Kinship and pedigree analysis: Methods and applications

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Exercise set VII. DNA based disaster victim identification

To get started, open RStudio and load the **pedsuite** packages used in these exercises:

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

If needed, download the datasets with the following command:

```
download.file("https://magnusdv.github.io/pedinr/datasets/data.zip", dest = "data.zip")
unzip("data.zip")
```

Exercise VII-1 (MPI case study)

In this exercise we will work through a case study in missing person identification. Genotypes and allele frequencies are given in the files `mpi-example.ped` and `mpi-example.freq`, included in the `data` folder.

a) Load the data in R with the code below, and inspect it using `summary()` and `plotPedList()`.

```
mpi = readPed("data/mpi-example.ped")
mpi = setFreqDatabase(mpi, "data/mpi-example.freq")
```

b) Use the following code to plot the hypotheses. How many reference individuals have been typed, and what are their relationships to the missing person?

```
ref = mpi$Reference
missingPersonPlot(ref, missing = "MP")
```

c) We want to test if POI1 is the missing person. Find the LR and give a conclusion after running

```
poi1 = mpi$POI1
mpiTest1 = missingPersonLR(ref, missing = "MP", poi = poi1)
mpiTest1
```

d) Use the code below to plot the LR for each marker. Which markers have LR = 0?

```
lr1 = mpiTest1$LRperMarker
cols = ifelse(lr1 > 1, 8, 2)
barplot(lr1, col = cols, ylab = "LR", las = 2, cex.names = 0.8)
abline(h = 1, lty = 2)
```

e) Find the LR for POI2. Which marker gives the largest LR?

f) Explain why marker D18S51 gives a high LR for POI2, but not for POI1.
Hint: Look at `plotPedList(mpi, marker = "D18S51")` and `afreq(mpi, marker = "D18S51")`.

g) Find the *exclusion power* by running the code below. Interpret the output.

```
ep = missingPersonEP(ref, missing = "MP")
ep
```

h) Find the *inclusion power* by running the code below. Interpret the output.

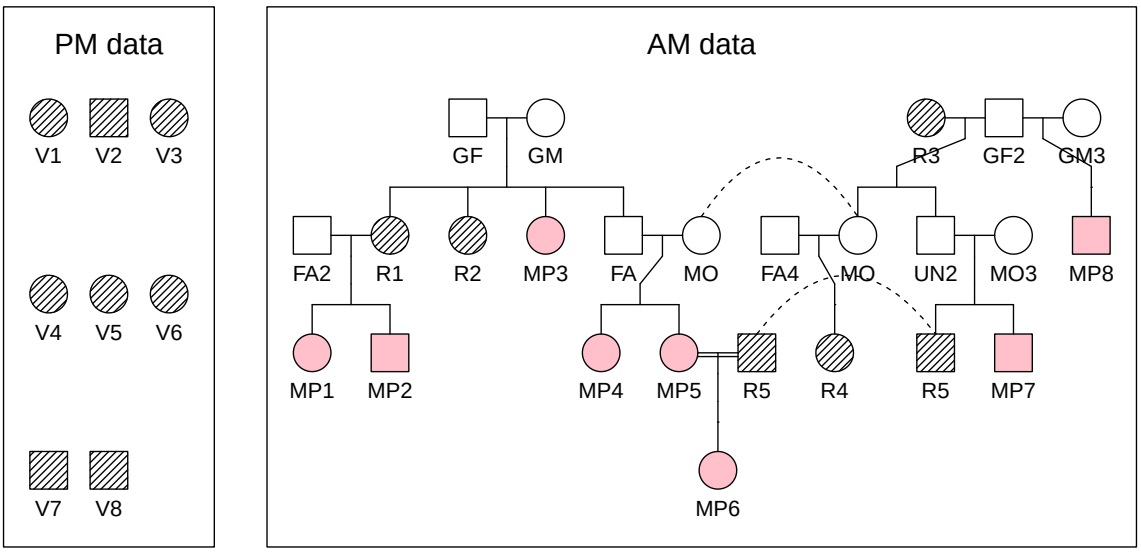
```
ip = missingPersonIP(ref, missing = "MP", nsim = 1000, threshold = 10000, seed = 17)
ip
```

i) Present the two power analyses in a *power plot* and comment on the output.

```
powerPlot(ep, ip)
```

Exercise VII-2 (DVI analysis, grave data)

We will analyse the DVI dataset `grave` included in the **dvir** package. This is a complex case and some commands may take a few minutes to run. Here is an overview of the data:



a) Familiarise yourself with the dataset by printing the `grave` object to the console, and also its main components `pm`, `am` and `missing`.

```
grave
grave$pm
grave$am
grave$missing
```

