GHEP-ISFG Collaborative Exercise on Mixture Profiles of Y-Chromosome STRs (GHEP-MIX05_HAP2): Results and evaluation (mock case #2).

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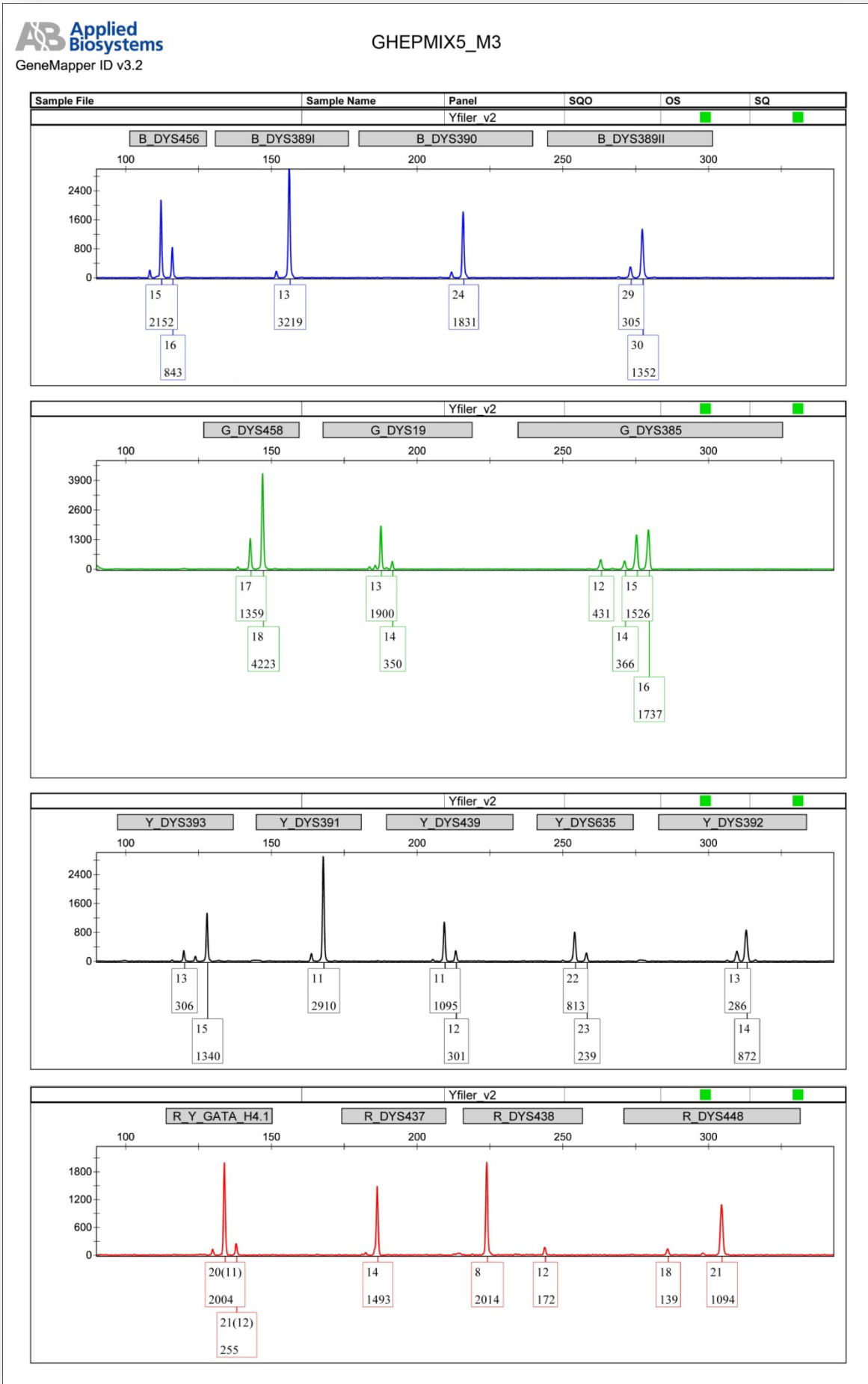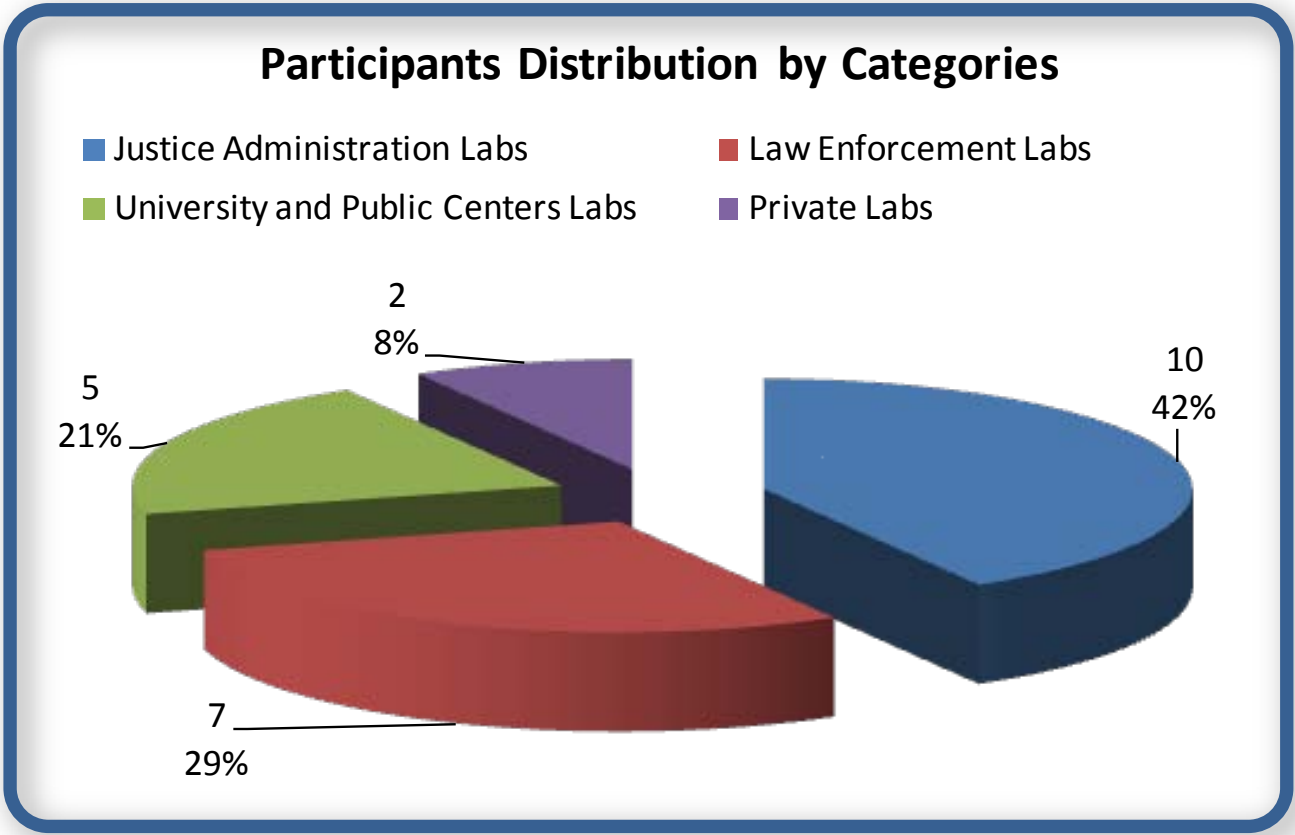on behalf of GHEP-MIX working commission

