ISFG 2024 Pre-congress Workshop

Kinship Statistics and Pedigree Analysis: Basic

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Exercise set I. Pedigrees and measures of relatedness

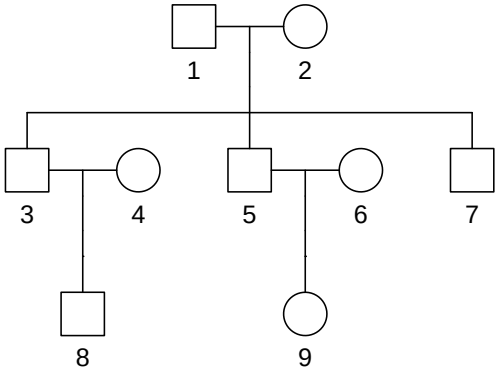
Welcome to the exercises! To get started, open RStudio and load the **pedsuite** packages:

```
library(pedsuite)
```

If this gives you an error, you may need to run `install.packages("pedsuite")` first.

Exercise I-1 (Building pedigrees in R)

To illustrate a typical workflow for building pedigrees in R, we will create this family:



a) Use the following code to create the pedigree. *Plot the pedigree after each line to track the progress.*

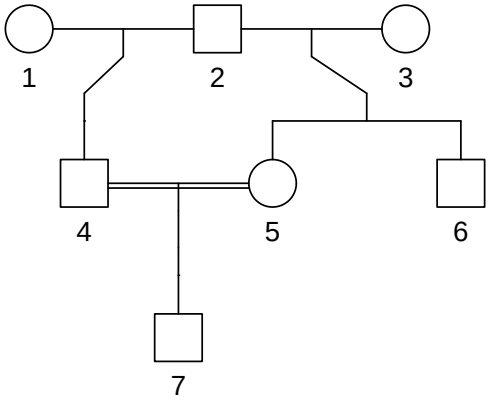
```
x = nuclearPed(nch = 3)
x = addSon(x, 3)
x = addDaughter(x, 4)
x = relabel(x)  # relabel according to plotting order
```

b) The **pedsuite** works well with *pipe* operator `|>` in R. The pipe makes the result of the previous command become the first argument of the next. For example, the code in a) may be rewritten to this:

```
x = nuclearPed(nch = 3) |>
  addSon(3) |>
  addDaughter(4) |>
  relabel()
```

Verify this by running the code and plot the result.

Exercise I-2 (QuickPed)



a) Create the above pedigree in QuickPed (https://magnusdv.shinyapps.io./quickped/).
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.

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.

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 - cont.)

We will now use the **ibdsim2** Shiny app to analyse the incest case from the previous exercise. To open the app, you have two choices:

- Go to the website https://magnusdv.shinyapps.io./ibdsim2-shiny/
- Or open the app from R with the command `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 does this after a few minutes 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 (Bonus exercise: A double relationship)

This exercise can be solved either with QuickPed or in R (or by hand if you really want to show off!)

a) Adrian and Belinda are maternal half siblings, and their fathers are also maternal half siblings. Draw the pedigree.
b) What is the kinship coefficient of Adrian and Belinda?
c) Compute the IBD coefficients $(\kappa_0, \kappa_1, \kappa_2)$ and plot the corresponding point in the IBD triangle.