Ayudas para asistencia a las Jornadas del GHEP-ISFG

Communications

Communications

i. STR mutations modelling

O12: "How often have X- and aut-STRs mutations equivocal parental origin assigned?"
Sofia Antão-Sousa, Eduardo Conde-Sousa, Leonor Gusmão, António Amorim, **Nádia Pinto**

P100: "How frequently are aut- and X-STRs multistep mutations perceived as single-step?"
Sofia Antão-Sousa, Eduardo Conde-Sousa, Leonor Gusmão, António Amorim, Nádia Pinto

Communications

ii. Weighing genetic relatedness considering X-chromosome aneuploidies

P.95: "Novel mathematical framework for genetic relatedness analysis involving X- chromosome aneuploidies"

Marisa Faustino, Chiara Turchi, Daniel Kling, Leonor Gusmão, Antonio Amorim, Nádia Pinto

Communications

iii. Quantification of forensic genetic evidence

O32: "Statistical analysis tools of mixture DNA samples: When the same software provide different results"

Camila Costa, Carolina Figueiredo, António Amorim, Lourdes Prieto, Sandra Costa, Paulo Miguel Ferreira, Nádia Pinto

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29th Congress of the International Society for Forensic Genetics

Statistical Analysis Tools Of Mixture DNA Samples: When The Same Software Provides Different Results

Camila Costa, Carolina Figueiredo, António Amorim, Lourdes Prieto, Sandra Costa, Paulo Miguel Ferreira, Nádia Pinto

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REPÚBLICA PORTUGUESA | CIÊNCIA, TECNOLOGIA E ENSINO SUPERIOR

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FCT Fundação para a Ciência e a Tecnologia

PhD Studentship reference:2021.05655.BD

INTRODUCTION | Quantifying the Genetic Evidence

Statistical models

Qualitative

- Qualitative information

Semi-continuous

LRmix Studio; Lab Retriever

Quantitative

- Qualitative information
- Quantitative information

Continuous

EuroForMix; STRmix

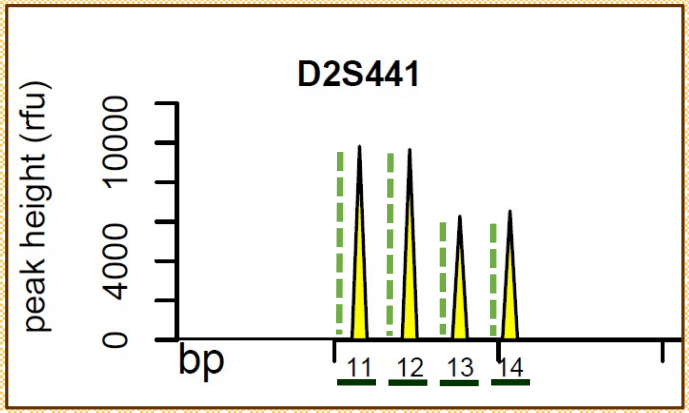

Figure 1: Representation of a two contributors' mixture for the marker D2S411.

SHORT COMMUNICATION |

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Short communication

Quantification of forensic genetic evidence: Comparison of results obtained by qualitative and quantitative software for real casework samples

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SHORT COMMUNICATION |Aims + Material and Methods

LRmix Studio v.2.1.3

EuroForMix v.3.4.0

STRmix™ v.2.7

Qualitative

Quantitative

156 anonymized mixture/single contributor sample pairs from Portuguese *Laboratório de Polícia Científica - Polícia Judiciária* (LPC-PJ) former cases

78 with mixtures with 2 estimated contributors

(21 markers analyzed)

