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Workshop session I. Basic pedigree analysis in R

Before you start

The aim of this session is to acquaint you with the **pedsuite** and show how to create and analyse pedigrees in R. For more information, see the pedsuite homepage.

To get started, open Rstudio and load the **pedsuite**, and also the **ibdsim2** package.

```
library(pedsuite)
library(ibdsim2)
```

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!
```

Note: If you find the exercises difficult and need a break from R, then jump ahead to Exercises I-5 and I-6. They can be solved in QuickPed without any coding!

Exercise I-1   (Building 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) Check that the following code produces the same pedigree.
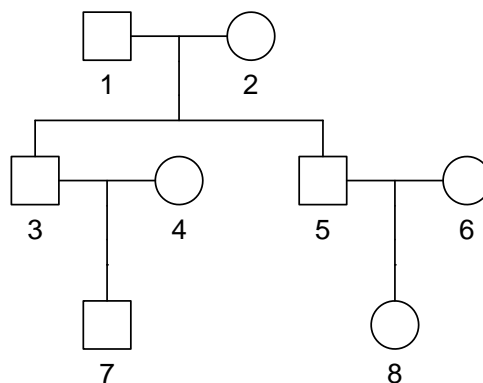
```
x = cousinPed(degree = 1)
x = swapSex(x, ids = 8)
```
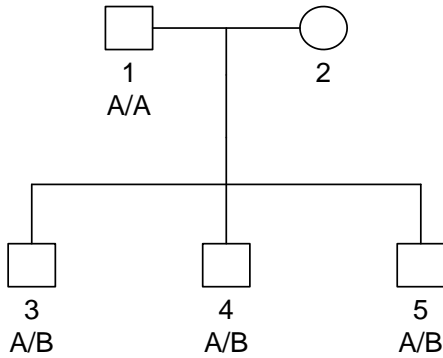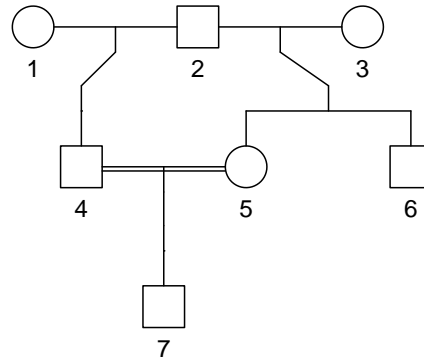

Figure 1

Figure 2

Figure 3

Exercise I-2   (Marker genotypes. Plot options)

a) Use the code below to create the pedigree in Figure 2.

```
x = nuclearPed(3)
x = addMarker(x, geno = c("A/A", NA, "A/B", "A/B", "A/B"))
plot(x, marker = 1)
```

b) With `x` as above, try the following commands and make sure you understand how the different options work. The help page for pedigree plotting contains additional information: type `?plot.ped` to open it.

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

Exercise I-3   (Pedigree likelihood)

Suppose the marker in Figure 2 is an autosomal SNP in Hardy-Weinberg equilibrium, with allele frequencies $P(A) = 0.9$ and $P(B) = 0.1$.

a) With `x` as in the previous exercise, use `afreq(x, marker = 1)` to inspect the allele frequencies.

b) Set the correct allele frequencies and compute the likelihood as follows.

```
x = setAfreq(x, marker = 1, afreq = c(A = 0.9, B = 0.1))
likelihood(x, marker = 1)
```

c) (Optional) Validate the likelihood by manual calculation.

Exercise I-4   (Creating and loading a ped file)

a) Create the pedigree shown in Figure 3 using QuickPed (https://magnusdv.shinyapps.io/quickped/).

b) What is the relationship between individuals 6 and 7? Use QuickPed to check your answer.

c) Store the pedigree as a ped file named "fig3.ped" somewhere on your computer.

d) Load the pedigree into R using the `readPed()` function, and check that it looks like Figure 3:

```
x = readPed("path/to/your/folder/fig3.ped")
plot(x)
```

Relatedness coefficients

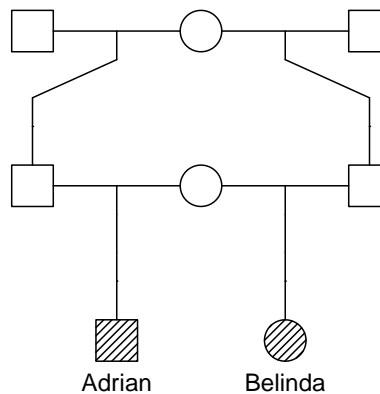The next two exercises can be solved either in R or with QuickPed.

Exercise I-5 (Inbred pedigree)

In a case of incest, a man had a child by his own granddaughter.

a) Create and plot the pedigree.
b) What is the inbreeding coefficient of the child?

Exercise I-6 (A double relationship)

Consider the relationship between Adrian and Belinda:

a) Describe the relationship. Are Adrian and Belinda inbred?
b) Create and plot the pedigree. *Hint if you use R*: `x = halfSibStack(2)`.
c) Compute the kinship coefficient between Adrian and Belinda.
d) Compute the IBD coefficients $(\kappa_0, \kappa_1, \kappa_2)$ of the relationship.
e) Plot the corresponding point in the IBD triangle.

Realised relatedness

Exercise I-7 (Realised inbreeding)

a) Use the following code to estimate the distribution of the realised inbreeding coefficient in a child of uncle–niece parents.

```
x = cousinPed(deg = 0, removal = 1, child = TRUE)
plot(x)
```

```
sims = ibdsim(x, N = 500, ids = 7)
real = realisedInbreeding(sims)
hist(real$perSimulation$fReal)
```

Comment on the result.

b) Make a similar histogram for the number of autozygous segments.
(Hint: Use one of the other columns of `real$perSimulation`.)

Exercise I-8   (Zero IBD)

Suppose that you are a 10th generation, male-line descendant of Napoleon Bonaparte.

a) What is the probability that none of your (autosomal) DNA originates from Napoleon? Use the following code to give a realistic estimate:

```
x = linearPed(10)
sims = ibdsim(x, N = 500, ids = c(1, 21))
zeroIBD(sims)
```

b) Repeat the analysis for a female line of 10 generations. Comment on the result.