Kinship and pedigree analysis: Methods and applications

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Exercise set V. Realised relatedness

This exercise set is about realised relatedness, and how it may deviate from the pedigree-based expectation. We will use the pedsuite package **ibdsim2** to simulate recombination patterns and investigate distributions of realised inbreeding and relatedness.

```r
library(pedsuite)
library(ibdsim2)  # not a core package; must be loaded separately
```

Exercise V-1

In a case of incest a man had a son by his own granddaughter. The purpose of this exercise is to explore the distribution of the realised inbreeding in the offspring.

a) Create and plot the pedigree in R with the following code.

```r
x = linearPed(2, sex = 2) |> addSon(parents = c(1, 5))
plot(x)
```

b) What is the (pedigree-based) inbreeding coefficient of the child?

c) Run the code below to simulate 500 realisations of the recombination in the pedigree. (Note the use of `seed` for reproducibility.)

```r
sims = ibdsim(x, N = 500, seed = 111)
```

d) Plot the autozygous segments of the child in the first simulation.

```r
sim1 = sims[[1]]
segs = findPattern(sim1, pattern = list(autozygous = "6"))
karyoHaploid(segs, title = "Autozygous segments")
```

e) For a more detailed picture, plot the full IBD pattern of the first chromosome:

```r
haploDraw(x, sim1, chrom = 1)

# Alternative versions using optional parameters
haploDraw(x, sim1, chrom = 1, pos = c(2,4,2,4,4,4))
haploDraw(x, sim1, chrom = 1, pos = c(2,0,0,4,4,4),
          col = c("#FFC1C1", "#B20000", rep("gray95", 4)))
```

f) Use the code below to create a histogram of the realised inbreeding in the 500 simulations. Comment on the result.

```
r = realisedInbreeding(sims, id = 6)
fReal = r$perSimulation$fReal
hist(fReal, main = "Realised inbreeding")
abline(v = 0.125, col = 2, lwd=2)
```

g) Find the standard deviation of the realised inbreeding coefficients.

h) How many autozygous segments will the child typically have? (Hint: `r$perSimulation$nSeg`.)

Exercise V-2 (Realised inbreeding - cont.)

We will now use the **ibdsim2** Shiny app to analyse the incest case from the previous exercise. To open the app, you have two choices:

- Go to the website https://magnusdv.shinyapps.io./ibdsim2-shiny/

- **OR**: Open the app from R with the command `ibdsim2::launchApp()`.
  Note: You may be asked to install some additional packages the first time you do this.

a) From the list of built-in pedigrees, select `Grandfather incest`. (Leave pedigree 2 empty.)
b) In the Settings frame, select `Autozygosity` analysis, and click `Simulate!`. Study the plots.
c) How many autozygous segments does the child typically have?
d) Select `Grandmother incest` as the second pedigree and simulate this as well. Comment on the results.

Exercise V-3 (Zero IBD)

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

a) What is the probability that some 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))
z = zeroIBD(sims)
1 - z$zeroprob
```

b) Repeat the analysis for a female line of 10 generations. Why is the result different?

Exercise V-4 Bonus exercise: Fly forensics

A certain species of flies has two autosomal chromosomes of lengths approx. 107 and 110 cM. Simulate 500 pairs of fly siblings, and plot their realised κ coefficients in the IBD triangle. Comment on the result.

Hint: Reuse code from the lecture, but use the following map in the `ibdsim()` command:

```
mapFly = list(uniformMap(cM = 107, chrom = 1),
              uniformMap(cM = 110, chrom = 2))
```