**GRUPO DE HABLA ESPAÑOLA Y PORTUGUESA DE LA ISFG**

**GRUPO DE LÍNGUAS ESPANHOLA E PORTUGUESA DA ISFG**

ghep-isfg

SPANISH AND PORTUGUESE SPEAKING WORKING GROUP OF ISFG

## Forensic paper challenge, advanced level
## GHEP 2012

Lourdes Prieto

Forensic paper challenge, advanced level
GHEP 2012

▸ Participants and aims
▸ Part I: what samples would you analize?
  ▸ Approach, answers and comments
▸ Part II: evaluations
  ▸ Approach, answers and comments
▸ Suggestions
▸ Conclusions

▸

## Participants and aim

- 43 Laboratories
- Objectives:
  - To know the handling of missing person samples (personal effects)
    - Are they considered as reference samples?
    - Should we «authenticate» them?
  - To know the types of comparisons (MIs and/or LRs) that the labs carry out
    - maternity?
    - identity?
    - both?
    - aSTRs?
    - mtDNA?
    - both?
  - To know the handling of point heteroplamies
  - To know if labs combine aSTR and mtDNA information

PART I

PART II

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## Approach part I

▸ A mother denunciates a missing son (Individual A). In order to facilitate a possible identification of this individual A in the future, this mother leave a biological sample (**sample 1**) and also provides personal belongings of the missing son (**sample 2**).
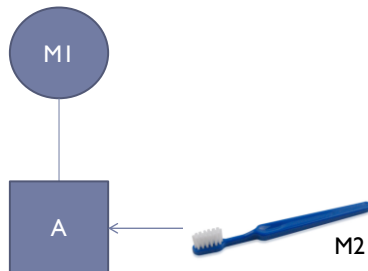


▸ Part I: samples that you would analyze

## Approach part I
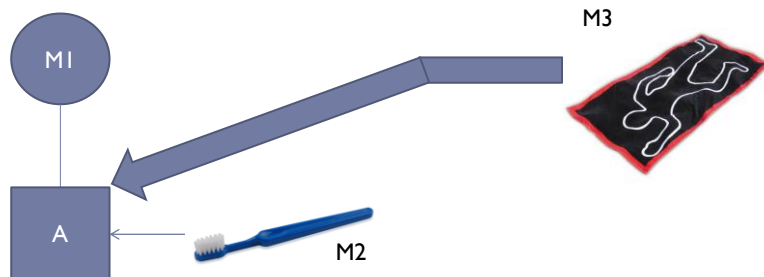
▸ Would you analyze both samples?
  ▸ YES
  ▸ NO
▸ If your response above is NO, please indicate which samples would you analyze:
  ▸ Sample of the mother of the missing son (sample 1)
  ▸ Personal belonging of the missing son (sample 2)
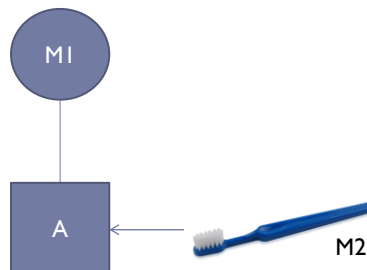


▸ Part I: samples that you would analyze

## Approach part I

▸ Two months later, an unidentified human body is found in Madrid. The authorities consider the hypothesis that these remains could belong to the son of the mother that denunciated its disappearance. The authorities take a biological simple from the body (sample 3)



▸ Parte I: samples that you would analyze

## Answers part I

▸ Would you analyze both samples?

37 | 6

▸ If your response above is NO, please indicate which samples would you analyze: **M1 (all labs)**



| LAB |
|---|
| 20166 |
| 20197 |
| 20218 |
| 20264 |
| 20269 |
| 20280 |

▸ Part I: samples that you would analyze

# Only analysis on M1 and M3: comments

▸ 20166 → **«lack of authentication of M2»**
  ▸ «The personal effect (sample 2) was not analysed since «authentication» is needed in Colombia, with the porpoer chain of custody and the sample should be endorsed by judicial police under the supervision of the prosecutor».

