Kinship and pedigree analysis: Methods and applications

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Solutions for exercise set IV. Kinship testing in Familias

The parts omitted below are explained in the lecture `Kinship testing in Familias`.

Exercise IV-1

f) The LR comparing H1 to H3 is 1.96.

Exercise IV-2

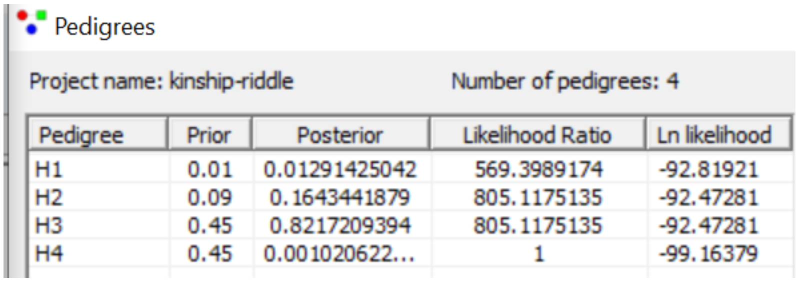
a) Use `scale` to get H4 in the denominator
b) We find

```r
LRs = c(569.399, 805.118, 805.118, 1)
round(LRs/sum(LRs), 3)
```

```
## [1] 0.261 0.369 0.369 0.000
```

Interpretation: the posterior probability of H3 is 0.369.

c) We modify the priors in `Familias` and find



Pedigrees

Project name: kinship-riddle

Number of pedigrees: 4

Pedigree	Prior	Posterior	Likelihood Ratio	Ln likelihood
H1	0.01	0.01291425042	569.3989174	-92.81921
H2	0.09	0.1643441879	805.1175135	-92.47281
H3	0.45	0.8217209394	805.1175135	-92.47281
H4	0.45	0.001020622...	1	-99.16379

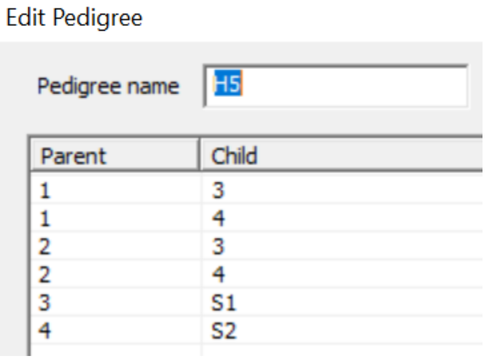
Figure 1: Familias output in IV-1 c)

Alternatively, we can use Bayes theorem:

```r
prior = c(0.01, 0.09,0.45, 0.45)
LRs = c(569.399, 805.118, 805.118, 1)
round(prior[3]*LRs[3]/sum(prior * LRs), 3)
```

```
## [1] 0.822
```

d) We find LR = 110.0958, as in the previous exercise. Here's a specification of H5 in Familias:



Pedigree name: H5

Parent	Child
1	3
1	4
2	3
2	4
3	S1
4	S2

Figure 2: Specifying H5 in Familias

Exercise IV-3

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

```
## Deprecated, use ‘pedFamilias::readFam()‘ instead.
## Reading from URL: http://familias.name/norbisRelatedness/paternityCase.fam
## Familias version: 3.2.8
## Read DVI: No
##
## Number of individuals (excluding ’extras’): 4
##   Individual ’CH’: Genotypes for 21 markers read
##   Individual ’AF’: Genotypes for 21 markers read
##   Individual ’MO’: Genotypes for 0 markers read
##   Individual ’TF’: Genotypes for 0 markers read
##
## Number of pedigrees: 2
##   Pedigree ’H1: father’ (0 extra females, 0 extra males)
##   Pedigree ’H2: not father’ (0 extra females, 0 extra males)
##
## Database: C:\Users\Thore\Dropbox\familias\sanAndres\data\ExS9Frequencies.txt
## Number of loci: 21
##   D3S1358: 11 alleles, unisex mut model = equal, rate = 0
##   THO1: 8 alleles, unisex mut model = equal, rate = 0
##   D21S11: 19 alleles, unisex mut model = equal, rate = 0
##   D18S51: 20 alleles, unisex mut model = equal, rate = 0
##   PENTA_E: 18 alleles, unisex mut model = equal, rate = 0
##   D5S818: 9 alleles, unisex mut model = equal, rate = 0
##   D13S317: 9 alleles, unisex mut model = equal, rate = 0
##   D7S820: 15 alleles, unisex mut model = equal, rate = 0
##   D16S539: 9 alleles, unisex mut model = equal, rate = 0
##   CSF1PO: 10 alleles, unisex mut model = equal, rate = 0
##   PENTA_D: 16 alleles, unisex mut model = equal, rate = 0
##   VWA: 12 alleles, unisex mut model = equal, rate = 0
##   D8S1179: 10 alleles, unisex mut model = equal, rate = 0
##   TPOX: 8 alleles, unisex mut model = equal, rate = 0
##   FGA: 21 alleles, unisex mut model = equal, rate = 0
##   D12S391: 19 alleles, unisex mut model = equal, rate = 0
##   D1S1656: 14 alleles, unisex mut model = equal, rate = 0
```

```
##   D2S1338: 11 alleles, unisex mut model = equal, rate = 0
##   D22S1045: 8 alleles, unisex mut model = equal, rate = 0
##   D2S441: 9 alleles, unisex mut model = equal, rate = 0
##   D19S433: 11 alleles, unisex mut model = equal, rate = 0
##
## Converting to ‘ped‘ format
```

b) See exercise.