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SPANISH AND PORTUGUESE-SPEAKING WORKING GROUP OF ISFG

Introduction

One of the main goals of the Spanish and Portuguese-Speaking Group of the International Society for Forensic Genetics (**GHEP-ISFG**) is to promote and contribute to the development and dissemination of scientific knowledge in the field of forensic genetics. Due to this fact, GHEP-ISFG holds different working commissions that are set up to develop activities in scientific aspects of general interest. One of them, the Mixture Commission of GHEP-ISFG, has organized annually, since 2009, a collaborative exercise on analysis and interpretation of autosomal short tandem repeat (STR) mixture profiles. Until now, six exercises have been organized.
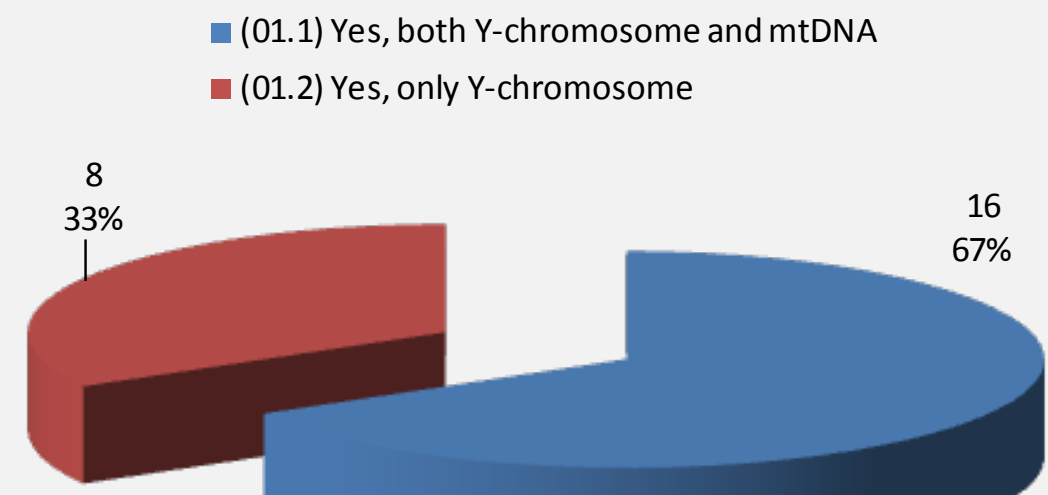
In the 2015 edition (**GHEP-MIX05**), with 24 participant laboratories from 7 different countries (Spain, Chile, Argentina, Dominican Republic, Colombia, Italy, Czech Republic), one of the exercise aims was to give a general vision about mixed profiles of Y-chromosome STRs analysis. Through the proposal of mock cases, edition and statistical treatment were assessed. Additionally, the exercise included a questionnaire where various issues were asked that dealt with the characteristics of the laboratory, as well as the technical criteria used in the interpretation of haploid profiles and statistical estimation. With this poster we will show the results obtained from the statistical evaluation for one of the mock cases (**mock case #2**).
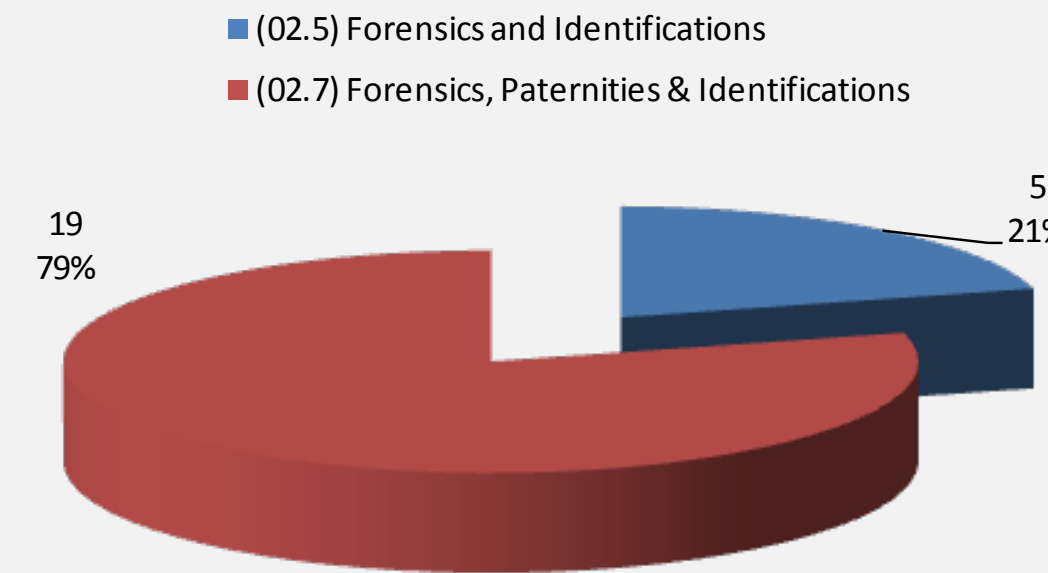
Participants Distribution by Categories