▸ 20218 → **it's better to analyze M2**
  ▸ It is better to choose the sample 2 compared to 1 because:
    ▸ the LRs is higher than that M1
    ▸ the mitochondrial DNA analyses are the same between sample 2 and sample 3 (no heteroplasmy)

▸ Part I: samples that you would analyze

# Only analysis on M1 and M3: comments

▸ 20264 → **availability of full profiles of M1 and M3 samples**
  ▸ Firstly, being available the sample from the mother we would not analyze the personal effect from the missing son. Secondly, as a full aSTR profile was obtained from the unidentified body sample, we would proceed to the maternity investigation by using aSTRs results from the mother and the body.
▸ 20184 → without comments
▸ 20197 → without comments
▸ 20269 → without comments

▸ Part I: samples that you would analyze

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## Approach part II

▸ The genetic analysis results are displayed in the following tables:

| SAMPLE | aSTRs | mtDNA |
|---|---|---|
| MOTHER (M1) | FULL PROFILE (17) | 16024-576 |
| PERSONAL EFFECT (M2) | PARTIAL PROFILE (9) | 16024-576 |
| BODY (M3) | FULL PROFILE (17) | 16024-576 |

WITHOUT EXCLUSIONS

MOTHER: HG T2 with **152Y**
BODY AND EFFECT: HG T2 without polymorphism in 152

▸ Part II: evaluation

# Approach part II

▸ Compute LRs and/or MIs that you would consider needed according to question 1, specifying your hypotheses

▸ Posibilities:
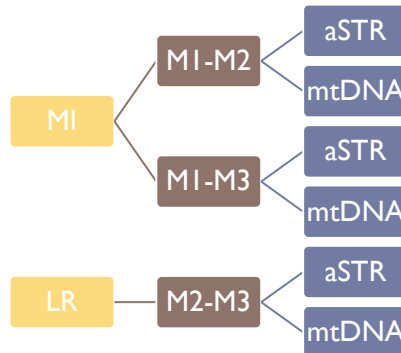


▸ Part II: evaluation

# Answers II:

MIndex ➡ MI-M3 ➡ aSTR

DATO CONSENSUADO: ~148.900

| MIndex | Num LABS |
|--------|----------|
| ~148.900 | 33 |
| 5.540 | 1 |
| 61.081 | 1 |
| 75.400 | 1 |
| 153.320 | 1 |
| 804.707 | 1 |
| Partials | 1 |
| Not reported | 4 |
| TOTAL | 43 |

2 • Calculate Maternity index of M2 and M3

1 • Report calculations including M1, M2 and M3 (LR =3,1484e+12)

1 • Report Probability of Maternity without specifying prior (99,9999948%)
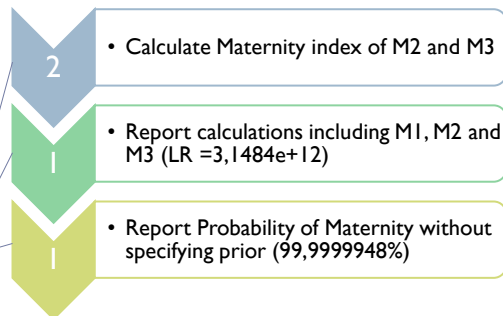
▸ Part II: evaluation

# Answers II:

M Index ➡ MI-M3 ➡ mtDNA

**MOTHER**
- 16126C 16294T 16304C 16519C 73G **152Y** 263G 315.1C

**EFFECT**
- 16126C 16294T 16304C 16519C 73G 263G 315.1C

**BODY**
- 16126C 16294T 16304C 16519C 73G 263G 315.1C

▸ Searching parameters
  ▸ Using EMPOP
  ▸ Disregard indels in length variants at positions 16193, 309, 455, 573
  ▸ Geographic affiliation: Europe, Metapopulation: all

▸ Part II: evaluation

# Answers II:

M Index ➡ MI-M3 ➡ mtDNA

| M Index | Num LABS |
|---|---|
| Not reported | 26 |
| Frequency | 3 |
| 473 | 1 |
| 361-325 | 1 |
| 315 | 2 |
| **296** | **2** |
| **283** | **2** |
| 257 | 3 |
| 250 | 1 |
| 236 | 1 |
| 165 | 1 |
| TOTAL | 43 |

LACK OF CONSENSUS

- The mt haplotype was observed in 2,764 x 10EXP-3 times in the database containing 3100 haplotypes. With a frequency of 8,8387 x10exp-7

- Haplotype frequency of the body (2.823E-03)

- Haplotype frequency of Sample 1 (3,8801E-03)

▸ Part II: evaluation

## Answers II:

M Index ➡ MI-M3 ➡ mtDNA

RESULTS OF QUERIES

| EMPOP R-6 (until 24-4-12) | EMPOP R-7 (from 25-4-12) |
|---|---|
| Sample1 (with 152Y) 10/2834 | Sample1 (with152Y) 11/3256 |
| Freq. = 3,529e-3 M Index = 283,4 | Freq. = 3,378e-3 M Index = 296 |

▸ Part II: evaluation

## Answers II:

M Index ➡ MI-M3 ➡ mtDNA

▸ **Do not report: 26 labs**
  ▸ Without comments: 21
  ▸ Make any comment about the difference in 152 and maternity was not excluded: 3
  ▸ «mtDNA haplotype would not be used in the identification» (calculate LR for M2-M3)
  ▸ «As aSTRs results are informative sufficiently, we would not analyze mtDNA»

