

Familias and Forensic genetics

Thore.Egeland@umb.no
Norwegian University of Life Sciences

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Purpose

"..practical workshop about the statistical evaluation of forensic cases (paternity and other family relationships) by using Familias"

Contents

- Principles
- Statistical evaluation
 - LR (Paternity index)
 - Non-standard cases
 - Theta-correction
 - Mutations
 - Complex pedigrees
 - Silent alleles.
- Familias. Exercises
- Course homepage arken.umb.no/~theg/familias/
user: *familias*
pass word: *andres*

Part I: 09-10

- Review of basic forensic genetics
- Spanish/Portuguese summary

Principles

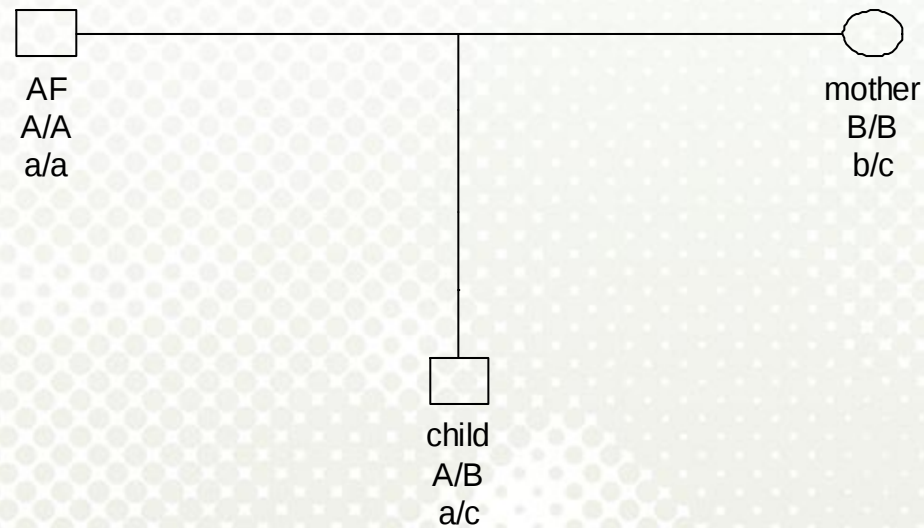
1. To evaluate the uncertainty of any given proposition it is necessary to consider at least one alternative proposition.
2. Scientific interpretation is based on questions of the kind: "What is the probability of the evidence given the proposition?"
3. Scientific evidence is conditioned not only by the competing propositions, but also by the framework of circumstances within which they are to be evaluated

Books: Evett&Weir, Balding, Buckleton et al.

Exercise S1 Standard paternity case

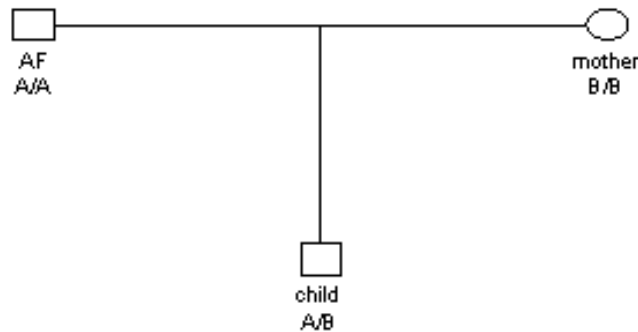
- H_1 : The alleged father (AF) is the real father
- H_2 : AF and the child are unrelated.

Figure1. Standard paternity case. Two markers



Exercise S1 a) Calculate LR. First marker

Figure1. Standard paternity case



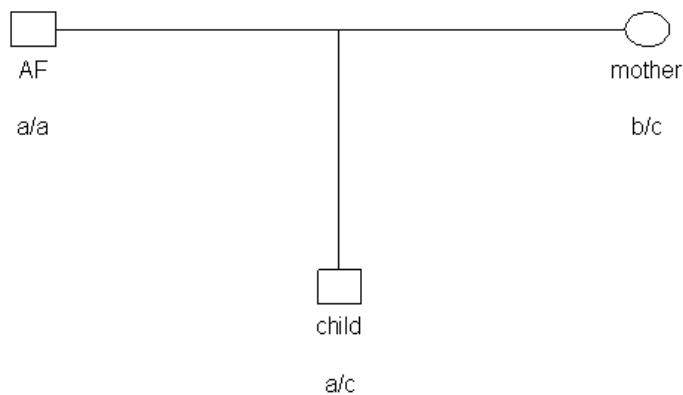
$$LR = \frac{P(\text{child} \mid \text{mother}, \text{AF})}{P(\text{child} \mid \text{mother})} = \frac{1}{p_A} = \frac{1}{0.05} = 20.$$

- *Interpretation:*

“The data is 20 times more likely assuming AF to be the father compared to the alternative that some unknown man is the father”.