- Justice Administration Labs
- Law Enforcement Labs
- University and Public Centers Labs
- Private Labs

Labs Characterization

1. Does your laboratory usually analyze haploid markers?

- (01.1) Yes, both Y-chromosome and mtDNA
- (01.2) Yes, only Y-chromosome

2. The analysis of haploid markers, your laboratory uses it in cases:

- (02.5) Forensics and Identifications
- (02.7) Forensics, Paternities & Identifications

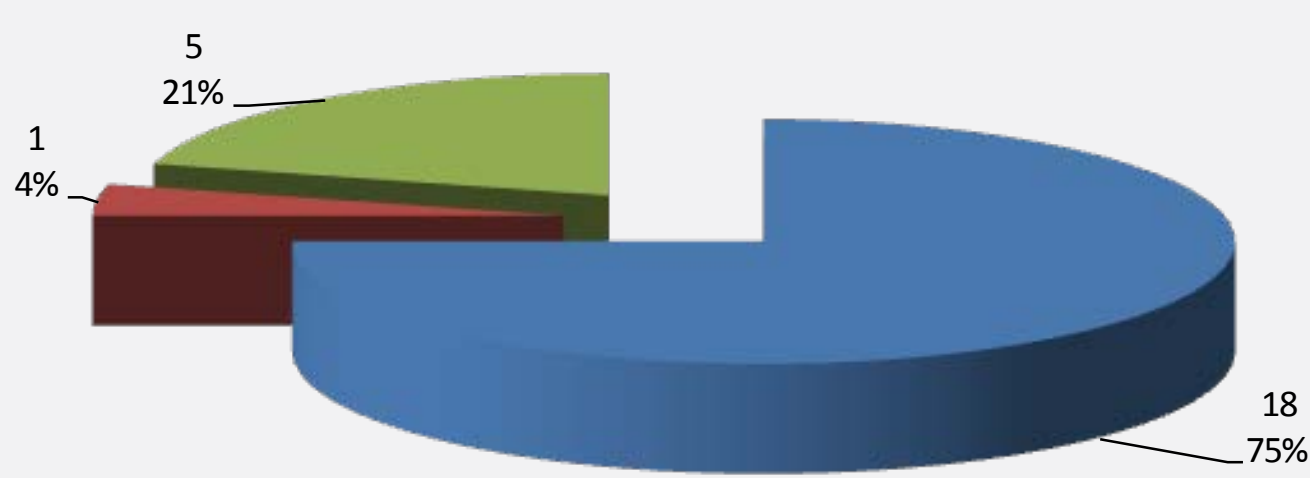The participants had to indicate if, in daily casework, they emits results for:

7. ... haploid markers:

