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Research article

GEP-ISFG proficiency testing programs: 2007 update

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Abstract

This year the annual GEP-ISFG proficiency test consisted of two practical and three theoretical exercises. The practical exercise included a deficiency maternity test for which blood samples from a woman and two alleged children were analyzed and a forensic case that included a cigarette butt for STR typing and two hair shafts sent specifically for mitochondrial DNA analysis, both to be compared with a reference blood sample from a suspect. In the theoretical approach of the exercise, three different proposals were offered (one included in the certificate of participation, and two optional); the most relevant was a paper challenge on mixtures prepared with the idea to review some interesting aspects of the recent ISFG recommendations on mixtures interpretation and specially the likelihood ratio method calculation under both the unrestricted and the restricted combinational approaches. A total of 122 laboratories belonging to 16 different countries received the samples, from which, 109 submitted results. Around 50% of participating labs performed both the paternity and the forensic trials, while 52 laboratories performed only the paternity test. Other working groups of the GEP-ISFG have also organized other collaborative exercises during this year. These included a collaborative exercise with a battery of STR from X chromosome to improve standardization and the first collaborative exercise on non-human (dog) mtDNA sequencing coordinated by the Sexual Chromosomes Working Group and the Non-Human Forensic Genetics Working Group of the GEP-ISFG, respectively.
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1. Introduction

For more than 10 years, the GEP-ISFG has been offering an annual exercise involving genetic and also statistical analysis of paternity and forensic cases. The 2007 GEP-ISFG proficiency testing program consisted of two practical and three theoretical tests. In this paper we summarize the results submitted this year for the practical test with a brief analysis of the discordances and causes of error observed. A special consideration to the theoretical exercises and other collaborative tests organized by the GEP-ISFG is also commented.

2. Materials and methods

2007 practical exercise consisted in six samples: three blood reference samples for a maternity test (M1 mother, M2 and M3 alleged sons) and for the forensic case a cigarette whose butt was covered with 50 μl of saliva (from an unrelated child) and the investigation of two hair shafts (M6, from M4 donor) with a blood reference sample M4.

All labs were asked to report the methods used including forensic preliminary analysis, the typing results of STRs (autosomal, X- and Y-STRs) and mitochondrial DNA (mtDNA) as well as to interpret results including statistical evaluation. Electropherograms and analytical results were required to obtain the participation certificate.

3. Results and discussion

STR loci included in GEP-ISFG 2007 is listed at the table below, where only consensus markers are listed. Consensus for

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Table 1
Details of total number of determinations for STR profiling and discordance rate (%) observed depending on the detection procedure (manual/automated)

	Manual				Automated		
	System	*n*	Discordance	(%) discordance	*n*	Discordance	(%) discordance
2007 maternity exercise: participating labs: 108, number of samples: 3, total determinations: 7975	Autosomal	741	24	3.24	4659	29	0.62
	Y-STRs	184	8	4.43	1503	7	0.47
	Others	36	0	0	852	3	0.35
	Total	961	32	3.32	7014	39	0.56

a specific marker is reached when at least 70% of a minimum of five labs reports the same result. In this exercise a significant increased participation at X-STR markers was observed. More than 50 other markers not included in the table are reported by a lower number of laboratories.

The forensic sample was analyzed by 54 laboratories; only 16 of them described preliminary tests. The sample contained saliva from a unique donor (different from the suspect, M4) from which reference sample was not sent. All labs discarded M4 donor as its contributor. It has been shown a correlation

Autosomal STRs			Y-STRs		X-STRs
FES/FPS	FGA	Penta D	DYS19	DYS456	DXS8378
F13A01	D13S317	Penta E	DYS385	DYS458	DXS9898
F13B	D16S539		DYS389 I	DYS460	DXS7133
LPL	D18S51		DYS389 II	DYS461	GATA31E08
ACTBP2(SE33)	D19S433		DYS390	GATA A10	GATA172D05
D1S1656	D21S11		DYS391	DYS635	DXS7423
D12S391	D2S1338		DYS392	GATA H4.1	DXS6809
CSF1PO	D3S1358		DYS393	DYS439	DXS7132
TH01	D5S818		DYS437		DXS9902
TPOX	D7S820		DYS438		DXS6789
VWA	D8S1179		DYS448		HPRTB
Gender determination			AMELOGENIN		

A relevant number of discordances were detected in the exercise (see Tables 1 and 2). 21 out of 108 participating labs showed discordances in the maternity test exercise. A similar proportion of labs reported discordances in the forensic exercise (9 out of 54). Discordances are concentrated at a few laboratories while more than 10 laboratories presented only a single discordance. One of the causes of errors is attributable to methodological aspects. Maternity exercise results in Table 1 show that laboratories performing manual detection techniques presented a higher error rate.

between discordances and labs with deficient technical procedures (absence of preliminary analysis, absence of human specific quantification techniques, and incorrect procedures for allelic assignment). Three laboratories had reported 70% of the discordances observed in this sample due to these deficient procedures. An example of this is displayed in Table 2, where those labs performing human DNA quantification showed lower discordance rate when compared with those labs that do not use human specific DNA quantification (0.098% vs. 3.07%).

Table 2
Details of total number of determinations for forensic sample STR profiling and discordance rate (%) observed related to the quantification procedure (human specific/no quantification, non-human specific)

	Human specific				Non-human specific		
	System	*n*	Discordance	(%) discordance	*n*	Discordance	(%) discordance
2007 forensic exercise: participating labs: 54; number of samples: 2; total determinations: 3489	Autosomal	597	1	0.17	1254	56	4.47
	Y-STR	481	0	0	853	14	1.64
	Others	136	0	0	173	0	0
	Total	1214	1	0.098	2280	70	3.07

Concerning mtDNA hair analysis, 2007 results showed an increased participation in these analyses. The consensus haplotype (73G, 146C, 153G, 189G, 235G, 263G, 315.1C, 16051G, 16223T, 16290T, 16293G, 16319A, 16362C) was reported by 27 out of 32 of the labs and discordances observed were mainly due to nomenclature or clerical errors. Additionally, two labs reported A/G at position 215, while two others reported G at the same position and one lab reported a C in position 199.

The different theoretical tests proposed in 2007 exercise generated the largest discussion. 107 labs sent results to the paternity test included in the certificate. 58 labs participated in a fatherless case in relation to a putative daughter having only available the STR profiles from the mother and two brothers. Results dispersion makes difficult to establish a consensus IP value and evidenced different statistical procedures.

Laboratories were also encouraged to participate in a mixture interpretation challenge planned with the intention to follow the ISFG recommendation on mixture interpretation paper recently published [1]. 23 out of 31 participating obtained a consensus response for the mixture proposed under the unrestricted combinatorial approach but when the restricted combinatorial approach were applied only 13 laboratories participated, showing uniform results for most markers analyzed but data dispersion in some of them.

Other collaborative exercises have been organized during 2007 by the GEP-ISFG working groups: The Non-Human Forensic Genetics Working Group sent a dog blood sample to 13 participating labs that were asked to type the sample for mt D-loop region (15372–16083). Guidelines for PCR analysis and sequence edition were provided to labs on the website (http://www.gep-isfg.org) and their results will be reported soon. The Sexual Chromosomes Working Group prepared two blood samples and provided primer mix for 10 X-STRs: DXS8378, DXS9898, DXS7133, GATA31E08, GATA172D05, DXS7423, DXS6809, DXS7132, DXS9902, DXS6789. Primer sequence and PCR conditions were specified at the website. The results sent by 32 participating labs were summarized in a poster [2] at the 22nd ISFG Congress.

Conflict of interest

None.

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