Exercise S1 c) Calculate LR. Second marker

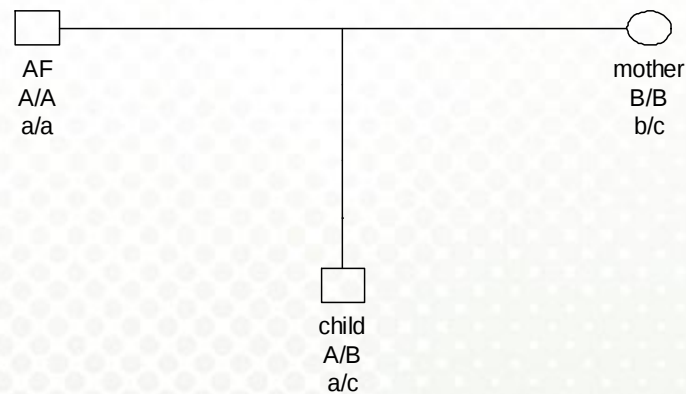
Figure1. Standard paternity case



$$LR = \frac{P(\text{child} \mid \text{mother}, \text{AF})}{P(\text{child} \mid \text{mother})} = \frac{0.5}{0.5p_a} = \frac{0.5}{0.5 * 0.1} = 10.$$

Exercise S1 c) Calculate LR. Two markers

Figure1. Standard paternity case. Two markers



$$LR = 20 * 10 = 200.$$

- *Interpretation:*
"The data is 200 times more likely assuming AF to be the father compared to the alternative that some unknown man is the father".
- *Assumptions?*

Non-standard cases

- Complications
 - Theta-correction
 - Mutations
 - Complex pedigrees
 - Silent alleles

Part II: 10-1115: Introduction to Familias. Demo.

- Exercises S1-S3, S5. *Plenum*
- *Videos available*
- Spanish/Portuguese summary

Part III: 1145-1230: Hands on Familias exercises

- Exercises S1-S6

Part IV: 14-16
Repetition.
Exercises S4, S6. Plenum
Non-standard cases

Non-standard cases

- Complications
 - Theta-correction
 - Mutations
 - Complex pedigrees
 - Silent alleles

Theta - correction

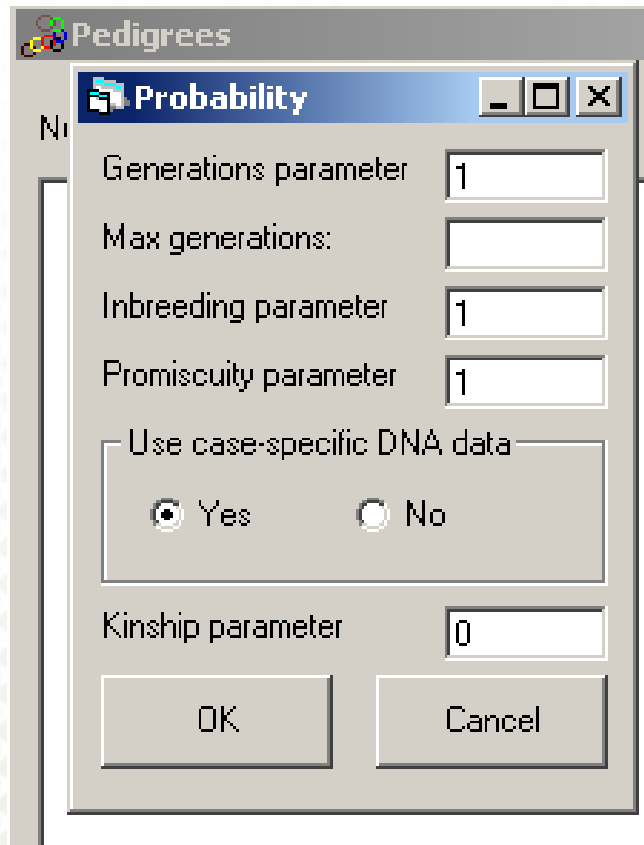
Homozygous A, A: $\theta p_A + p_A^2(1 - \theta)$,

Heterozygous A, B: $2 p_A p_B(1 - \theta)$.