- (07.1) Yes, both for Y-chromosome and mtDNA markers
- (07.2) Yes, both for Y-chromosome and mtDNA markers, only if reference samples are available
- (07.3) Yes, only for Y-chromosome
- (07.7) No
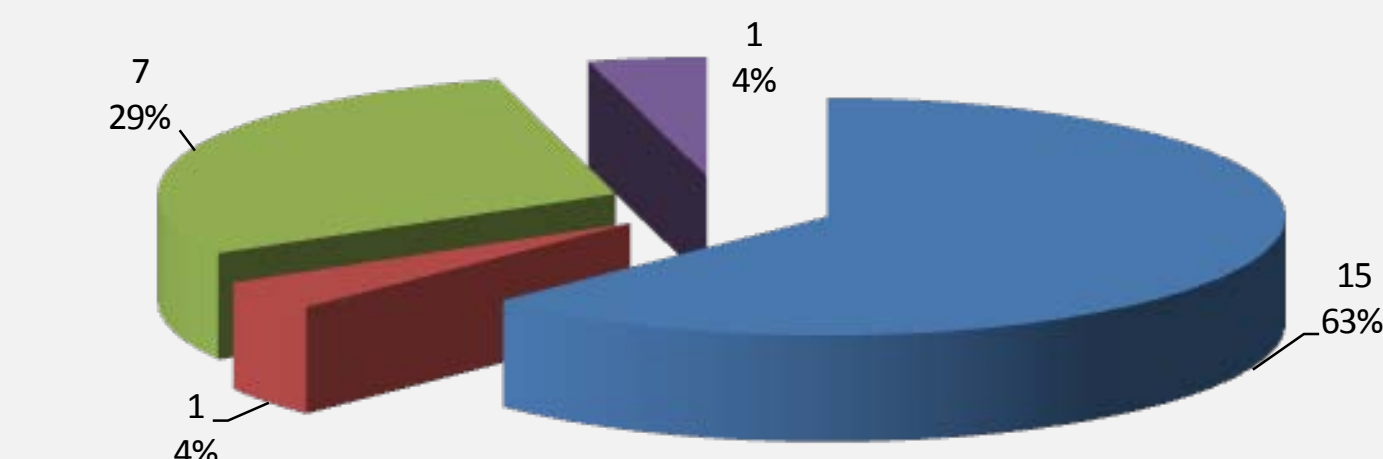
8. ... mixed profiles of haploid markers of Y-chromosome:

- (08.1) Yes
- (08.2) Only in case of having reference samples
- (08.3) No

Applied Biosystems GeneMapper ID v3.2 — GHEPMIX5_M3

	Person 1			Person 2
DYS456	15		DYS456	16
DYS389I	13		DYS389I	13
DYS390	24		DYS390	24
DYS389II	30		DYS389II	29
DYS458	18		DYS458	17
DYS19	13		DYS19	14
DYS385	15-16		DYS385	12-14
DYS393	15		DYS393	13
DYS391	11		DYS391	11
DYS439	11		DYS439	12
DYS635	22		DYS635	23
DYS392	14		DYS392	13
GATAHA.1 (H4)	20(11)		GATAHA.1 (H4)	21(12)
DYS437	14		DYS437	14
DYS438	8		DYS438	12
DYS448	21		DYS448	18

Background of mock case #2:

In a case of alleged sexual assault, the victim claimed that she was raped with vaginal penetration by 1 assailant, without condom. She also stated that a couple of hours before, she had had sexual relations with her regular partner, also without condom. During the medical examination, the coroner made a vaginal swab to the victim, which was sent to the forensic lab along with a reference sample of the victim and her regular partner (labeled as "Person 2"), for its exclusive use as a discard.

After some time, the police arrested a suspect, and after informed consent, a reference sample was taken (labeled as "Person 1"), which was also sent to the lab.

Analytical background of mock case #2:

After the analysis of the vaginal swab, a small amount of sperm was visualized under the microscope. Differential lysis was performed and, after quantification of the sample, it was detected that the disproportion between total DNA and male DNA was about 1:30. Autosomal genetic markers were analyzed, revealing the impossibility of detecting autosomal DNA of masculine origin. When not being able to separate male DNA from female DNA, the decision was made to analyze Y-chromosome markers. After this genetic analysis, the haploid profile of the EPG (**GHEPMIX5_M3**) was obtained, at final point and without the possibility of carrying out additional analysis.

Likewise, the reference haploid genetic profiles of the two presumed males involved in the case were obtained: the victim's regular partner ("Person 2"), and the suspect arrested ("Person 1"). These haploid genetic profiles were listed in the left tables.