Finally, use the function `plotDVI()` to reproduce the plot shown above.

b) Find the total number of *a priori* possible solutions, using the `ncomb()` function as follows. Explain the input to the function.

```
ncomb(5, 5, 3, 3)
```

c) Describe the relationship between the parents of MP6. Find the inbreeding coefficient of MP6.

d) Use `pairwiseLR()` to compute the matrix of pairwise likelihood ratios. Explain what the entries of this matrix are. What is the LR for the pairing V1 = MP1?

```
prw = pairwiseLR(grave)
prw$LRmatrix
```

e) Explain the output from

```
excl = findExcluded(grave)
excl$exclusionMatrix
```

f) Explain the output from

```
und = findUndisputed(grave)
und$undisputed
```

g) Use `jointDVI()` to find the optimal (joint) solution, and inspect the top five alternatives. Comment on your findings.

h) Use the function `plotSolution()` to visualise the best joint solution.

Exercise VII-3 (DVI analysis continued, grave data)

This exercise continues on the previous. We will now use the function `dviSolve()`. This wraps several other functions into a complete pipeline for solving a DVI case.

a) Run

```
gr = dviSolve(grave)
```

and comment on the whether there are any

- problems with the data,
- nonidentifiable missing persons,
- exclusions.

b) Explain why a joint analysis is needed to find MP4 = V4 and MP5 = V5.

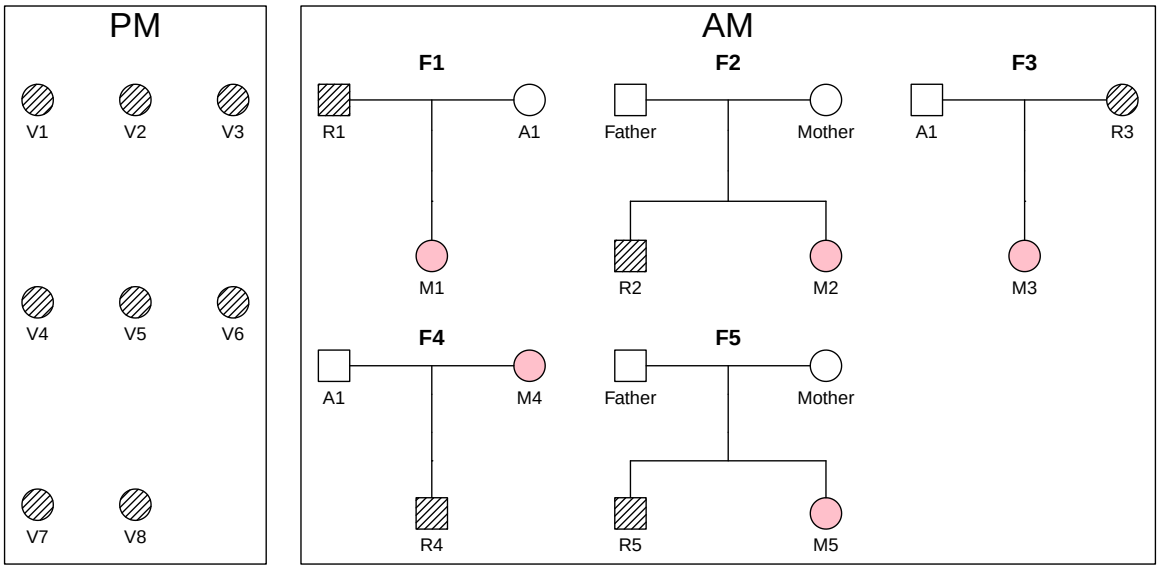
c) Assume we cannot trust the information on sex and choose to run

```
gr = dviSolve(grave, ignoreSex = T)
```

What conclusion can now be drawn about the missing persons `MP1` and `MP2`?

Exercise VII-4 (DVI analysis, Familias)

Consider a simulated crash of a plane with 10 passengers. We have obtained 8 PM samples, presumably from 8 different deceased individuals. There are 5 reference families, each with one missing person, see below figure.



a) Load the file planecrash.fam (available from the course home page) into Familias. Has a mutation model been specified? *Hint*: `File > Open` and then `Tools > General DNA data`.

b) Perform a search within the PM samples to see if there parent-offspring relationships, siblings or direct matches (identical samples). Use `Match threshold` 1000 and leave remaining parameters unchanged. *Hint*: `Tools > DVI module > Unidentified persons`and then `Blind search`.

c) Perform the search and formulate your conclusion. Use LR threshold 10000 and leave remaining parameters unchanged. *Hint*: `Tools > DVI module > Search` and then `Search`.