Special cases:

$$\theta=0$$

$$\theta=1$$

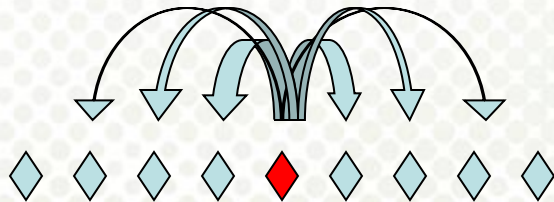


← θ

Revisit Exercise S1h (plenum)

Modelling mutations

- Mutation rate varies with
 - Sex of parent and locus.
 - Alleles tend to mutate to close alleles:
 - Several models



Mutations

Female mutations:

Mutation rate:

☒ Prob. decreasing with range (stable)
☐ Prob. decreasing with range (equal)

Mutation range:

☐ Probability proportional to frequency (stable)
☐ Equal probability (simple and fast)

Male mutations:

Mutation rate:

☒ Prob. decreasing with range (stable)
☐ Prob. decreasing with range (equal)

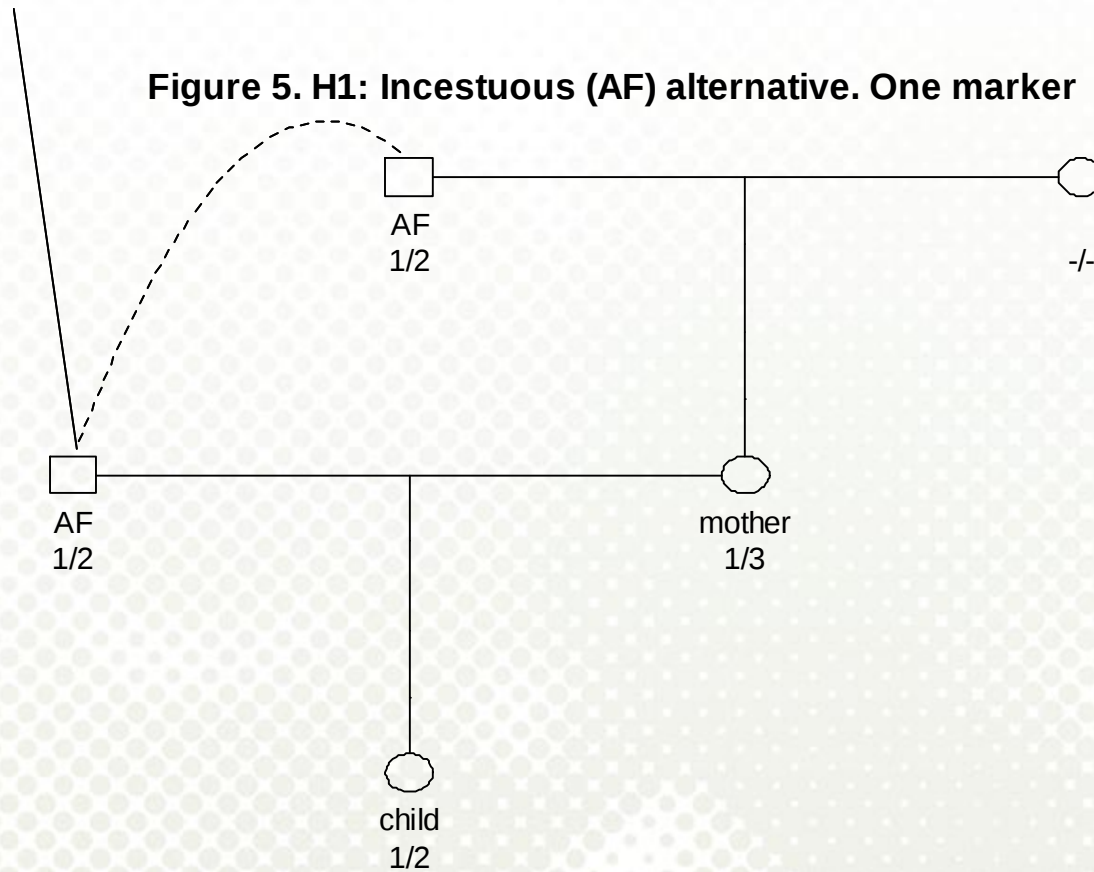
Mutation range:

☐ Probability proportional to frequency (stable)
☐ Equal probability (simple and fast)

Revisit Exercise S2, S7

Complex pedigrees I. Exercise S6

Figure 5. H1: Incestuous (AF) alternative. One marker



Complex pedigrees II. Exercise S6

familias - ExS6 - [Pedigree]

File Window Help

File Edit View Options Database Help

Casenotes

Pedigree Name:

