A contribution of mtDNA to a particular case of identification

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Scientific identification is a probabilistic approach

Population

Threshold

Reduction factors

Individual

Two kinds of reference for a DNA identification
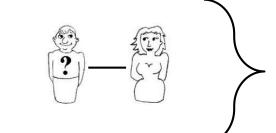
Post-mortem

- ✓ blood
- ✓ saliva
- ✓ nail
- ✓ muscle
- ✓ bone
- ✓ tooth

Ante-mortem

Personal reference

« civil identity »

Familial reference

« biological identity »

The forensic medicine unit asked for a DNA identification

- ✓ Blood on FTA from (?) Miss Tick (alleged identity)
- ✓ Buccal swab from Mr T., Miss Tick's brother

- ➢ Fast DNA analyses : NGM Select + ESI17

	Mr. T.	Sister ?
D10S1248	13-14	13-15
vWA	14-17	16-18
D16S539	8-12	11-12
D2S1338	17-18	22-24
Amelogenine	XY	XX
D8S1179	13-13	13-13
D21S11	29-31	29-30
D18S51	10-14	13-14
D22S1045	11-16	15-16
D19S433	13-13	13-14
TH01	6-9	6-9
FGA	21-25	20-24
D2S441	10-11	11-11.3
D3S1358	15-17	16-16
D1S1656	12-18.3	14-17
D12S391	18-19	19-20
SE33	25.2-29.2	15-23.2

Using a bayesian approach

H1 : The deceased and Mr T. are full siblings
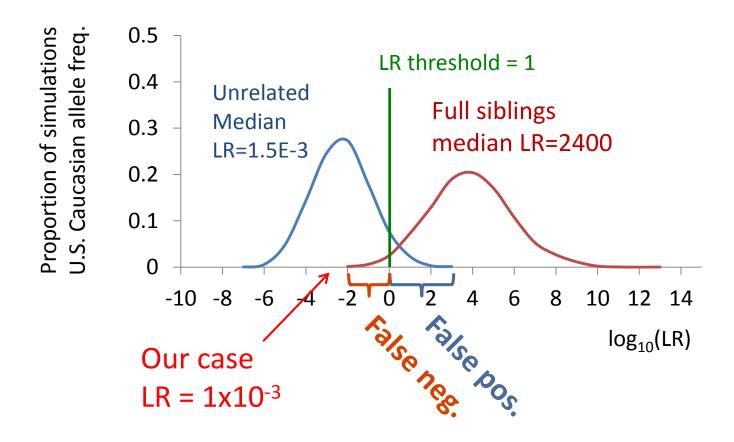**H2 : The deceased and Mr T. are unrelated**

Thomas Bayes
1701-1761

$$\textbf{\textit{nDNA LR}} = \frac{\Pr(E \mid H1, I)}{\Pr(E \mid H2, I)} = \textbf{0.001}$$

The Swiss allele frequencies (Gehrig *et al.* Int. J. Legal Med. 128 (2014) 461–465) and classical formulas without F_{ST} correction were used for assigning the LR!

Our results support H2. Our findings are about 1'000 times more probable if the deceased and Mr T. are unrelated rather than full siblings !

LR distributions for full siblings and unrelated individuals from 5'000 simulations (J.M. Butler, Advanced Topics in Forensic DNA Typing: Interpretation, Academic Press, 2015)
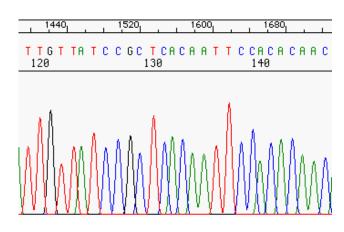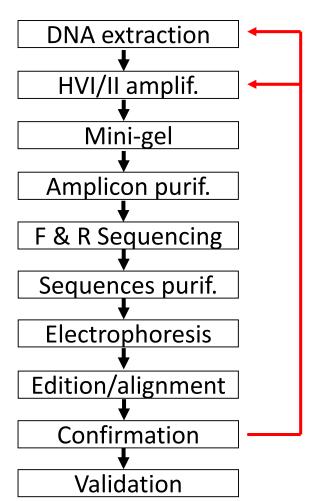
What about contextual information ?