▸ **Other labs make also comments:**
  ▸ «In this case the values are not significant (referred to mt LR), and do not supply additional information to the aSTR results»

▸ Part II: evaluation

# Dangers: not real matches

| Lourdes Profile | ➡ | 15 aSTR | ➡ | Database 180.404 | ➡ | 30 sons |

| | MI | MP (%) | | MI | MP (%) | | MI | MP (%) |
|---|------|--------|---|------|--------|---|-------|--------|
| 1 | 12 | 92,664 | 1 | 410 | 99,757 | 1 | 1475 | 99,932 |
| 1 | 46 | 97,893 | 1 | 425 | 99,766 | 1 | 1551 | 99,936 |
| 1 | 62 | 98,437 | 1 | 435 | 99,771 | 1 | 2040 | 99,95 |
| 3 | 64 | 98,47 | 1 | 444 | 99,776 | 1 | 2375 | 99,96 |
| 1 | 102 | 99,04 | 2 | 448 | 99,778 | 1 | 2455 | 99,96 |
| 1 | 124 | 99,2 | 1 | 457 | 99,782 | 1 | 2792 | 99,964 |
| 1 | 128 | 99,23 | 1 | 580 | 99,828 | 1 | 4764 | 99,98 |
| 1 | 188 | 99,473 | 1 | 770 | 99,87 | 1 | 5916 | 99.983 |
| 1 | 343 | 99,71 | 1 | 1007 | 99,90 | 1 | 8131 | 99,988 |
| 1 | 406 | 99,75 | 1 | 1182 | 99,92 | 2 | 18662 | 99,995 |

**mtDNA data could be important to exclude not real compatibilities**

▶ Part II: evaluation

# Answers II:

| LR | ➡ | M2-M3 | ➡ | aSTR |

**CONSENSUS DATA: ~ 2,20E+09**

| LR | Num LABS |
|----------|----------|
| ~ 2,20E+09 | 27 |
| 1,82E+09 | 1 |
| 4,39E+09 | 1 |
| 1,01E+10 | 1 |
| 3,15E+12 | 1 |
| Not reported | 7 |
| TOTAL | 38 |

- **3** • Without comment
- **1** • Only M2 profile would be used to perform a query in the missing person database
- **1** • Both samples were analyzed in order to determine the fact that Samples 1 belongs to the mother of the misisng person.
- **1** • The personal effect was used to confirm the results in the sample from the body, but it was not used to determine the identification, as it is an unknown sample
- **1** • The profile from the body is identical to the one from the personal effect in 9 STR, Amelogenine marker and mtDNA.

▶ Part II: evaluation

## Answers II:

LR → M2-M3 → mtDNA

| LR | Num LABS |
|---|---|
| Not reported | 25 |
| Frequency | 1 |
| 678 | 1 |
| 473 | 1 |
| **361-325** | **1** |
| **354** | **2** |
| 345 | 1 |
| **326** | **1** |
| 315 | 3 |
| 296 | 1 |
| 190 | 1 |
| TOTAL | 38 |

- The estimated frequency of the M2 and M3 haplotypes is 2,1164E-03 (EMPOP, counting method).

1

LACK OF CONSENSUS

▸ Part II: evaluation

## Answers II:

LR → M2-M3 → mtDNA

RESULTS OF QUERIES

### EMPOP R-6 (until 24-4-12)

Samples 2 y 3 (without polymorphism in 152) 8/2834

Freq. = 2,383e-3
MI = 354,25

### EMPOP R-7 (from 25-4-12)

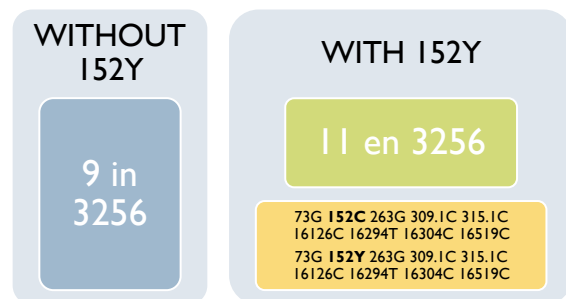Samples 2 y 3 (without polymorphisms in 152) 9/3256

Freq. = 2,764e-3
MI = 361,8

▸ Part II: evaluation

## Answers II:

LR ➡ M2-M3 ➡ mtDNA

▸ Do not report: 25 labs
  ▸ Without comments: 23
  ▸ Say «matching haplotypes, no exclusion»: 1
  ▸ «same result incluyending 152 heteroplasmy and considering T in that position»: not true

| WITHOUT 152Y | WITH 152Y |
|---|---|
| 9 in 3256 | 11 en 3256 |
| | 73G **152C** 263G 309.1C 315.1C 16126C 16294T 16304C 16519C |
| | 73G **152Y** 263G 309.1C 315.1C 16126C 16294T 16304C 16519C |

