

# Kinship and pedigree analysis: Methods and applications

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## Exercise set III. Kinship testing

Before you start, run the following commands to download some of the datasets used in this course.

```
url = "https://magnusdv.github.io/pedinr/datasets/data.zip"
download.file(url, destfile = "data.zip")
unzip("data.zip")
```

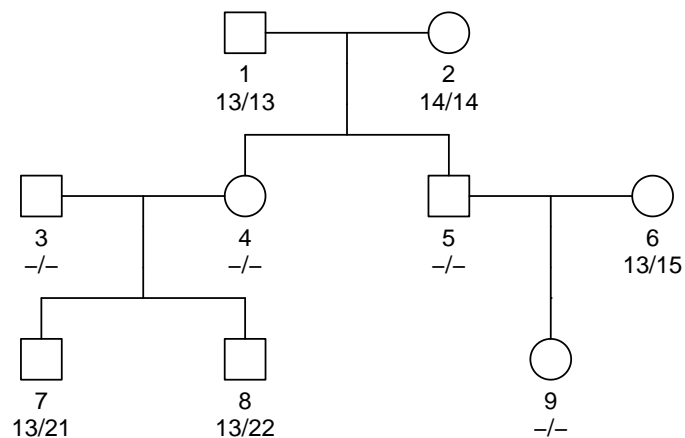
*Note:* The files are put in a subfolder `data` in the current working directory. You can identify this by with the command `getwd()`.

You should also load the `pedsuite` packages:

```
library(pedsuite)
```

### Exercise III-1

Consider the following pedigree, in which some members have been typed with a single DNA marker.



- What kind of marker is this: SNP or STR? Autosomal or X-linked? How do you know?
- How many different alleles are observed in the family. What do the allele labels (e.g. 13) mean?
- What are the genotypes of individuals 4 and 5?
- Can you determine the genotype of individual 3?
- What are the possible genotypes for individual 9, and how likely is each of them?

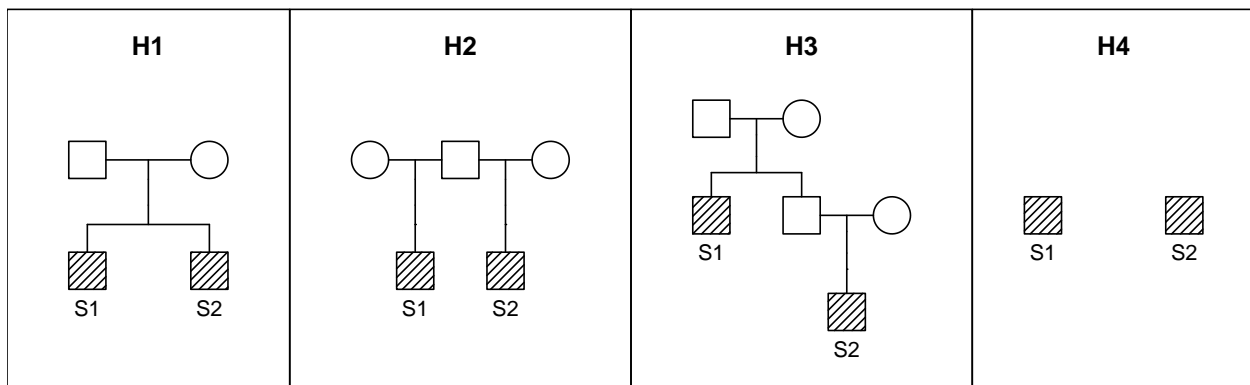
**Exercise III-2**

In a paternity case, the alleged father has genotype a/b for a certain marker, while the child, has genotype a/c. The mother is not genotyped.

- Formulate the natural hypotheses H1 and H2, and create the corresponding pedigree objects in R.
- Use `plotPedList()` to plot H1 and H2.
- Use `kinshipLR()` to compute the LR if the allele frequencies are  $p_a = 0.01$ ,  $p_b = 0.3$  and  $p_c = 0.69$ .
- (Optional) Find a general formula for LR expressed by the allele frequency  $p_a$ .

**Exercise III-3**

In this exercise we will analyse the relationship between two males, S1 and S2, who are genotyped with 15 STR markers. The hypothesised pedigrees are shown below.



- Define the first three pedigrees by running:

```
ids = c("S1", "S2")
H1 = nuclearPed(children = ids)
H2 = relabel(halfSibPed(), old = 4:5, new = ids)
H3 = relabel(avuncularPed(), old = c(3,6), new = ids)
```

- Load the marker data and check that the result contains S1 and S2 as singletons.

```
H4 = readPed("../data/kinship-riddle.ped")
H4 = setFreqDatabase(H4, database = NorwegianFrequencies)
summary(H4)
```

- Find the LRs using H4 as the reference by running

```
lr = kinshipLR(H1, H2, H3, H4, verbose = TRUE, ref = 4)
```

- Notice that H2 and H3 give identical LRs. Do you think this is a coincidence? Explain!
- Include a hypothesis specifying that S1 and S2 are first cousins, and find the LR against H4. *Hint:*

```
H5 = relabel(cousinPed(1), old = 7:8, new = ids)
```