allele selected randomly from $i$ and an allele selected randomly from the same autosomal locus of $j$ are ibd.
**Inbreeding coefficient** \( f_i \): The inbreeding coefficient \( f_i \) of an individual \( i \) is the probability that his two alleles at any autosomal locus are ibd. If \( f_i > 0 \), \( i \) is said to be **inbred**.

**Relation between kinship and inbreeding coefficients:**

\[
\Phi_{ii} = \frac{1}{2}(1 + f_i), \quad f_i = \Phi_{kl},
\]

where \( k \) and \( l \) are parents of \( i \).

- **Calculation of kinship coefficients**
  
  - **Simple examples**

    *Parent-offspring.* \( \Phi_{ij} = 1/4 \).

    *Full sibs.* \( 1, 2: \) parents of \( 3, 4 \).

    \[
    \Phi_{34} = \frac{1}{2}\Phi_{31} + \frac{1}{2}\Phi_{32} = \frac{1}{4}.
    \]
**Half sibs.** 4: Child of 1 and 2; 5: Child of 2 and 3.

\[
\Phi_{45} = \frac{1}{2} \Phi_{42} + \frac{1}{2} \Phi_{43}
\]

\[
= \frac{1}{2} \times \frac{1}{4} + 0 = \frac{1}{8}.
\]

**First cousins.** 3,4: children of 1 and 2; 7: child of 3 and 5; 8: child of 4 and 6.

\[
\Phi_{78} = \frac{1}{2} \Phi_{74} + \frac{1}{2} \Phi_{76}
\]

\[
= \frac{1}{2} \Phi_{74} + 0
\]

\[
= \frac{1}{2} \left( \frac{1}{2} \Phi_{34} + \frac{1}{2} \Phi_{54} \right)
\]

\[
= \frac{1}{2} \left( \frac{1}{2} \Phi_{34} + 0 \right)
\]

\[
= \frac{1}{2} \times \frac{1}{4} = \frac{1}{16}.
\]
○ General algorithm

i) Any person should have either both or neither of his or her parents in the pedigree.

ii) Members in the pedigree are numbered in such a way that every parent precedes his or her children.

iii) The kinship coefficients between any two persons in the pedigree are computed in a symmetric matrix from left top downwards recursively.

Recursive calculation formulas:

a) For $\Phi_{ii}$: if $i$ is a founder, $\Phi_{ii} = 1/2$, otherwise, $\Phi_{ii} = \frac{1}{2} + \frac{1}{2} \Phi_{kl}$, where $k$ and $l$ are parents of $i$.

b) For $\Phi_{ij}$, $(i > j)$: if $i$ is a founder, $\Phi_{ij} = 0$, otherwise, $\Phi_{ij} = \frac{1}{2} \Phi_{jk} + \frac{1}{2} \Phi_{jl}$, where $k$ and $l$ are parents of $i$.

Basic Rule: Substitution of parental alleles for the allele of the child.
A Brother-sister mating example

\[ \Phi = \begin{pmatrix}
\frac{1}{2} & 0 & \frac{1}{4} & \frac{1}{4} & \frac{1}{4} & \frac{1}{4} \\
0 & \frac{1}{2} & \frac{1}{4} & \frac{1}{4} & \frac{1}{4} & \frac{1}{4} \\
\frac{1}{4} & \frac{1}{4} & \frac{1}{2} & \frac{1}{4} & \frac{1}{8} & \frac{1}{8} \\
\frac{1}{4} & \frac{1}{4} & \frac{1}{2} & \frac{1}{4} & \frac{1}{8} & \frac{1}{8} \\
\frac{1}{4} & \frac{1}{4} & \frac{1}{8} & \frac{1}{8} & \frac{3}{8} & \frac{3}{8} \\
\frac{1}{4} & \frac{1}{4} & \frac{1}{8} & \frac{1}{8} & \frac{3}{8} & \frac{3}{8}
\end{pmatrix} \]
A remark on the substitution rule

The substitution of parental alleles in the calculation of the kinship coefficient between two persons should always operate on the higher numbered person.

A counterexample:

\[ \Phi_{35} = \frac{1}{2} \Phi_{33} + \frac{1}{2} \Phi_{34}, \]

\[ \Phi_{35} \neq \frac{1}{2} \Phi_{15} + \frac{1}{2} \Phi_{25}. \]

Note: If replace the allele of 3 with its parents’ alleles, then sampling from 3 and sampling from 5 are not independent. Sampling from 5 depends on what have been sampled for 3.
§4.2. Identity states and identity coefficients

- Detailed identity states
• Condensed identity states

\[ S_1 = S_1^*, \quad S_2 = S_6^*, \quad S_3 = S_2^* \cup S_3^* \]
\[ S_4 = S_7^*, \quad S_5 = S_4^* \cup S_5^* \]
\[ S_6 = S_8^*, \quad S_7 = S_9^* \cup S_{12}^* \]
\[ S_8 = S_{10}^* \cup S_{11}^* \cup S_{13}^* \cup S_{14}^* \]
\[ S_9 = S_9^* \]