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A report of the 2002–2008 paternity testing workshops of the English speaking working group of the International Society for Forensic Genetics

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ABSTRACT

The English Speaking Working Group (ESWG) of the International Society for Forensic Genetics (ISFG) offers an annual Paternity Testing Workshop open to all members of the group. Blood samples, a questionnaire and a paper challenge are sent to the participants. Here, we present the results of the 2002–2008 Paternity Testing Workshops with the objective to evaluate the uniformity of DNA-profiling and conclusions of the participating laboratories as well as to clarify tendencies in typing strategies and biostatistical evaluations of the laboratories. The numbers of participating laboratories increased from 46 in 2002 to 68 in 2008. The results showed an increasing degree of concordance concerning methods and DNA systems used and a high degree of uniformity in typing results with discrepancies in 0.1 and 0.3 % of all submitted PCR-based results. The paper challenges showed uniformity in the calculation of the weight of evidence for simple cases with straight-forward genetic constellations. However, a high degree of variation existed in complex scenarios with rare genetic constellations such as genetic inconsistencies/possible silent alleles, rare alleles and haplotypes.

1. Introduction

Since 1991, The English Speaking Working Group (ESWG) of the International Society for Forensic Genetics (ISFG) has organized an annual, collaborative workshop concerning genetic analysis in paternity testing [1–4]. The workshops are performed with the aim to enable inter-laboratory comparison, which is essential for modern, accredited laboratories. The workshop is divided into three parts. One part is the paternity testing exercise, in which blood samples from fictive paternity cases are distributed to the participating laboratories that are asked to perform genetic investigations according to their usual protocols. With the aim to compare laboratory strategies and biostatistical evaluations among the participants, the second part of the workshop is a questionnaire. The third part is a paper challenge concerning biostatistical calculations. As laboratories use different systems for typing as well as different frequency-databases for their calculations, comparisons of calculated likelihood ratios (LR) of the performed paternity tests are unattainable. Thus, from 2000, a paper challenge has been included in the workshop. This allows for comparison of the biostatistical calculations of both routine combinations and rare events such as genetic inconsistencies/possible silent alleles and haplotypes.

Here, we present the results of the 2002–2008 Paternity Testing Workshops of the ESWG. The report describes tendencies in methods and kits used for DNA-typing, information concerning strategies for biostatistic calculations of the weight of evidence and requirements for issuing a report with an excluded/non-excluded man. Also, concordances/discordances in phenotyping results are presented. Finally, the divergence in biostatistical calculations of the weight of evidence among the laboratories, highlighted by the paper challenges, is presented.

2. Material and methods

Blood samples for the paternity testing exercise were distributed to the participants along with paper challenges and questionnaires. The laboratories were asked to perform testing according to their usual strategies and methods. Until year 2004, the participants reported the results of the paternity tests in their report. From 2005, the participants have reported the results, the answers to the questionnaire and the paper challenge online. The participating laboratories are listed in Appendix A. The results were analysed and presented at the annual ESWG meetings (Appendix B).

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Table 1
Methods available for genetic investigations in paternity testing.

Methods	2002 (%) N^a = 46	2003 (%) N^a = 51	2004 (%) N^a = 55	2005 (%) N^a = 62	2006 (%) N^a = 64	2007 (%) N^a = 69	2008 (%) N^a = 68
Autosomal STR kits	91	100	100	98	100	100	99
Y-chromosomal STRs	20	39	64	71	78	86	81
kits only	–	18	45	61	67	78	77
X-chromosomal STRs	–	8	15	10	20	26	35
kits only	–	–	–	4	13	19	32
VNTR-systems (RFLP)	43	25	29	18	16	13	12
mtDNA sequencing	7	16	36	31	34	30	29
Autosomal SNPs	–	–	9	9	5	7	7
Y-chromosomal SNPs	–	–	11	11	8	6	6
X-chromosomal SNPs	–	–	2	3	–	1	–
mtDNA SNPs	–	–	2	–	2	4	4

[a] Number of participating laboratories.

