Pedigrees and kinship analysis in forensic genetics

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Exercise set V. IBD segments and realised relatedness

The **ibdsim2** Shiny app is accessible in two ways:

- *Online*: The app is available online at https://magnusdv.shinyapps.io/ibdsim2-shiny/.
- *From R*: Run `ibdsim2::launchApp()` in a fresh R session. (You may be asked to install some additional packages the first time you do this.)

Exercise V-1 (Realised inbreeding with the ibdsim2 app)

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

We will first use the **ibdsim2** Shiny app to analyse the case.

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-2 (Realised inbreeding in R)

In this exercise we analyse the same relationship as in the previous exercise, but this time in R. This requires a bit of coding, but allows us to go much deeper. Along the way we will practise our pedigree skills in R.

Start by loading the necessary packages:

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

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 inbreeding coefficient of the child?
(Hint: Try `inbreeding(x)`)

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)
```

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

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

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

```
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("gray90", 4)))
```

c) 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)
```

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

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

Exercise V-3

Two bodies are found in an ancient grave. The evidence suggests that they are related, but how? Using sequencing, researchers have established that the individuals share 3 IBD segments on the X chromosome, of lengths 10 cM, 15 cM and 60 cM. Unfortunately, the sex of the individuals are 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 IBD distributions on X for the alternatives and conclude which is best fit.