Mock case #2 Hypotheses

- No LR performed
- (P1/U)
- (P1/U) + (P1+P2/P2+U)
- (P1+P2/P2+U)
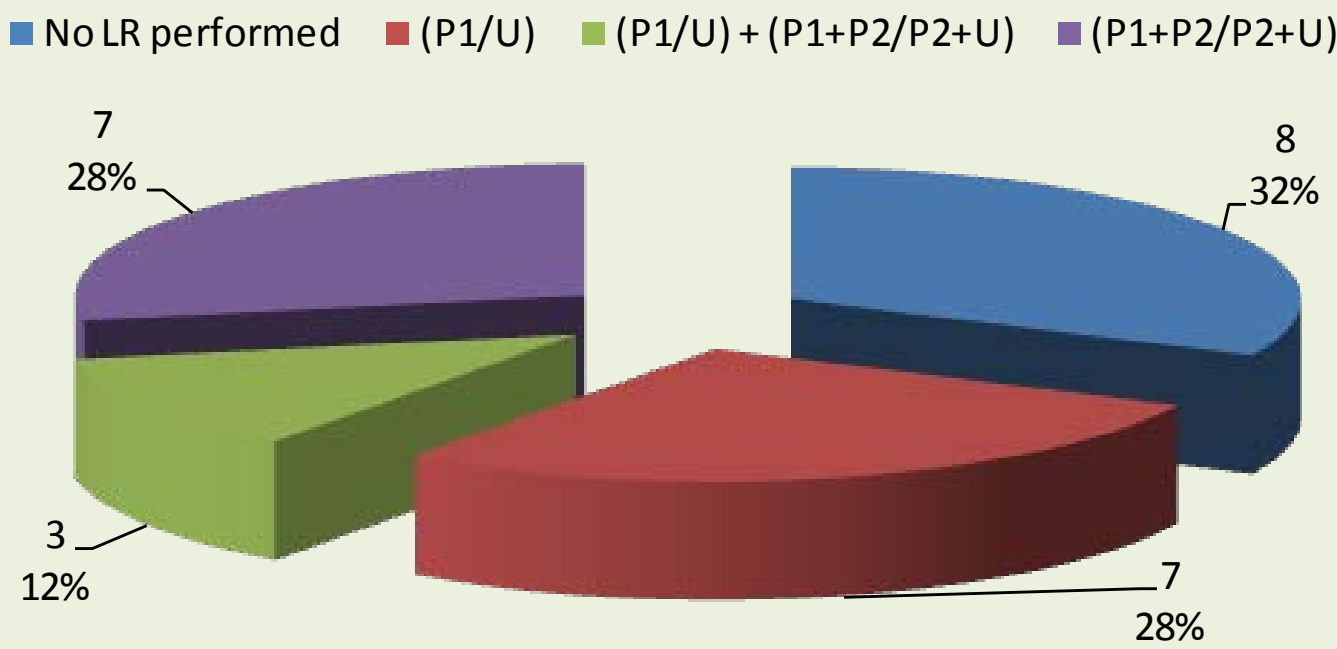
Hypotheses	LR	n	%
(S1/U)	6.8800E+02	1	10.0%
	1.5798E+03	1	10.0%
	6.2570E+03	1	10.0%
	1.1317E+04	1	10.0%
	1.8388E+04	2	20.0%
	2.2633E+04	1	10.0%
	8.4257E+04	1	10.0%
	9.1232E+04	1	10.0%
	2.5077E+05	1	10.0%
(S1+S2/S2+U)	1.0137E+04	5	50.0%
	1.2037E+04	1	10.0%
	2.8126E+04	1	10.0%
	4.5620E+04	1	10.0%
	9.1743E+04	1	10.0%
	2.5059E+05	1	10.0%

Conclusions

All the GHEP-ISFG labs that participated in the **GHEP-MIX5_HAP** exercise use haploid markers (Y-chromosome and/or mtDNA) in their casework. From these, only one lab does not emit results for this type of markers. Nevertheless, five labs do not emit results for mixed profiles of haploid markers of Y-chromosome, although 60% of the participants did not perform the statistical evaluation in these cases. It is possible, although these laboratories indicate in their reports that they have obtained a mixed profile for Y-chromosome markers, due to their complexity, they prefer not to do their statistical evaluation.

Notwithstanding the above, against the mock case #2 proposed in this exercise, up to 68% of the participants made a statistical assessment through LR calculation, although setting different hypotheses. The differences in the LR value detected within each pair of hypotheses could be explained as a consequence of the reference metapopulation used, as well as the frequency source when not observed in the YHRD database.

Taking these data into account, the main conclusions obtained from this exercise module may be summarized in the need for continuing education in the analysis and interpretation of mixture DNA profiles, in particular, in the case of Y-chromosome mixture profiles.

Acknowledgements