- Deceased alone within her apartement.

- Doors locked from the inside.

- She slit her wrist and wrote a suicide note.

- Age (76 y.o.), height, weight and hairs corresponding to those of Miss Tick.

- No other missing person in the village !

- Miss Tick has only 2 brothers, none of them had children !

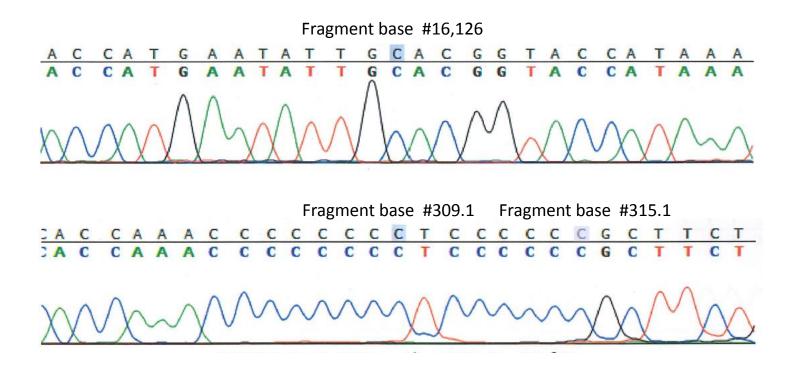Mitochondrial DNA is maternally inherited. **Siblings have the same mitotype !**

Sanger sequencing

DNA extraction

↓

HVI/II amplif.

↓

Mini-gel

↓

Amplicon purif.

↓

F & R Sequencing

↓

Sequences purif.

↓

Electrophoresis

↓

Edition/alignment

↓

Confirmation

↓

Validation

The deceased and Mr T. have the same mitotype !

HVI : 15928A, **16126C**, 16153A, 16242T, 16294T, 16320T
HVII : 73G, 150T, 152C, 263G, **309.1C**, **315.1C**

Fragment base #16,126

Fragment base #309.1   Fragment base #315.1

Using a bayesian approach

H1 : The deceased and Mr T. are full siblings
H2 : The deceased and Mr T. are unrelated

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1701-1761

$$mtDNA\ LR = \frac{Pr(E \mid H1, I)}{Pr(E \mid H2, I)} = \frac{n + 3}{x + 2} = 2'200$$

x = 0 observation within a database with n = 4'374 European mitotypes (http://empop.org) ; LR assignment using a bayesian estimator (J. Buckleton, M. Triggs, S. Walsh, Forensic DNA evidence interpretation, CRC press, 2005)

Our results support H1. Our findings are about 2'200 times more probable if the deceased and Mr T. are full siblings rather than unrelated !

Combining our analytical results.

Our findings are about **2 times** more probable if the deseased and Mr T. are full siblings rather than unrelated. A very weak support to H1...

What if the deceased and Mr T. would be half siblings ?

H1 : The deceased and Mr T. are half siblings
H2 : The deceased and Mr T. are unrelated

$$nDNA\ LR = \frac{Pr(E \mid H1, I)}{Pr(E \mid H2, I)} = 0.1$$

H2 is still moderaltly supported ; but the **combined LR = 200**

Conclusions

➢ Final report : our nDNA results do not support the full sibling relationship, but our mtDNA results support the hypothesis that the deceased and Mr T. belong to the same matriline. **The forensic pathologist therefore concluded that the deceased was Miss Tick**.

➢ MtDNA may represent a usefull complement to nDNA analyses for some parentage and identification cases.

➢ Familial references used for cadaver identification have limitations (false positives or negatives, adoption, gamete donation, adultery,…).

➢ Pr(E | H, I) : **DNA evidence has to be assessed taking into account the context of the case**.