

# ISFG summer school - virtual edition 2021

## Pedigree analysis in R

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### Exercise set II. Working with pedigrees and markers in R

#### Before you start

The aim of these exercises is to acquaint you with the **pedsuite**, which is a collection of R packages for pedigree analysis. For now we focus on two of the packages, **pedtools** (creating and plotting pedigrees) and **pedprobr** (probability computations). For more information, see the [pedsuite homepage](#).

To get started, open Rstudio and load the **pedsuite**.

```
library(pedsuite)
```

If this gives an error like **there is no package called pedsuite**, you must first install it:

```
install.packages("pedsuite") # don't forget the quotes!
```

The **pedtools** package contains much more than we have time to cover in this course. A more comprehensive overview is available in the [user manual](#).

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#### Exercise II-1 (Creating pedigrees)

This exercise displays some common methods of creating pedigrees with **pedtools**. I recommend running `plot(x)` after each line of code, to get a feeling of what's going on.

- a) Use the code below to create the pedigree in Figure 1. Plot the pedigree after each line.

```
x = nuclearPed(nch = 2)
x = addSon(x, 3)
x = addDaughter(x, 4)
x = relabel(x, "asPlot") # relabel according to plotting order
```

- b) Use the code below to create the pedigree in Figure 2. Plot the pedigree after each line.

```
x = nuclearPed(nch = 1)
x = addParents(x, id = 1)
x = addParents(x, id = 2)
x = relabel(x, "asPlot")
```

#### Exercise II-2 (Pedigree shortcuts)

Several shortcuts for common pedigree structures are available in **pedtools**. In many cases one of these will give you the pedigree you want in a single command.

- a) Check that the following code produces the pedigree in Figure 1.

```
x = cousinPed(degree = 1)
x = swapSex(x, ids = 8)
```

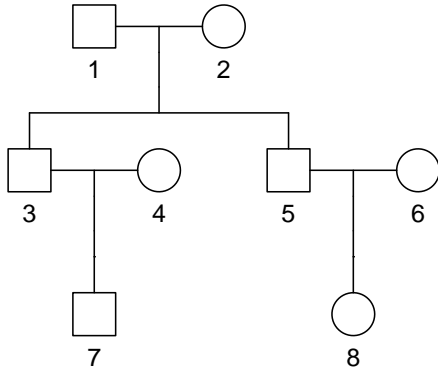


Figure 1

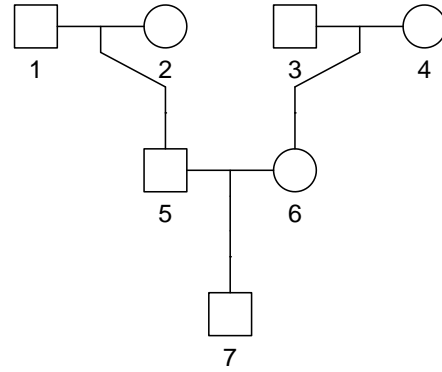


Figure 2

- b) Check that the following code produces the pedigree in Figure 2.

```
x = ancestralPed(g = 2)
```

### Exercise II-3 (Marker genotypes and plotting options)

- a) Use the code below to create the pedigree in Figure 3.

```
x = nuclearPed(3)
m = marker(x, geno = c("A/A", NA, "A/B", "A/B", "A/B"))
plot(x, marker = m)
```

- b) With  $x$  and  $m$  as above, try the following commands. Experiment with different plot options on your own. The help page for pedigree plotting contains additional information: type `?plot.ped` to open it.

```
plot(x, title = "MY PED", hatched = 3:5, deceased = 1:2)
plot(x, col = list(red = 1:2, blue = 3:5), labs = NULL)
plot(x, marker = m, showEmpty = T, missing = "*", sep = ":")
plot(x, cex = 2)
```

- c) Try to recreate the version shown in Figure 4 (use  $x$  and  $m$  as above).

