

Pedigree analysis and relatedness inference

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Exercise set I: Pedigrees and measures of relatedness

The following online apps are used in these exercises:

- **QuickPed**: <https://magnusdv.shinyapps.io/quickped>
- **ibdsim2**: <https://magnusdv.shinyapps.io/ibdsim2-shiny>

Note: **ibdsim2** can also be run locally from R. To set this up, install the package:

```
install.packages("ibdsim2", dep = T)
```

You can then start the app with a single command:

```
ibdsim2::launchApp()
```

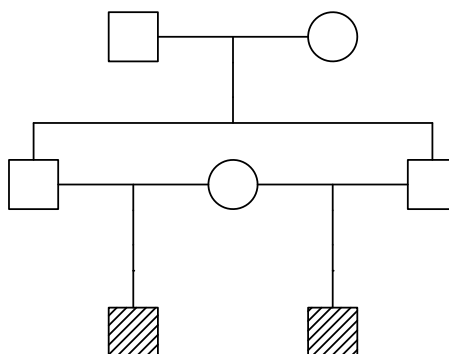
Exercise I-1

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

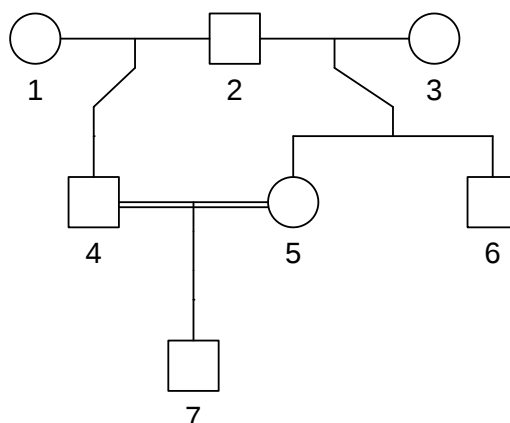
- Draw the pedigree in QuickPed (or in R).
- Compute the inbreeding coefficient of the child.
- What is the formula connecting inbreeding coefficients and kinship coefficients? Check that it holds in this case.

Exercise I-2

Consider the following pedigree:



- Describe the relationship between the children. Are they inbred?
- Use QuickPed to show that their IBD coefficients are $\kappa = (\frac{3}{8}, \frac{1}{2}, \frac{1}{8})$.
- Show the relationship in the IBD triangle.
- This relationship is sometimes called *3/4-siblings*. Why?

Exercise I-3

- Create the above pedigree in QuickPed. (Hint: Start with **Half siblings (pat)**.)
- Describe the relationship between 6 and 7.
- What is the kinship coefficient between 6 and 7?
- What is the inbreeding coefficient of individual 7?
- Click on the “R code” button and verify that the code produces the same pedigree in R.

Exercise I-4

- What is the definition of the kinship coefficient φ between two individuals?
- What is the kinship coefficient between monozygotic twins?
- Give examples of relationships where $\varphi \geq 0.5$.
- Discuss: Describe a relationship with $\varphi = 1$. How can this be achieved in practice? (Hint: It is best to imagine other species than humans for this one!)

Exercise I-5

In this exercise we use the **ibdsim2** app to study the distribution of IBD segments in grandparent–grandchild relationships.

- From the list of built-in pedigrees, select **Grandparent (male line)** and click **Simulate!** (Leave pedigree 2 empty and all settings at their default values.)
- How many IBD segments do they typically share? What is the typical average segment length?
- On average, approximately how much of the genome (in %) is covered by IBD segments? How does that compare with the theoretical coefficient κ_1 for this relationship?
- Select **Grandparent (female line)** as the second pedigree and simulate this as well. Comment on the results. Why are the distributions different?
- Change the second pedigree to **Grandparent (male line)** and select individuals 2 and 5 (i.e., using the grandmother instead of the grandfather). Before simulating, make a guess about how this will compare with the first pedigree. Do the simulations and comment on the result.

Bonus exercise if you have time

Draw your own family tree in QuickPed.