

# Statistical methods in genetic relatedness and pedigree analysis

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## Solutions for exercise set III

*Note:* Some answers are given in the exercise and therefore omitted here.

### Exercise III-1

- a) There are two paths of length three, giving  $\varphi = 2\left(\frac{1}{2}\right)^{3+1} = \frac{1}{8}$ .
- b) There is a single path of length 4, which gives  $\varphi = \left(\frac{1}{2}\right)^{4+1} = \frac{1}{32}$ .

### Exercise III-2

- a)  $f = \frac{1}{8} = 0.125$ , as shown e.g. by

```
x = avuncularPed("uncle", "niece")
x = addSon(x, c(3, 6))
plot(x)
inbreeding(x)
```

```
##      1      2      3      4      5      6      7
## 0.000 0.000 0.000 0.000 0.000 0.000 0.125
```

- b)  $f = \frac{1}{32} = 0.03125$ , as shown e.g. by

```
x = cousinPed(1, half = TRUE, child = TRUE)
plot(x)
inbreeding(x, id = 10)
```

```
## [1] 0.03125
```

The answers are the same as in the previous exercise because the inbreeding coefficient of an individual always equals the kinship coefficient of the parents.

### Exercise III-3

The inbreeding coefficient is  $f = \frac{1}{8} = 0.125$ , which can be computed in R as follows:

```
x = linearPed(2, sex = 2)
x = addSon(x, parents = c(1, 5))
plot(x)
inbreeding(x, id = 6)
```

```
## [1] 0.125
```

### Exercise III-4

- a) Maternal half siblings, and also first cousins through the fathers. The children are not inbred.
- b) One path of length 2, and two of length 4, gives  $\varphi = \left(\frac{1}{2}\right)^{2+1} + 2\left(\frac{1}{2}\right)^{4+1} = \frac{3}{16}$ .

c)  $(\frac{3}{8}, \frac{1}{2}, \frac{1}{8})$ . Starting with  $\kappa_2$ , we observe that

$$\kappa_2 = P(\text{same from mother})P(\text{same from fathers}) = \frac{1}{2} \cdot \varphi_S = \frac{1}{8},$$

where  $\varphi_S = \frac{1}{4}$  is the kinship coefficient between the fathers, being full siblings. Similarly, we find  $\kappa_0$  by

$$\kappa_0 = P(\text{different from mother})P(\text{different from fathers}) = \frac{1}{2} \cdot (1 - \varphi_S) = \frac{3}{8}.$$

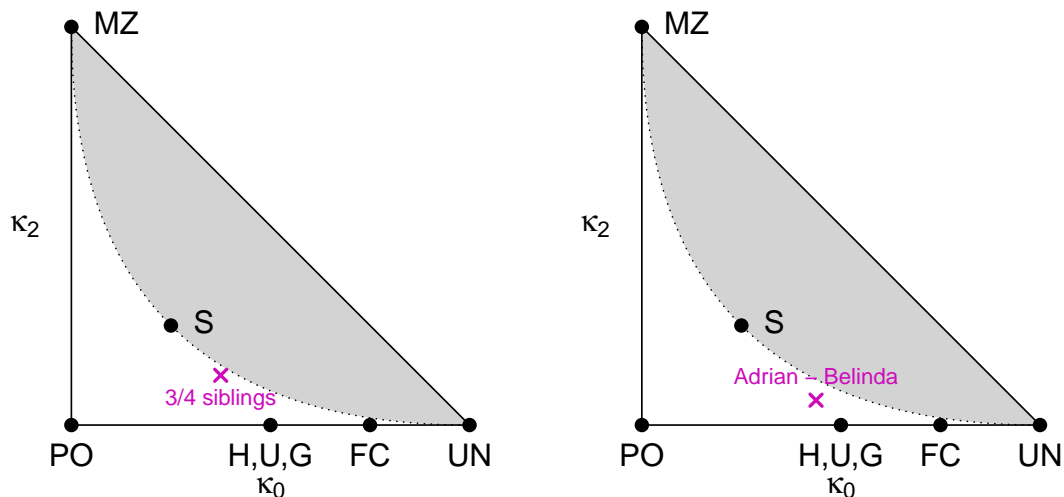
Finally we get  $\kappa_1 = 1 - \kappa_0 - \kappa_2 = \frac{1}{2}$ .

d) We find that  $\frac{1}{4}\kappa_1 + \frac{1}{2}\kappa_2 = \frac{1}{4} \cdot \frac{1}{2} + \frac{1}{2} \cdot \frac{1}{8} = \frac{3}{16}$ , which agrees with our value for  $\varphi$  above.

e) The point is shown in the left-most triangle below. R code:

```
showInTriangle(kappa = c(3/8, 1/2, 1/8), label = "3/4 siblings")
```

f) The point is exactly halfway between H (half siblings) and S (full siblings). (And  $\frac{3}{4}$  is halfway between  $\frac{1}{2}$  and 1.)