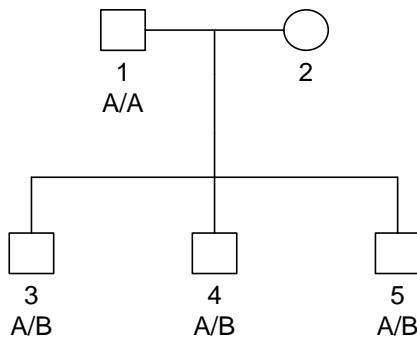


Figure 3

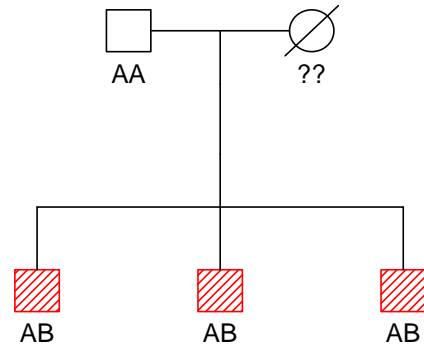


Figure 4

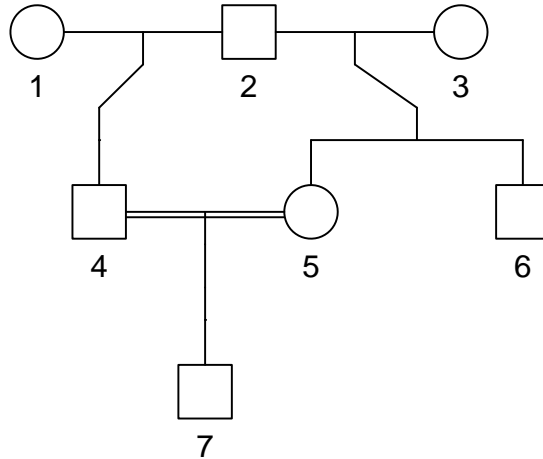


Figure 5

#### Exercise II-4 (Creating and loading a pedigree file)

- Use QuickPed (<https://magnusdv.shinyapps.io/quickped/>) to create the pedigree in Figure 5, and store it as a ped file named “fig5.ped” somewhere on your computer.
- Load the pedigree into R using the `readPed()` function:

```
x = readPed("path/to/your/folder/fig5.ped")
```

Plot the pedigree and check that it looks like Figure 5.

#### Exercise II-5 (Inbred pedigrees)

- Use R to create and plot a pedigree showing father-daughter incest.
- Use R to create and plot a pedigree showing brother-sister incest.

#### Exercise II-6 (Pedigree likelihoods)

Figures 6 and 7 show pedigrees with genotypes for an autosomal SNP with allele frequencies  $P(A) = 0.9$  and  $P(B) = 0.1$ . We assume Hardy-Weinberg equilibrium.

- Use the following code to compute the pedigree likelihood in Figure 6.

```

# Create pedigree
x = nuclearPed(2)
m = marker(x, geno = c("A/A", "B/B", "A/B", "A/B"), afreq = c(A = 0.9, B = 0.1))
plot(x, marker = m)

# Compute likelihood
likelihood(x, m)
  
```

- Compute the likelihood of the pedigree in Figure 7.
- (If you have time) Validate the results in a) and b) by calculating the likelihoods manually.

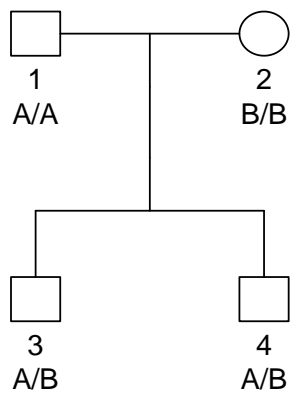


Figure 6

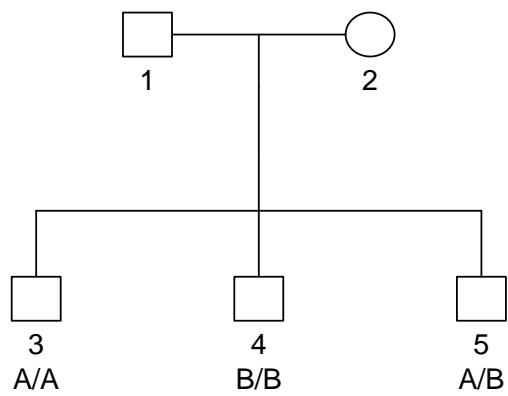


Figure 7