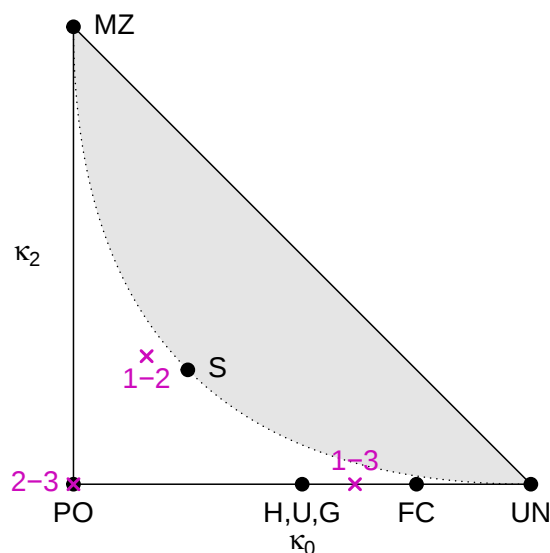


Pedigree analysis and relatedness inference

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Exercise set III: Relationship inference and pedigree reconstruction

Exercise III-1

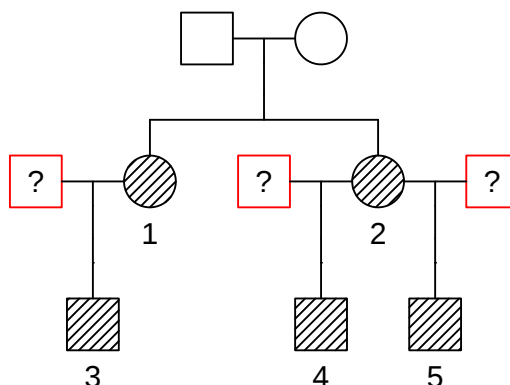


- The triangle above shows pairwise kappa estimates for three male individuals, labelled 1, 2 and 3. How do you interpret the relationship between each pair? How confident are you in each case?
- Use the pairwise estimates to deduce the pedigree connecting the individuals.

Exercise III-2

Required files: `reconstruct-fathers.ped`, `reconstruct-fathers.freq`

The following is based on an actual case from Australia. Genotypes are available from two sisters and their children. The first sister has one child, the other has two children. The question we must answer is: *Do any of the children have the same father?*



- Load the data with the commands below, and use `summary()` to inspect the data. Check that the labels and sexes match the figure. How many markers are used?

```
x = readPed("data/reconstruct-fathers.ped") |>
  setFreqDatabase("data/reconstruct-fathers.freq")
```

- b) Use **pedbuildr** to perform a pedigree reconstruction on the data set. *Hint*: Use undisputed parts of the family to restrict the search space. For example, consider the parameters **knownPO** and **noChildren**.
- c) Plot the six most likely pedigrees and study the paternity constellations. What is your conclusion?

Exercise III-3

Two bodies are found in an ancient grave. The evidence suggests that they are related, but how? It is found that the individuals share 3 IBD segments on the X chromosome, of approximate lengths 10 cM, 15 cM and 60 cM. Unfortunately, the sex of the individuals is not known with certainty.

The following relationships are particularly relevant:

- Paternal half siblings
- Maternal half siblings
- Paternal grandparent-grandchild
- Maternal grandparent-grandchild

- a) Which of the above can be ruled out without any statistical analysis? Explain.
- b) Use **ibdsim2** to simulate relevant IBD distributions on X, and give a conclusion.

Exercise III-4

In a case of suspected incest, a woman has given birth to a child whose father may be either the woman's father or her brother. After sequencing the child's genome, the investigators identify 25 autozygous segments with a total length of 850 cM. Use **ibdsim2** to determine which of the two hypotheses is most likely given these observations.