### Exercise III-5

a) The pedigree and plot is produced with the following code:

```
x = halfSibStack(2) |> swapSex(8)
plot(x, hatched = leaves, labs = c(Adrian = 7, Belinda = 8))
```

b)  $\frac{5}{32} = 0.15625$ , as obtained by

```
kinship(x, c(7,8))
```

```
## [1] 0.15625
```

Manual calculation: One path of length 2, and one of path 4, gives  $\varphi = (\frac{1}{2})^{2+1} + (\frac{1}{2})^{4+1} = \frac{5}{32}$ .

c)  $(\frac{7}{16}, \frac{1}{2}, \frac{1}{16}) = (0.4375, 0.5, 0.0625)$ , as found by

```
k = kappaIBD(x, c(7,8))
k
```

```
## [1] 0.4375 0.5000 0.0625
```

- d) The point is shown in the right-most triangle above. R code:

```
showInTriangle(k, label = "Adrian - Belinda", pos = 3)
```

- e) The identified point can be written as  $\frac{3}{4}H + \frac{1}{4}S$ , where  $H$  and  $S$  corresponds to half and full siblings, respectively. Thus, arithmetically speaking, their “fraction of siblingship” is  $\frac{3}{4} \cdot \frac{1}{2} + \frac{1}{4} \cdot 1 = \frac{5}{8}$

### Exercise III-6

- a) Outbred monozygotic twins have kinship coefficient  $\varphi = 0.5$ .  
 b) A kinship coefficient of  $\varphi = 1$  is possible only asymptotically, for example with repeated sibling matings. The *realised* kinship coefficient may be 1 in a finite pedigree, however. More about this later in the course!

### Exercise III-7

- a) First observe that  $\kappa_2 = 0$  since the fathers are unrelated. To find  $\kappa_0$ , we note that the mother cannot be autozygous if the half sibs have IBD status 0. Hence

$$\kappa_0 = P(\text{mother not autozygous}) \cdot P(\text{IBD} = 0 \mid \text{mother not autozygous}) = (1 - f) \cdot \frac{1}{2}.$$

Finally we compute  $\kappa_1 = 1 - \kappa_0 = \frac{1}{2}(1 + f)$ .

- b) On the  $\kappa_0$ -axis, more specifically on the line segment between H,U,G (when  $f = 0$ ) and P0 (when  $f = 1$ ).  
 c) For  $f = 1$ , the relationship is genetically equivalent to a parent-child relationship.

### Exercise III-8

Discussed in class.

### Exercise III-9

- a) Possible R code:

```
x = fullSibMating(1)
plot(x)
identityCoefs(x, ids = 5:6)
```

```
## [1] 0.06250 0.03125 0.12500 0.03125 0.12500 0.03125 0.21875 0.31250 0.06250
```

- b)  $\Delta_9 = 0.0625$  is much smaller than  $\kappa_0 = 0.25$  for outbred siblings. This quantifies the intuitive fact that the inbreeding increases the overall probability of IBD sharing.

### Exercise III-10

- a) Possible R code:

```
x = cousinPed(1, child = T)
x = addSon(x, parents = 7:8) # add second child
plot(x)
identityCoefs(x, ids = c(9, 10))
```

```
## [1] 0.015625 0.000000 0.031250 0.015625 0.031250 0.015625 0.234375 0.468750
## [9] 0.187500
```

- b) State two is impossible. It would require  $\text{IBD} = 2$  between the parents, but first cousins have  $\kappa_2 = 0$ .

## Bonus exercises

### Exercise III-11

- b) Calculation in R (see triangle plot further down):

```
dscA = doubleCousins(2,2)
kA = kappaIBD(dscA, 17:18)
kA
```

```
## [1] 0.87890625 0.11718750 0.00390625
```

- c)  $kA[2]^2 == 4 * kA[1] * kA[3]$

```
## [1] TRUE
```

- d) The fathers are double first cousins, while the mothers are unrelated. In R the pedigree can be made as follows:

