Pedigree analysis and relatedness inference

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Exercise set II: Kinship testing

Some exercises below use **KLINK**, which is an extension of Familias handling pairwise linked markers. For more details, see the KLINK paper here: 10.1016/j.fsigen.2026.103578.

The online version of **KLINK** is available here: https://magnusdv.shinyapps.io/klink.

For optimal performance (and with sensitive case data) we recommend running **KLINK** locally from RStudio. To set this up, first install the R package:

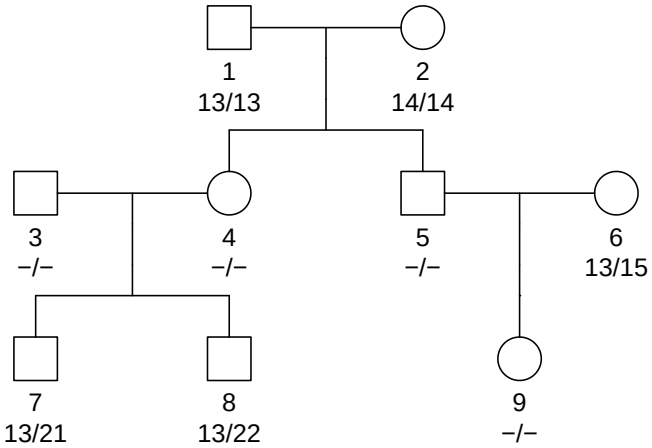
```
install.packages("KLINK")
```

After installation you can launch the app:

```
KLINK::launchApp()
```

Exercise II-1

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



a) What kind of marker is this: SNP or STR? Autosomal or X-linked? How do you know?

b) How many different alleles are observed in the family? What do the allele labels (e.g. `13`) mean?

c) What are the genotypes of individuals 4 and 5?

d) Can you determine the genotype of individual 3?

e) What are the possible genotypes for individual 9, and how likely is each of them?

Exercise II-2

Required file: `halfsib_case.fam`

a) Load the Familias file `halfsib_case.fam` into KLINK and study the data.
   i. Describe the kinship case. What are the hypotheses?
   ii. Where are the two markers located? Are they linked? (Hint: Look at *Linkage map*.)
   iii. What is the centiMorgan distance between the markers?

b) Calculate the LR in KLINK.
   i. What is the LR ignoring linkage?
   ii. What is the LR accounting for linkage? How much (in %) does it differ from the unlinked LR?

c) Set the mutation model to *Simple*.
   i. What is the mutation rate with this model? (Hint: Look at *Marker data*.)
   ii. How much does the LR change?

Exercise II-3

In this exercise we analyse one of the built-in examples in KLINK. To get started, open KLINK and click the green button labelled "Example 2".

a) Describe the kinship case. What are the hypotheses?

b) How many markers are used? How many linked pairs are there?

c) Some chromosomes have more than two markers. How does KLINK handle this?

d) Compute the LRs. What is the total LR with and without linkage?

e) Turn off mutation modelling and recalculate the LRs. What happens? Explain why.

f) Re-do the LRs using the original mutation models, and download the results. Examine the different sheets of the Excel document.