Kinship statistics and pedigree analysis

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Exercise set I. Pedigrees and measures of relatedness

Software used in these exercises

- **QuickPed**: Online app available at https://magnusdv.shinyapps.io/quickped.
- **ibdsim2**: Online app available at https://magnusdv.shinyapps.io/ibdsim2-shiny
- **R/pedsuite**: To get started, open RStudio and load the **pedsuite** packages:

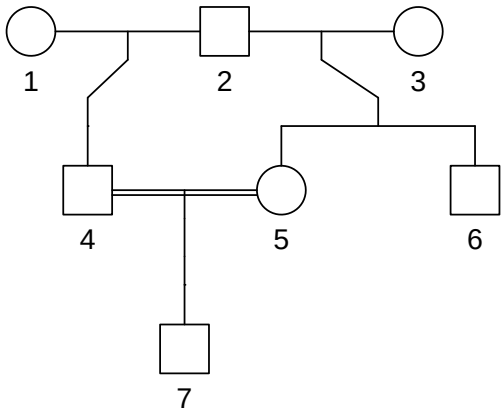
```
library(pedsuite)  # if error, try `install.packages("pedsuite")` first
```

Exercise I-1 (Basic pedigrees)

Draw pedigrees by hand illustrating the following relationships.

a) Grandaunt – grandnephew.
b) First cousins twice removed.
c) Half second cousins once removed.

Exercise I-2 (QuickPed)



a) Create the above pedigree in QuickPed. (Hint: Start with `Half siblings (pat)`.)
b) Describe the relationship between 6 and 7. (Hint: There is a button for that!)
c) What is the kinship coefficient between 6 and 7?
d) What is the inbreeding coefficient of individual 7?
e) Click on the "R code" button and verify that the code produces the same pedigree in R.

Exercise I-3 (Realised inbreeding)

In a case of incest a man had a son by his own granddaughter. The purpose of this exercise is to explore the distribution of the *realised* inbreeding in the offspring. Along the way we will practise our pedigree skills in R.

a) Create and plot the pedigree in R with the following code.

```
x = linearPed(2, sex = 2) |> addSon(parents = c(1, 5))
plot(x)
```

b) What is the inbreeding coefficient of the child?

c) Run the code below to simulate 500 realisations of the recombination in the pedigree. (Note the use of `seed` for reproducibility.)

```
library(ibdsim2)
sims = ibdsim(x, N = 500, seed = 111)
```

d) Plot the autozygous segments of the child in the first simulation.

```
sim1 = sims[[1]]
segs = findPattern(sim1, pattern = list(autozygous = "6"))
karyoHaploid(segs, title = "Autozygous segments")
```

e) For a more detailed picture, plot the full IBD pattern of the first chromosome:

```
haploDraw(x, sim1, chrom = 1)

# Alternative versions using optional parameters
haploDraw(x, sim1, chrom = 1, pos = c(2,4,2,4,4,4))
haploDraw(x, sim1, chrom = 1, pos = c(2,0,0,4,4,4),
          col = c("#FFC1C1", "#B20000", rep("gray90", 4)))
```

f) Use the code below to create a histogram of the realised inbreeding in the 500 simulations. Comment on the result.

```
r = realisedInbreeding(sims, id = 6)
fReal = r$perSimulation$fReal
hist(fReal, main = "Realised inbreeding")
abline(v = 0.125, col = 2, lwd = 2)
```

g) Find the standard deviation of the realised inbreeding coefficients.

h) How many autozygous segments will the child typically have? (Hint: `r$perSimulation$nSeg`.)

Exercise I-4 (Realised inbreeding with the ibdsim2 app)

We will now use the **ibdsim2** Shiny app to analyse the case from the previous exercise. To access the app, you can either use the online version (see URL above), or open it locally from R:

```
ibdsim2::launchApp()
```

The latter option may ask you to install some additional packages the first time. But it has the benefit that it works offline, and never disconnects. (The online version will disconnect after a period of inactivity.)

a) From the list of built-in pedigrees, select `Grandfather incest`. (Leave pedigree 2 empty.)
b) In the Settings frame, select `Autozygosity` analysis, and click `Simulate!`. Study the plots.
c) How many autozygous segments does the child typically have?
d) Select `Grandmother incest` as the second pedigree and simulate this as well. Comment on the results.

Exercise I-5 (A double relationship)

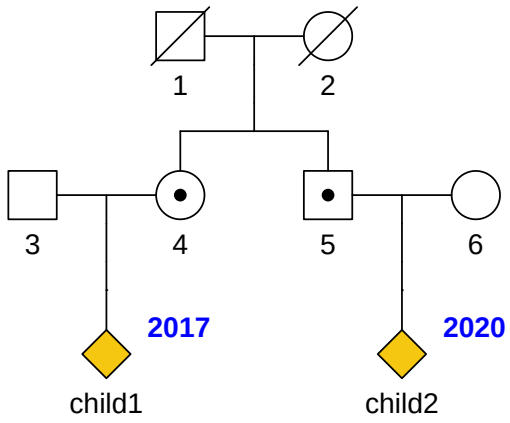
Adrian and Belinda have the same mother, and their fathers are maternal half siblings.

a) Draw the pedigree by hand. What is the relationship between Adrian and Belinda?
b) Draw the pedigree in QuickPed and use the "Describe relationship" button to verify your answer.
c) Compute their IBD coefficients $(\kappa_0, \kappa_1, \kappa_2)$ and plot the corresponding point in the IBD triangle.

Bonus exercises (if you have time)

Exercise I-6

Recreate the following pedigree plot in QuickPed as accurately as possible:



Exercise I-7

What is the relationship between individuals 7 and 8 in this pedigree?