In 2002, blood samples were drawn from a child, the biological mother and two alleged fathers. In 2003 and 2004, blood samples were drawn from a child, the biological mother and an alleged father. In 2005, blood samples were drawn from two children, the biological mother and an alleged father, who was the brother of the biological father. In 2006, blood samples from two twins, their biological mother and an alleged father were provided. In 2007 and 2008, blood samples were drawn from a child, the biological mother and an alleged father.

In the paper challenges, all laboratories investigated the same hypothesis using the same phenotyping data and information (numbers of alleles in a database), and it was left to the laboratories to use this information according to their usual procedures.

3. Results

3.1. Accreditation

The number of participating laboratories increased from 46 in 2002 to 68 in 2008. In this time span, the percentage of laboratories with accreditation increased from 46% to 59%. The main accreditation standard was ISO17025 according to which 95% of the laboratories were accredited in 2008 compared to 71% in 2004. In 2006 and 2007, 12% and 11%, respectively, were accredited according to the ISO15189 standard. In 2008, only 5% were accredited according to this standard.

3.2. Available methods

Table 1 shows the methods available for genetic investigations in the participating laboratories. Since 2003, all laboratories have analysed STR-systems. From 2002 to 2008, the use of RFLP-based VNTR analysis decreased notably from 43% to 12%. The use of HLA typing decreased from 17% in 2002 to 2% in 2004 and is no longer used for paternity testing by the participants. The availability of mtDNA sequencing as an additional test has increased from 7% of the laboratories in 2002 to 36% in 2004 and has remained constant since then. In 2003, the first laboratories started to report results of X-STR systems increasing to 35% in 2008. Likewise, Y-STR analysis was only available in 20% of the laboratories in 2002 compared to 86% and 81% in 2007 and 2008, respectively. As seen in Table 1, the use of SNP analysis as an additional test was first reported in 2004, but its use has not increased since then.

There is an obvious tendency towards the use of commercial kits both for autosomal STR-systems and for Y- and X-chromosomal STR systems. In 2002, 91% of the participants used commercial kits. Since 2003, all participants have used commercial autosomal-STR kits, except for a single laboratory in 2005 and 2008. Table 2 shows the most frequently used kits. The two autosomal kits, PowerPlex 16 System (Promega) and Amp*Fl*–STR Identifiler (Applied Biosystems-AB), are the most frequently used kits. The use of SGM Plus and Profiler Plus (AB) has decreased from 67% and 36%, respectively, in 2002 to 35% and 10% in 2008. The use

Table 2
The most frequently used commercial STR-kits for paternity testing.

STR-kits	2002(%)	2003(%)	2004(%)	2005(%)	2006(%)	2007(%)	2008(%)
Autosomal kits	N^a = 42	N^a = 51	N^a = 55	N^a = 61	N^a = 64	N^a = 69	N^a = 67
PowerPlex 16 (Promega)	48	55	64	68	70	71	76
Identifiler (AB[b])	10	27	49	52	45	57	57
SGM Plus (AB)	67	47	45	47	44	43	35
FFFL (Promega)	14	22	24	24	28	28	25
SEfiler (AB)	–	–	13	13	16	20	21
Profiler (AB)	14	–	18	15	11	12	13
Profiler Plus (AB)	36	16	20	18	9	10	10
Power ES System (Promega)	2	–	9	10	13	13	10
Humantype Chimera (Biotype)	–	–	–	2	6	10	9
MiniFiler (AB)	–	–	–	–	–	–	10
Y-chromosomal kits		N^a = 9	N^a = 29	N^a = 39	N^a = 48	N^a = 54	N^a = 55
Powerplex Y (Promega)	–	–	96	99	75	59	62
Y-filer (AB)	–	–	–	13	38	53	56
DYSplexI /II(Serac)	–	44	14	13	4	–	–
X-chromosomal kits				N^a = 6	N^a = 13	N^a = 16	N^a = 22
Mentype ArgusX-UL (Biotype)	–	–	–	100	100	100	92
Mentype ArgusX-8 (Biotype)	–	–	–	–	–	–	8

[a] Number of laboratories.
[b] Applied Biosystems.