▸ Part II: evaluation

## Answers II:

M Ind. ➡ M1-M2 ➡ aSTR

| LAB | M Index |
|---|---|
| 20216 | 103,86 |
| 20223 | 103,86 |
| 20259 | 561,298 |
| 20274 | 104 |
| 20278 | 103,86 |

**CONSENSUS DATA: 103,86 - 104**

| LAB | M Index |
|---|---|
| 20224 | «Both samples were analysed in order to determine that M1 is the biological mother of the missing son» |
| 20241 | «The sample from the personal effect was used as a confirmative test of the sample from the body, , but it was not used to determine the identification, as it is an unknown sample» |
| 20244 | Does not calculate but state the hypothesis |
| 20254 | «The profile from the body is identical to the one from the personal effect in 9 STR, Amelogenine marker and mtDNA» |

▸ Part II: evaluation

## Answers II: combining LRs

▸ Maternity Index M1/M3 (aSTRs and mtDNA)

| 20177 | 20209 | 20254 | 20272 |
|---|---|---|---|
| 2,4653 e+07 | 4,4074e+07 | 4,072e+08 | 6,3323e+11 |

▸ LR M2 / M3 (aSTRs and mtDNA)

| 20177 | 20257 |
|---|---|
| 4,1869e+11 | 1,4878e+12 |

▸ Part II: evaluation

## Answers II: combining LRs

▸ LR M1 / M2 / M3

| 20221 (aSTRs) LR (M2-M3)*MIndex(M1-M3)? | 20240 $LR_{aSTR}$(M1-M2)*$MIndex_{aSTR}$(M2-M3)*$LR_{mtDNA}$ (M2-M3) |
|---|---|
| 3,15E+12 | 5,12E+15 |

▸ Part II: evaluation

## Answers II: errors

▸ Wrong definition of LR / MIndex
  ▸ LR = H1 / H2
  ▸ MI = H1 / H2

| 20241 | 20245 | 20257 | 20281 |
|---|---|---|---|

▸ Wrong interpretation of M Index: XXX times more probable it is the mother to is not the mother

| 20166 | 20178 | 20252 | 20257 | 20268 | 20254 |
|---|---|---|---|---|---|

▸ Wrong interpretation of LR: XXX times more probable effect and body are the same individual

| 20170 | 20176 | 20178 | 20186 | 20257 | 20268 |
|---|---|---|---|---|---|

▸ Part II: evaluation

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## Suggestions from labs

▸ 20241: In item 5.2.1 it should be a third box for writting why one or both samples are used.

▸ 20254: Include a table for reporting partial LRs of each marker, as it was made in the case of other theoretical forensic exercises.

▸

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## Conclusions

▸ Only a few labs calculate the Maternity Index of M1 (mother) and M2 (personal effect)

▸ Many labs do not report M Index/LR fro mtDNA data
  ▸ What they do when only mtDNA results are available?

▸ Still many errors in the expression of the results

▸ Only a few labs combine aSTR and mtDNA LRs

▸

## Conclusions

▸ Forensic paper challenge 2011 was published:
  ▸ GHEP-ISFG proficiency test 2011: Paper challenge on evaluation of mitochondrial DNA results. Prieto L, Alves C, Zimmermann B, Tagliabracci A, Prieto V, Montesino M, Whittle MR, Anjos MJ, Cardoso S, Heinrichs B, Hernandez A, LÃ³pez-Parra AM, Sala A, Saragoni VG, Burgos G, Marino M, Paredes M, Mora-Torres CA, Angulo R, Chemale G, Vullo C, SÃ¡nchez-SimÃ³n M, Comas D, Puente J, LÃ³pez-CubrÃa CM, Modesti N, Aler M, Merigioli S, Betancor E, Pedrosa S, Plaza G, Masciovecchio MV, Schneider PM, Parson W. Forensic Sci Int Genet. 2012 May 19. [Epub ahead of print] PMID: 22613778 [PubMed - as supplied by publisher]

▸ Usefulnes of the current exercise:
  ▸ Publication? Too much authors?
  ▸ Report for each lab with an opinion about their results?

▸

## Forensic paper challenge, advanced level GHEP 2012

- ▶ Unknowledgments
  - ▶ Josefina Gómez
  - ▶ Victoria Lareu
  - ▶ Antonio Salas
  - ▶ CGPC



> **MANU THANKS FOR YOUR ATENTION AND PARTICIPATION**



▶