

Pedigree analysis: Basic

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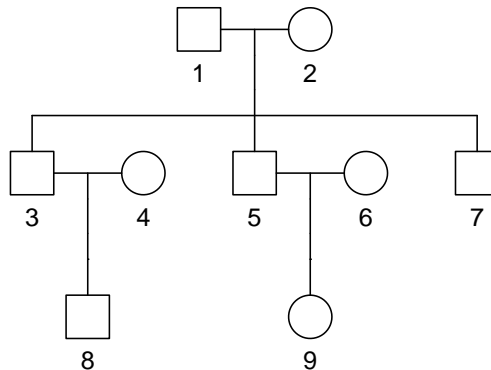
Exercise set I. Pedigrees and coefficients of relatedness

To get started, load the **pedsuite** packages in R:

```
library(pedsuite)
```

Exercise I-1 (Building pedigrees)

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.

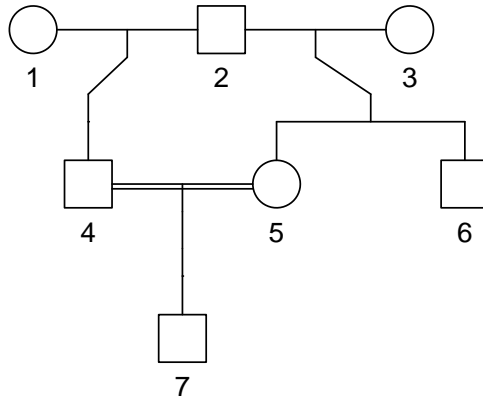
```
x = nuclearPed(nch = 3)
x = addSon(x, 3)
x = addDaughter(x, 4)
x = relabel(x) # relabel according to plotting order
```

- b) The **pedsuite** works well with *pipe* operator `|>` recently introduced 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)



- Use QuickPed (<https://magnusdv.shinyapps.io/quickped/>) to create the pedigree above.
- Describe the relationship between 6 and 7, and compute their kinship coefficient.
- What is the inbreeding coefficient of individual 7?
- Save the pedigree as a ped file named *quick1.ped* somewhere on your computer. Then load this file in R and check that it is the same pedigree:

```
x = readPed("../your path../quick1.ped")
plot(x)
```

Exercise I-3 (A double relationship)

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

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

Exercise I-4 (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.

- Create and plot the pedigree in R with the following code.

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

- What is the inbreeding coefficient of the child?

- a) 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)
```

- b) 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")
```

- c) 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), cols = c(8,2,8,8,8,8))
```

- d) 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)
```

- e) Find the standard deviation of the realised inbreeding coefficients.
f) How many autozygous segments will the child typically have?

Exercise I-5 (Bonus challenge for experts)

Use R to create a pedigree with *quadruple second cousins*.

Hint: Make the fathers double first cousins, and the same for the mothers. Use `doubleCousins()` twice and glue the parts with `mergePed()`. The final plot may look confusing, but you should verify the relationship with `verbalise()`. Check `?mergePed` for a similar example.