Parent	Child
mother	child
AF	child
AF	mother

OK

Persons

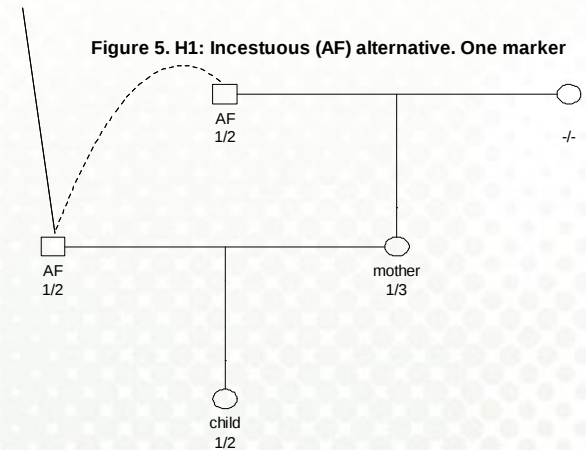
Remove

Add

Parent

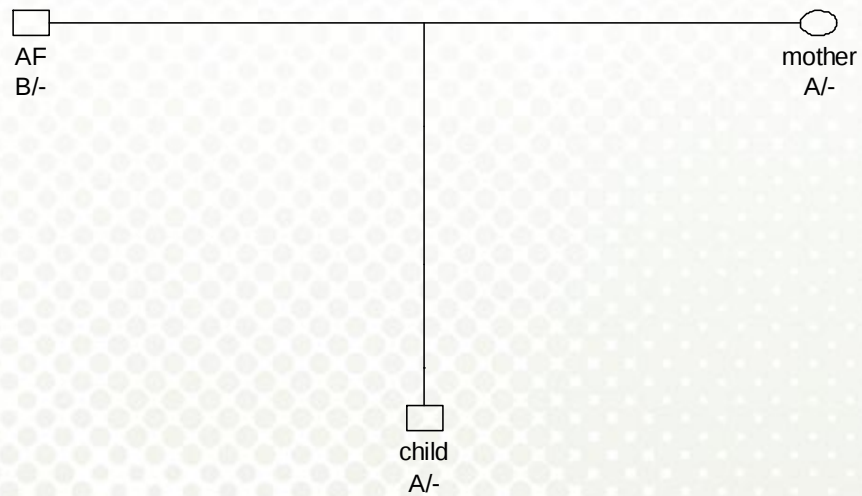
Child

Add



Silent alleles I. Exercise S11

Figure 5. Silent allele?



Silent alleles II. Familias

Silent
allele



Allele system

System name:

Silent frequency:

Alleles:	Frequencies:
A	0,1
B	0,1
Extra allele	0,75

Apply

OK

Remove Allele

Edit Allele

Allele name:

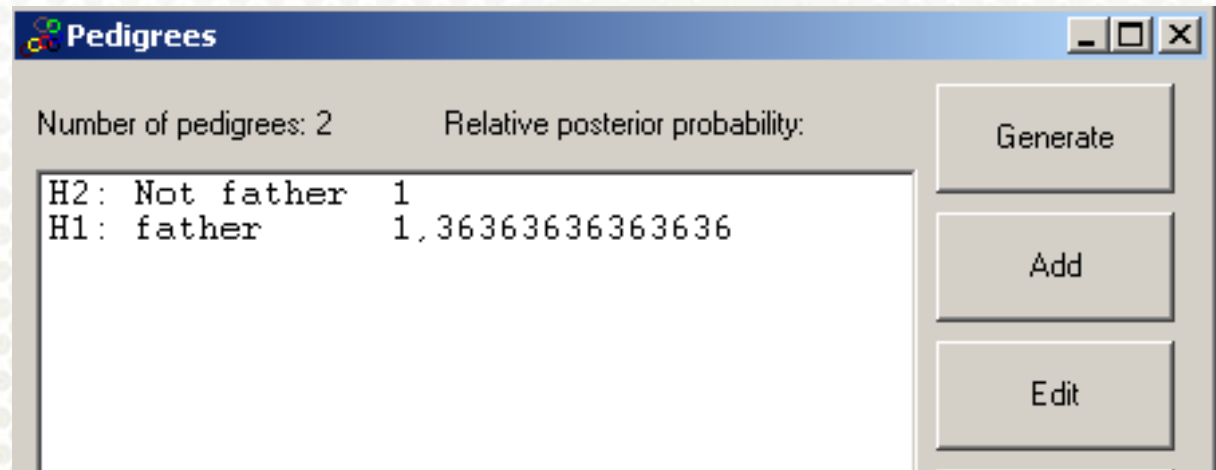
Frequency:

Add

Silent alleles III. Exercises S11 (S12 theoretical)

$$LR = \frac{p_s (p_A + p_s)}{(p_A + p_s)^2 (p_B + 2p_s) + p_s p_A (p_B + 2p_s)}$$

$$= \frac{0.05 \cdot 0.15}{0.15^2 \cdot 0.2 + 0.05 \cdot 0.1 \cdot 0.2} = 1.36.$$



Part V: 16-17

Hands on exercises: S9, S10, S11

If time: 'Extra' exercises on homepage:

arken.umb.no/~theg/familias/

Part VI: 17-18

Discussion of exercises.

Spanish/Portuguese summary.

- Questions?
- Exercises S1h, S4c, S5d, S9d

References

- Familias reference:
[Egeland, Mostad et al.](#), see also
<http://familias.name/>

