Pedigree analysis in R

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Solutions for exercise set III

Load libraries

```
library(pedsuite)
```

Some answers are given in the exercise. Here we add some comments and remaining answers.

Exercise III-1

a) We consider the following two hypotheses:

- H1: The alleged father (AF) is the biological father.
- H2: The alleged father and the child are unrelated.

b) We follow the approach of Lecture 3:

```
# Hypotheses
H1 = nuclearPed(fa = "AF", mo = "MO", child = "CH")
H2 = list(singleton("AF"), singleton("CH"))

# Allele frequencies
afr = c(a = 0.01, b = 0.30, c = 0.69)

# Create marker and attach to H1
m1 = marker(H1, AF = "a/b", CH = "a/c", afreq = afr)
H1 = setMarkers(H1, m1)

# Compute LR
kinshipLR(H1, H2, source = 1)
```
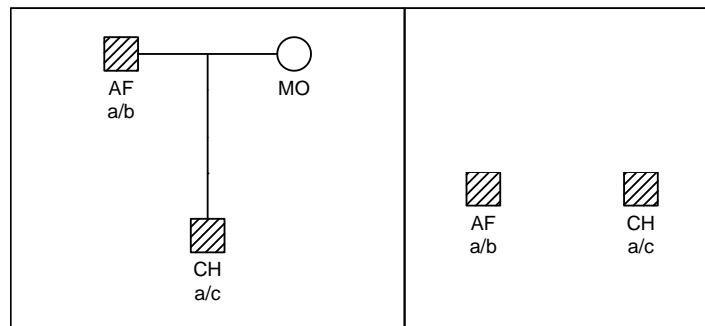
```
## H1:H2 H2:H2
##    25     1
```

c) A plot is produced by

```
plotPedList(list(H1, H2), marker = 1, source = 1, hatched = typedMembers)
```


d) We find LR $= \frac{2 p_a p_p b \frac{1}{2} p_c}{2 p_a p_p b 2 p_a p_c} = \frac{1}{4 p_a} = 25$.

Exercise III-2

Comment: You can alternatively download the zip file and extract the files, but there is no reason to do so unless you have problems with commands given in the exercise.

Exercise III-3

a) See exercise.

b) See exercise.

c) We find the LRs from

```
lr = kinshipLR(H1, H2, H3, H4, source = 4, verbose = T)

## Reference pedigree:  4
## Source pedigree:  4
## Number of markers:  15

lr
```

```
##     H1:H4     H2:H4     H3:H4     H4:H4
## 569.3989 805.1175 805.1175   1.0000
```

Comment: In this case only H4 has marker data attached. The argument `source = 4` secures that marker data from H4 are transferred to the other pedigrees. In this case the command would work also without using the `source` argument since, as stated in the documentation, then: "*If exactly one of the pedigrees have attached markers, these are transferred to the others.*" In the command above I included `source = 4` for clarity, although not strictly needed.

d) We add the pedigree and find the LRs:

```
H5 = relabel(cousinPed(1), old = 7:8, new = c("S1", "S2"))
lr = kinshipLR(H1, H2, H3, H4, H5, source = 4, verbose = T, ref = 4)

## Reference pedigree:  4
## Source pedigree:  4
## Number of markers:  15

lr
```

```
##     H1:H4     H2:H4     H3:H4     H4:H4     H5:H4
## 569.3989 805.1175 805.1175   1.0000 110.0958
```

Exercise III-4

a) *Comment*: Quite a bit of useful information is reported from the function. This information can be omitted by setting `verbose = F`.

b) See exercise.

c) The plot is produced by

```
H1 = dat[[1]]
H2 = dat[[2]]
plotPedList(list(H1, H2), shaded = typedMembers, marker = 1, titles = c("H1","H2"))
```

d) See exercise.

e) Marker 5, `PENTA_E`, has LR = 0. This is found by inspecting the LR-s for all markers:

```
res$LRperMarker
```

f) The answer can be obtained by running by running

```
prod(res$LRperMarker[-5])
```

```
## [1] 9611352944
```

Alternatively, marker 5 can be omitted in the LR calculation:

```
kinshipLR(H1, H2, markers = c(1:4, 6:21), ref = 2)
```

```
##       H1:H2        H2:H2
## 9611352944           1
```

Hence, excluding marker 5, `PENTA_E`, gives LR $= 9.61 \cdot 10^9$.

g) The total LR is 107148. For PENTA_E, we find LR $= 0.000011$.

h) We find

```
H2 = setMutationModel(H2, model = "proportional", rate = 0.00001)
lr2 = kinshipLR(H1, H2, ref = 2, source = 2)
lr2
```

```
##    H1:H2     H2:H2
## 107132.1     1.0
```

Comment: The total LR remains unchanged. There are minor differences for the individual markers as can be seen from

```
lr1$LRperMarker-lr2$LRperMarker
```

With a larger mutation rate, say 0.01, the LR would differ depending on whether mutation is modelled for all markers or not.

i) We find

```
H2 = setMutationModel(H2, model = "equal", rate = 0.00001)
kinshipLR(H1, H2, ref = 2, source = 2)
```

```
##    H1:H2     H2:H2
## 43253.77     1.00
```

j) The LR is evaluated for mutation rates $0.000001, 0.00001, \ldots, 0.1$. The resulting plot shows $\log 10(\mathrm{LR})$ as a function of $\log 10(rate)$ with variable names on the axis. The plot is improved below.
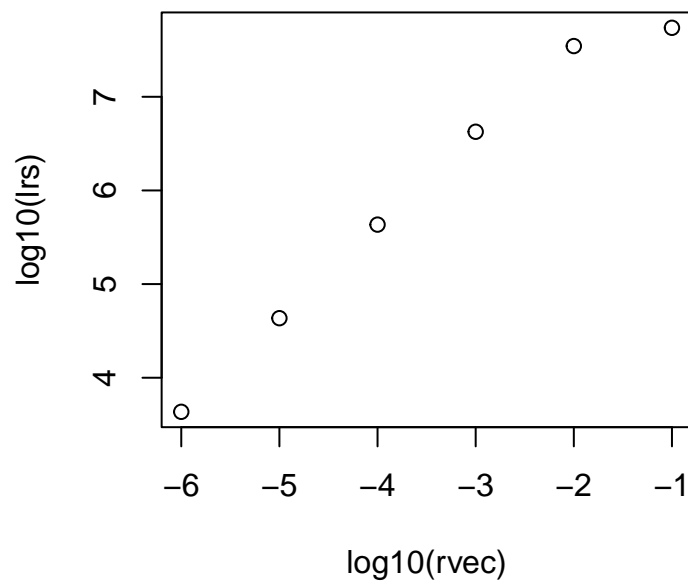
k) The plot command should be:

```
plot(log10(rvec), log10(lrs), xlab = "log10 mutation rate",
     ylab = "log10(LR)", main ="log10(lrMut). Equal model")
```