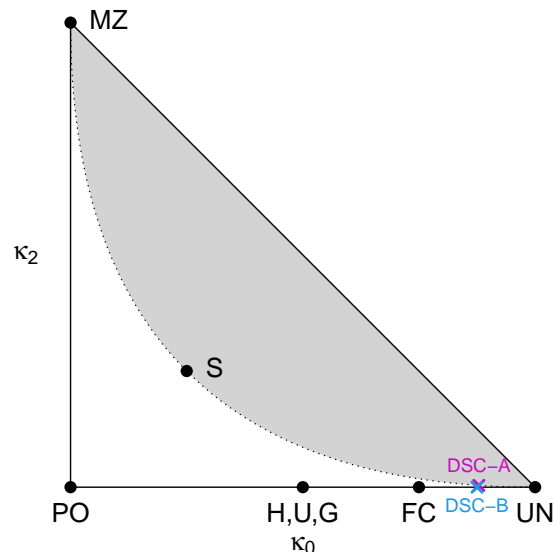
```
dscB = doubleCousins(1,1) |>
  addSon(9) |>
  addSon(10) |>
  relabel("asPlot")
```

- e)  $kB = \text{kappaIBD}(\text{dscB}, 13:14)$   
kB

```
## [1] 0.875 0.125 0.000
```

The following code plots both points in the same triangle:

```
showInTriangle(kA, label = "DSC-A", pos = 3)
showInTriangle(kB, label = "DSC-B", pos = 1, new = F, col = 4)
```

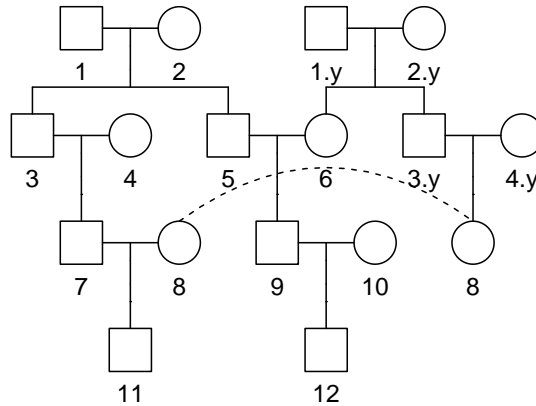


- f) Here is one solution: The fathers (7 and 9) are first cousins, while the mother (8) of one is the niece of the other's grandmother (6).

```
fa = cousinPed(2)
mo = avuncularPed("aunt", "niece", side = "left")
```

```
plotPedList(list(fa, mo))

dscC = mergePed(fa, mo, by = c("8" = "6", "6" = "5"))
plot(dscC)
```



Check that they are in fact DSC:

```
verbalise(dscC)
```

```
## Double second cousins
## 12-9-5-[1,2]-3-7-11
## 12-9-6-[1.y,2.y]-3.y-8-11
```

This type of DCS has the same IBD coefficients as type B:

```
kappaIBD(dscC, 11:12)
```

```
## [1] 0.875 0.125 0.000
```

### Exercise III-12

- $A/B - A/B$ : States 7, 8 and 9.
- $A/A - A/B$ : States 3, 4, 8 and 9.
- $13/17 - 15/15$ : States 6 and 9.
- $C/D - A/B$ : State 9.

### Exercise III-13

- $\Delta_8 = 1$ , all others are 0.
- The only possible states are  $S_5$ ,  $S_7$  and  $S_8$ . States 1–4 are impossible since the mother is not inbred;  $S_6$  and  $S_9$  are impossible because they lack vertical lines (which all parent-child relationships must have). Note that  $S_7$  is only possible if the relationship between the parents have  $\kappa_2 > 0$ .
- The identity coefficients are  $\Delta = (0, 0, 0, 0, \frac{1}{4}, 0, \frac{1}{4}, \frac{1}{2}, 0)$ . R code:

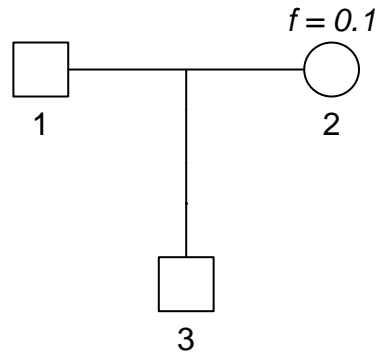
```
x = nuclearPed(1, sex = 2)
x = addSon(x, parents = c(1,3))
plot(x)
identityCoefs(x, ids = 3:4)
```

```
## [1] 0.00 0.00 0.00 0.00 0.25 0.00 0.25 0.50 0.00
```

- d) The child if not inbred. The nonzero identity coefficients between the mother and child are  $\Delta_3 = f$  and  $\Delta_8 = 1 - f$ .

Here's an example with  $f = 0.1$ :

```
x = nuclearPed(1)
founderInbreeding(x, 2) = 0.1
plot(x, margin = c(.5, .5, 1.5, .5))
```



The coefficients are as follows.

```
inbreeding(x) # just to check
```

```
## 1 2 3
## 0.0 0.1 0.0
```

```
round(identityCoefs(x, ids = 2:3), 10) # round to avoid rounding errors
```

```
## [1] 0.0 0.0 0.1 0.0 0.0 0.0 0.0 0.0 0.9 0.0
```