# Statistical methods in genetic relatedness and pedigree analysis

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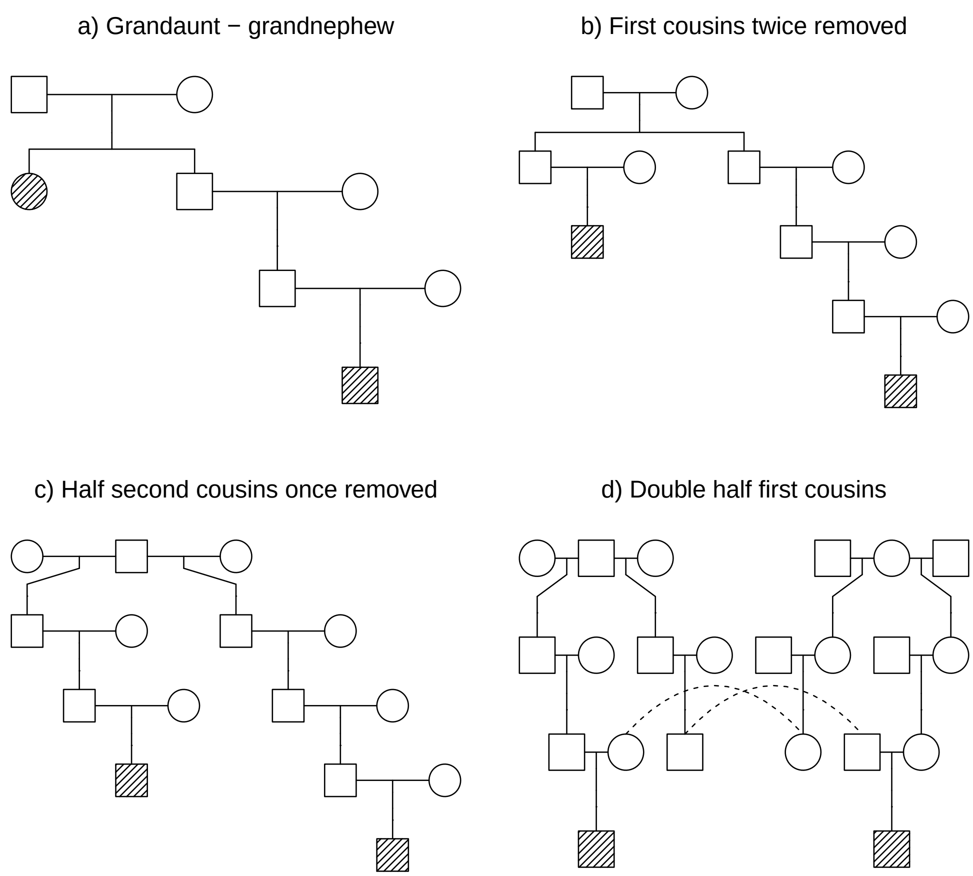
## Solutions for exercise set I

### Exercise I-1

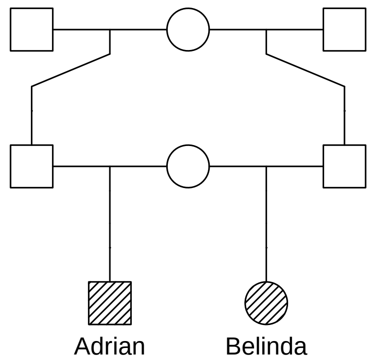
R versions of the pedigrees are shown below.




### Exercise I-2

a) Adrian and Belinda are half first cousins.

b) With the new information, Adrian and Belinda are simultaneous (maternal) half siblings and first half cousins.




**Exercise I-3**

a) Answer omitted.

b)
- 1 is a double grandfather of 7
- 3 is simultaneously the father and an uncle of 7
- 3 is a double uncle of 8
- 7 and 8 are quadruple first cousins

**Exercise I-4**

a) The marker is an autosomal STR. Reasons: heterozygous males; more than two alleles; numeric allele labels typical for STRs.
b) Five alleles (13, 14, 15, 21 and 22) are observed. The alleles names indicate the number of repeats.
c) Both 4 and 5 has genotype 13/14 (because of their parents).
d) Individual 3 has genotype 21/22 (because his children must have gotten 13 from their mother).
e) The possible genotypes are 13/13, 13/14, 13/15 and 14/15, each with probability $\frac{1}{4}$.

**Exercise I-5**

a) B (forced inheritance from the mother).
b) A/B (deduced from her children 5 and 6).
c) A/B. (She got B from her father, but has given an A to her son individual 10). She inherited A from her mother.
d) Neither genotype is possible to determine from the data.
e) 10 and 11 are maternal half siblings. 5 is the (maternal) uncle of 11. 4 and 9 are unrelated.

**Exercise I-6**

a) $p_A^3 p_B = 1/16$.
b) $\frac{1}{2} p_A^3 p_B = 0.03645$.

**Exercise I-7**

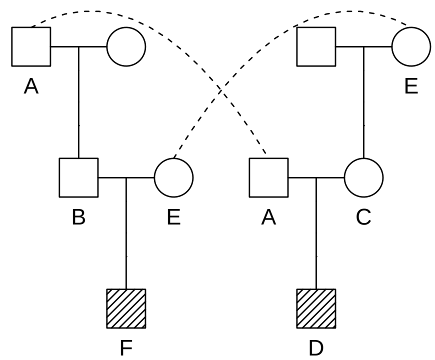
Let $p = P(A)$ and $q = P(B)$.

a) For a SNP in HWE the proportion of BB has expectation $q^2$. Setting $q^2 = 1/100$ gives $q = 1/10$, and thus $p = 9/10$. Our best guess is therefore $100 \cdot p^2 = 81$ individuals with $AA$, and $100 \cdot 2pq = 18$ individuals with $AB$.
b) Since $p^2 = 4q^2$, we find $p = 2q$. Inserting this into the identity $p + q = 1$, we get $2q + q = 1$, thus $p = 2/3$ and $q = 1/3$. The genotype frequencies for AA, AB and BB are therefore 4/9, 4/9 and 1/9.
c) It is not possible. If $P(AA) = P(BB)$ then $p^2 = q^2$, giving $p = q = 1/2$. But then $P(AA) = P(BB) = 1/4$ while $P(AB) = 2pq = 1/2$, contradicting the assumption.

# Bonus exercises

### Exercise I-8

a) Here is a plot of the pedigree:




b) Each is a half uncle of the other. This is the simplest example of a reciprocal asymmetric relationship.

### Exercise I-9

a) $P(A/A) = p$, $P(A/B) = q$, $P(B/B) = 0$.
b) $P(A/A) = p^2$, $P(A/B) = 2pq$, $P(B/B) = q^2$.
c) The conditional probabilities equal the Hardy-Weinberg proportions. This means that the genotypes of father and son does not tell us anything about the mother – her probabilities are exactly as for an unrelated person.

### Exercise I-10

a) Given that both parents are $A/B$, the genotype probabilities of each child are $P(A/B) = 0.5$ and $P(A/A) = P(B/B) = 0.25$. Thus the right-most pedigree is the most likely, since it has more heterozygous children than the left-most.
b) Once the parents are given, the children's genotypes depend only on them, and have nothing to do with HWE.