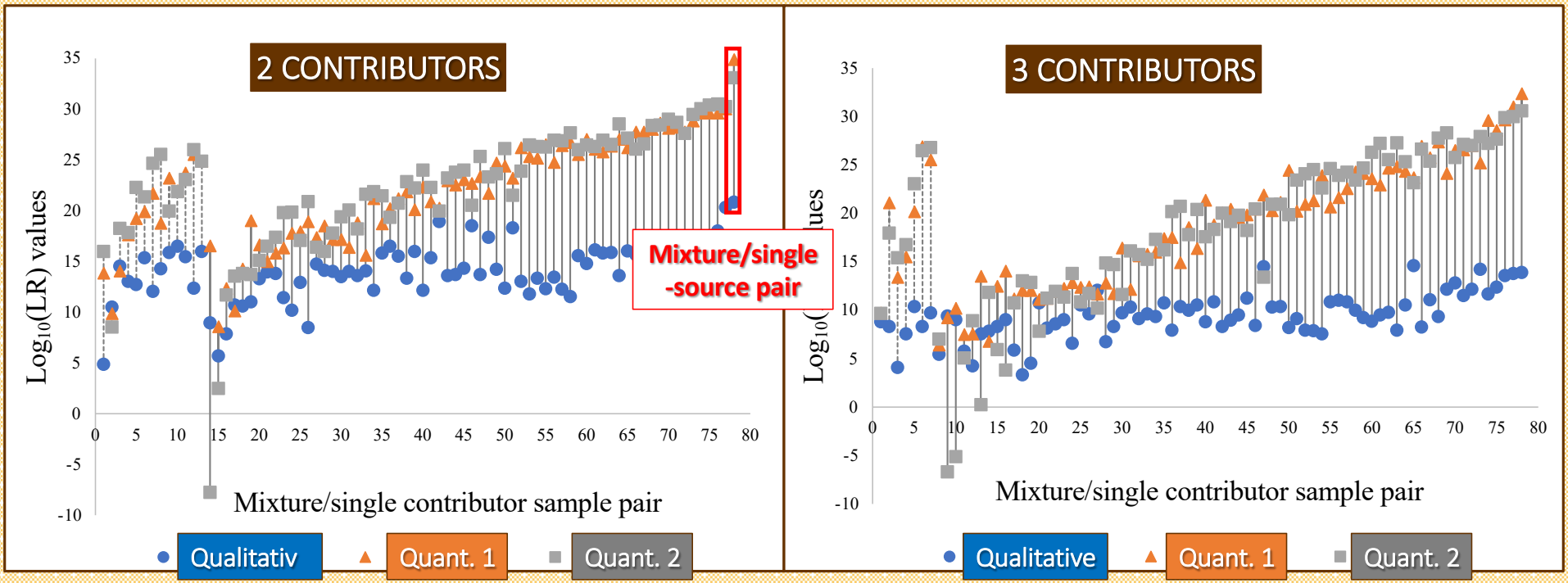
78 with mixtures with 3 estimated contributors

SHORT COMMUNICATION |Aims + Material and Methods

Parameters	Values		
	Qualitative	Quantitative 1	Quantitative 2
Number of contributors	Estimated	Estimated	Estimated
Co-ancestry Coefficient	0.010	0.010	0.010
Drop-in frequency	0.05	0.05	0.05
Drop-in parameters' distribution	N/A	λ: 0.01	Uniform
Drop-in cap	N/A	N/A	100
Dropout	0.1	[a]	[a]
Minimum allele frequency	0.001	0.001	[b]
Threshold detection	N/A	100	100
Stutters	No	Yes	Yes

[a] Dropout is directly estimated through the peak height distribution.
[b] Per locus specified by the software considering N=0
N/A: Not Applicable

SHORT COMMUNICATION |Results and Discussion



SHORT COMMUNICATION |Results and Discussion

$$D = ABS\,[Log_{10}(LR1) - Log_{10}(LR2)]$$

EXAMPLE (pair 78_2):	LR	$Log_{10}(LR)$	$Log_{10}(LR1) - Log_{10}(LR2)$	D
● QUALITATIVE	6.79E20	20.832	● - ▲ = -14.074	| ● - ▲ | = 14.074
▲ QUANTITATIVE 1	8.04E34	34.905	● - ■ = -12.245	| ● - ■ | = 12.245
■ QUANTITATIVE 2	1.19E33	33.077	■ - ▲ = -1.828	| ■ - ▲ | = 1.828

ABS → (Absolute value)

SHORT COMMUNICATION | Results and Discussion

$D = ABS [Log_{10}(LR1) - Log_{10}(LR2)]$	Estimated Number of Contributors					
	Two			Three		
	Qual. Quant. 1	Qual. Quant. 2	Quant. Quant. 2	Qual. Quant. 1	Qual. Quant. 2	Quant. 1 Quant. 2
0 < D < 2	8%	6%	73%	13%	9%	59%
2 < D < 4	13%	15%	19%	8%	8%	24%
4 < D < 6	19%	13%	4%	8%	9%	9%
6 < D < 8	5%	6%	3%	14%	10%	1%
8 < D < 10	18%	13%	0%	12%	10%	1%
D > 10	37%	46%	1%	46%	54%	5%
MAXIMUM DIFFERENCE (log_{10} units)	15.206	16.682	24.288	18.707	19.341	15.970

SHORT COMMUNICATION | Results and Discussion

Properties	QUANTITATIVE 1	QUANTITATIVE 2
Drop-in distribution	Exponential	Gamma or Uniform
Peak height distribution	Gamma	Log-normal
Inference approach	Bayesian or Maximum Likelihood Estimation (MLE)	Bayesian