c) The plot is produced by

```r
plotPedList(dat, hatched = typedMembers, marker = 1)
```

d) See exercise.

e) See exercise.

f) Marker 5, PENTA_E, has LR = 0, as can be seen by inspecting the individual LRs:

```r
res$LRperMarker
```

g) Excluding PENTA_E gives LR = $9.61 \cdot 10^9$. This can be found by the command

```r
kinshipLR(H1, H2, markers = -5)
```

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

Alternatively, we can multiply the LRs of all markers except PENTA_E:

```r
prod(res$LRperMarker[-5, 1])
```

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

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

```r
H2 = setMutmod(H2, marker = "PENTA_E", model = "proportional", rate = 0.00001)
lr = kinshipLR(H1, H2, source = 2)
lr
```

```
##  H1:H2  H2:H2
## 107148      1
```

```r
lr$LRperMarker["PENTA_E",]
```

```
##        H1:H2        H2:H2
## 1.114807e-05 1.000000e+00
```

i) We find LR = 107132:

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

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

Comment: The LR hardly changed when applying the model to all markers. This is reassuring, and supports that one should always do this whenever computationally feasible.
A marker-wise comparison between the two methods confirms the minuscule changes:

```
lr$LRperMarker / lr2$LRperMarker
```

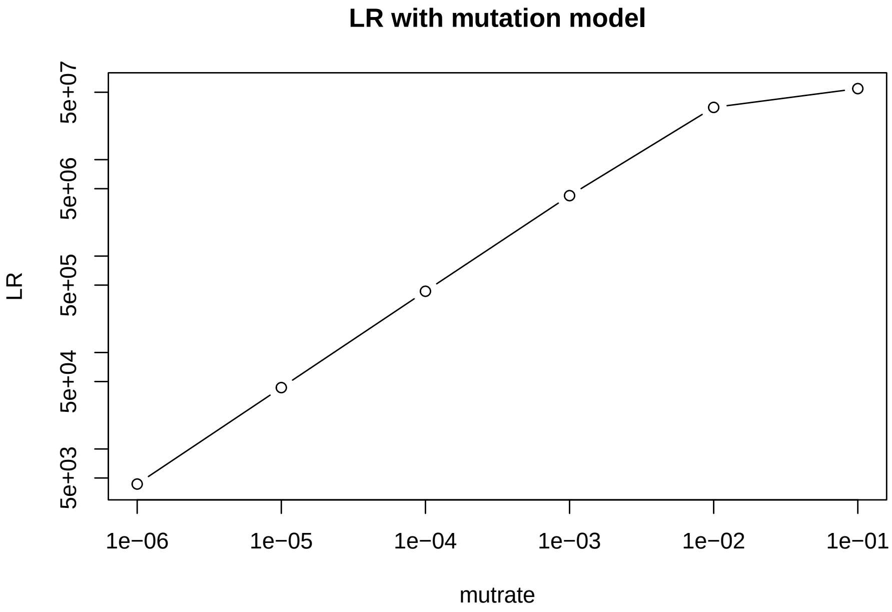
```
##             H1:H2 H2:H2
## D3S1358  1.000007     1
## TH01     1.000002     1
## D21S11   1.000001     1
## D18S51   1.000006     1
## PENTA_E  1.000000     1
## D5S818   1.000004     1
## D13S317  1.000009     1
## D7S820   1.000004     1
## D16S539  1.000011     1
## CSF1PO   1.000007     1
## PENTA_D  1.000011     1
## VWA      1.000010     1
## D8S1179  1.000011     1
## TPOX     1.000007     1
## FGA      1.000008     1
## D12S391  1.000006     1
## D1S1656  1.000008     1
## D2S1338  1.000008     1
## D22S1045 1.000013     1
## D2S441   1.000004     1
## D19S433  1.000009     1
```

j) With the `equal` model, we find

```
H2 = setMutmod(H2, model = "equal", rate = 0.00001)
lr3 = kinshipLR(H1, H2, source = 2)
lr3
```

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

k) The LR is evaluated for mutation rates 0.000001, 0.00001, . . . , 0.1. The resulting plot shows LR as a function of the mutation rate, using a logarithmic scale on both axes.


LR with mutation model
LR
5e+03
5e+04
5e+05
5e+06
5e+07
1e−06
1e−05
1e−04
1e−03
1e−02
1e−01
mutrate