

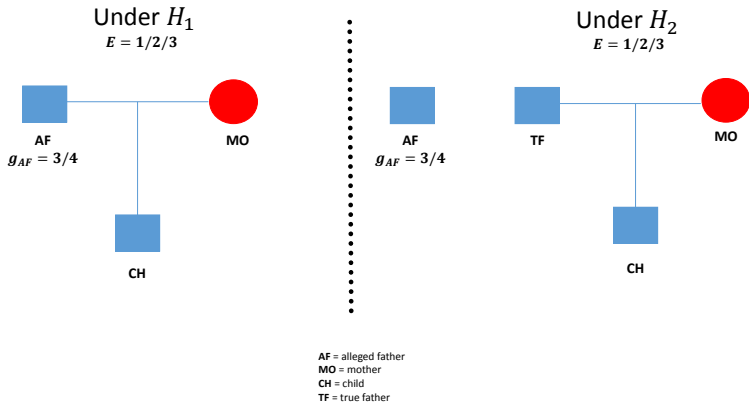
## Mixtures and relatives. reIMix demo. III.2

Thore Egeland<sup>(1),(2)</sup>, Klaas Slooten<sup>(3),(4)</sup>

(1) Norwegian University of Life Sciences, (2) NIPH, (3) Netherlands Forensic Institute, (4) VU University Amsterdam

Seoul, ISFG workshop, Aug 29 2017

# Combining mixtures and reference samples



## Notation [1]

- ▶  $E$  mixture, e.g.  $E = 1/2/3$ .
- ▶  $g_1, \dots, g_N$  reference samples, e.g.,  $g_1 = g_{AF} = 3/4$ .
- ▶  $u_1, \dots, u_K$  untyped, e.g.,  $u_1 = u_{MO}$ .

The basic calculation involves computing

$$\begin{aligned} P(\text{data} \mid H_t) &= P(E, g_1, \dots, g_N \mid H_t) \\ &= \sum_{u_1, \dots, u_K} P(u_1, \dots, u_K, g_1, \dots, g_N \mid H_t). \end{aligned}$$

Problem reduced to summing kinship likelihoods.

# relMix: R library for mixtures with relatives

```
> install.packages("relMix") #Installs R library relMix. Once  
Installing package into 'C:/R/library'  
(as 'lib' is unspecified)  
Warning: package 'relMix' is in use and will not be installed  
> library(relMix) # loads library relMix. Every R session  
> relMixGUI() # Starts GUI
```

The screenshot shows the relMix GUI window. The window title is "RelMix". The interface is organized into several sections:

- Import data:** Contains three buttons: "Import mixture profile", "Import reference profiles", and "Database".
- Mutations:** Contains one button: "Mutations".
- Pedigrees:** Contains two dropdown menus labeled "Pedigree 1" and "Pedigree 2".
- Contributors:** Contains one button: "Specify contributors".
- Dropout and drop-in:** This section is partially visible at the bottom of the window.

## Demo. Input files come with relMix

- ▶ Import mixture profile [mixture.txt](#):

```
SampleName Marker Allele1 Allele2 Allele3
relMix1 CSF1P0 10 12
relMix1 D10S1248 14 16
relMix1 D13S317 8 12 13
...
```

- ▶ Import reference profile [references.txt](#):

```
SampleName Marker Allele1 Allele2
Mother CSF1P0 12 12
...
Father CSF1P0 10 10
```

- ▶ Database [frequencies22Markers.txt](#)
- ▶ Mutations
- ▶ Pedigrees. Here default.
- ▶ Specify contributors: Mother and Child **both** hypotheses.
- ▶ Specify dropout and drop-in

# reMix: Output

## Database

Parameter	Value
theta	0
Silent allele freq.	0
Min. allele freq.	0.001

## Mutations

Parameter	Value
Model	Stepwise
Range	0.1
Female mut. rate	0.001
Male mut. rate	0.005

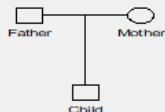
## Contributors

Pedigree 1	Pedigree 2
Mother	Mother
Child	Child

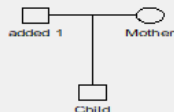
## Dropout and drop-in

Parameter	Value
-----------	-------

## Pedigree 1



## Pedigree 2

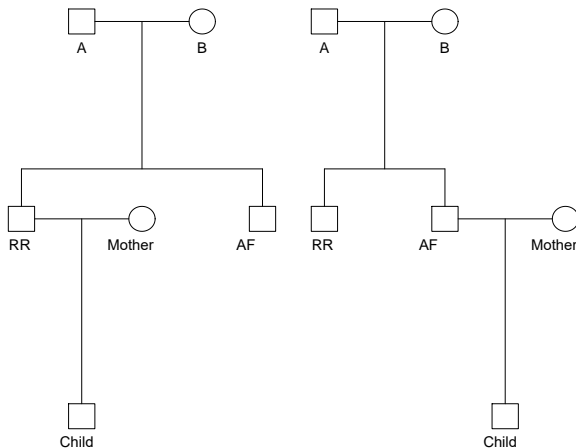


Total LR: 1.195e+05

Marker	LR	Rectangular Snip
D16S539	8.744e+00	
D19S433	1.952e+00	
D1S1656	1.761e+00	
D1S1677	2.110e+00	
D2S1338	4.128e+00	
D2S1776	1.549e+00	
D2S441	2.368e+00	
D3S1358	1.828e+00	
D4S2408	2.526e+00	
D5S2500	2.162e+00	
D5S010	1.107e+01	

## Example 2. Real case provided by Nadia Pinto.

- ▶ **Data:** Mother, Mother–Child mix, brothers RR and AF.
- ▶ **Which brother is the father?**
- ▶ Need to specify pedigrees on R Familias format ...



# reMix: Output

## Database

Parameter	Value
theta	0
Silent allele freq.	0
Min. allele freq.	0.001

## Mutations

Parameter	Value
Model	Equal
Female mut. rate	0.005
Male mut. rate	0.005

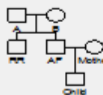
## Contributors

Pedigree 1	Pedigree 2
Child	Child
Mother	Mother

## Pedigree 1



## Pedigree 2



Total LR: 1.703e+18

Marker	LR
CSF1PO	1.000e+00
D10S1248	1.000e+00
D12S391	5.002e-01
D13S317	3.986e+02
D16S539	1.999e+00
D18S51	1.000e+00
D19S433	1.000e+00
D1S1656	1.998e+00





Navreet Kaur, Mariam M Bouzga, Guro Dørum, and Thore Egeland.

Relationship inference based on DNA mixtures.

*International Journal of Legal Medicine*, 130(2):323–329, 2016.