SHORT COMMUNICATION | Results and Discussion

$D = ABS [Log_{10}(LR1) - Log_{10}(LR2)]$	Estimated Number of Contributors					
	Two			Three		
	Qual. Quant. 1	Qual. Quant. 2	Quant. Quant.	Qual. Quant. 1	Qual. Quant. 2	Quant. 1 Quant. 2
0 < D < 2	8%	6%	73%	13%	9%	59%
2 < D < 4	13%	15%	19%	8%	8%	24%
4 < D < 6	19%	13%	4%	8%	9%	9%
6 < D < 8	5%	6%	3%	14%	10%	1%
8 < D < 10	18%	13%	0%	12%	10%	1%
D > 10	37%	46%	1%	46%	54%	5%
MAXIMUM DIFFERENCE (log_{10} units)	15.206	16.682	24.288	18.707	19.341	15.970

SHORT COMMUNICATION |Conclusions

- Software based on **different models** (Qualitative vs. Quantitative)
  Largest discrepancies between computed LR values
- Software based on the **same model** (Quantitative)
  ≠ mathematical, statistical and informatics models ≠ results
- Use of **more than one informatic tool** in forensic routine
  Confrontations between possibly different results
  Identify difficult cases

SAME SOFTWARE, DIFFERENT RESULTS | Software parameters

- Number of Contributors
- Drop-in
- Co-ancestry Coefficient
- Drop-out
- Threshold Detection
- Stutters

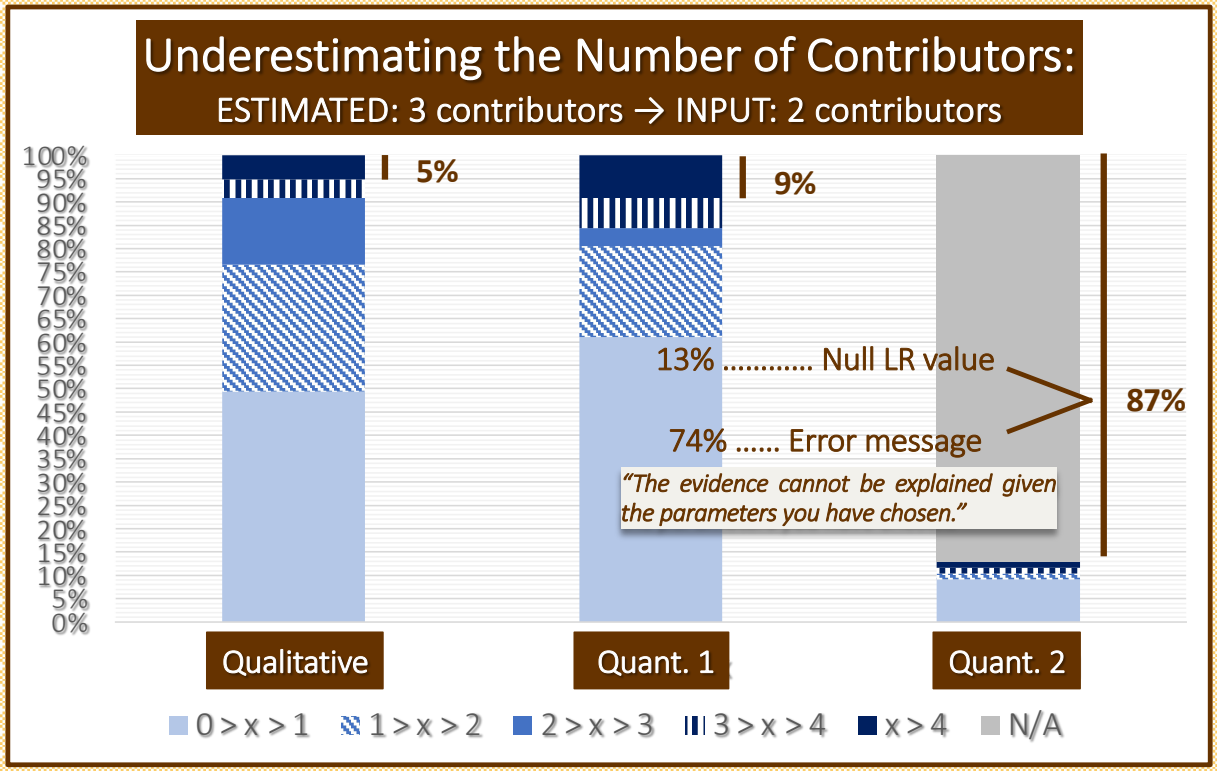
- Software based on the same model provides different results due to different mathematical computations

- Software parameters values must be established by the expert and enter on the software prior to any computation

Values / Parameters	LRmix Studio		EuroForMix		STRmix™	
	Default	Variation	Default	Variation	Default	Variation
Number of contributors (NOC)	Estimated	Estimated +1 Estimated -1	Estimated	Estimated +1 Estimated -1	Estimated	Estimated +1 Estimated -1
Co-ancestry Coefficient (F_{ST})	0.010	0.000 0.015 0.030	0.010	0.000 0.015 0.030	0.010	0.000 0.015 0.030
Drop-in frequency	0.05	0.00 0.10	0.05	0.00 0.10	0.05	0.00 0.10
Drop-in parameters' distribution	-	-	λ: 0.01	λ: 0.05	ɣ: (0.1,0.1)	Uniform
Threshold detection	-	-	100	150	100	150
Stutter	-	-	Yes.	No.	-	-

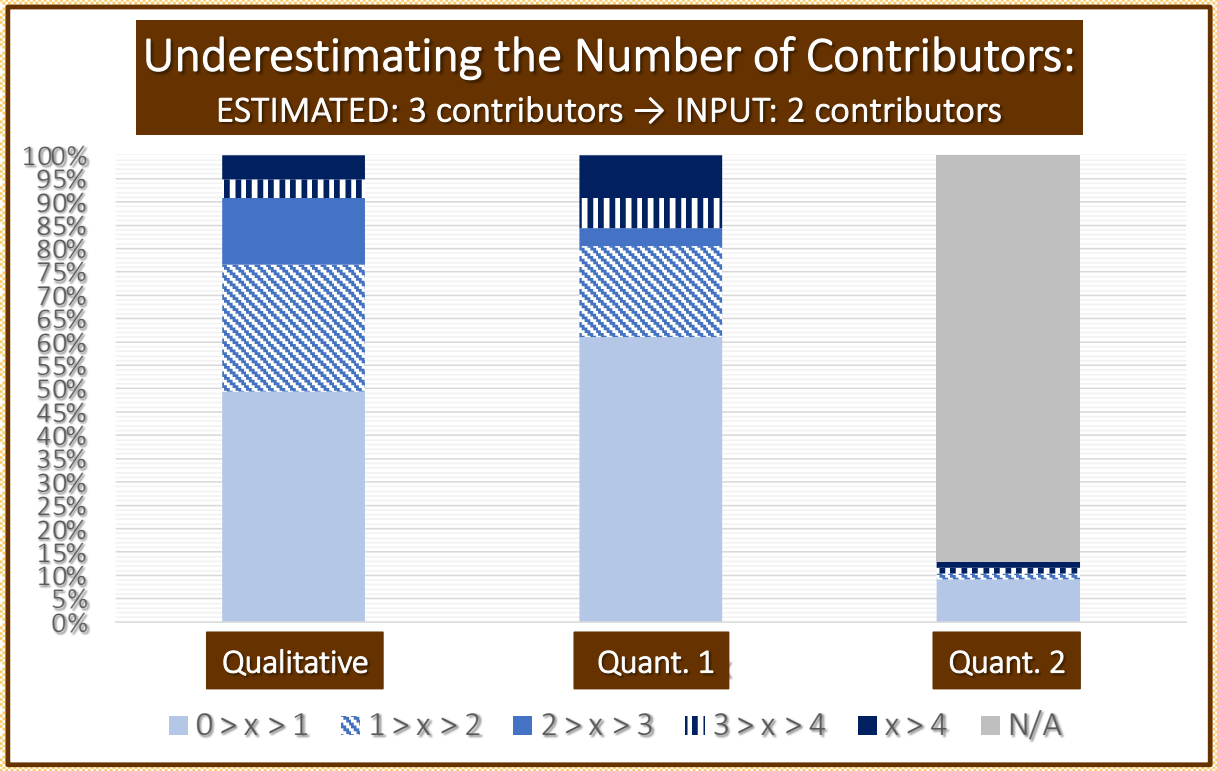
SAME SOFTWARE, DIFFERENT RESULTS | Software parameters

Number of Contributors



SAME SOFTWARE, DIFFERENT RESULTS | Software parameters

- Number of Contributors
- Drop-in
- Co-ancestry Coefficient
- Drop-out
- Threshold Detection
- Stutters



SAME SOFTWARE, DIFFERENT RESULTS | Different software versions

EXAMPLES:

	Quantitative 1 version 2016 Models Back stutter	Quantitative 1 version 2021 Models Forward stutter and Back stutter
2 CONTRIBUTORS (pair 24_2)	LR = 4.61E+15 Loglik = -670.3 P.H. variability = 0.258	LR = 6.28E+17 Loglik = -654.9 P.H. variability = 0.199
3 CONTRIBUTORS (pair 42_3)	LR = 5.62E+15 Loglik = -982.4 P.H. variability = 0.283	LR = 1.15E+20 Loglik = -944.7 P.H. variability = 0.175

The model is a GOOD FIT

↑ Loglik
↓ P.H. variability

FINAL REMARKS |

- Different software compute different LR results, regardless of whether the informatics tools consider the quantitative information of the electropherogram or not.
- Use of more than one informatic tool in forensic routine to confront obtained results and identify difficult cases.
- Importance of a correct software parameter estimation, so that the computation of the LR value is as accurate as possible.
- Awareness of updates made to software in use.

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