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# A report of the 2000 and 2001 Paternity Testing Workshops of the English Speaking Working Group of the International Society for Forensic Genetics

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### 11 Abstract

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13 During the last 10 years, the English Speaking Working Group (ESWG) of the International Society for Forensic Genetics (ISFG) has once a year arranged a Paternity Testing Workshop in which blood samples as well as a questionnaire concerning 14 15 laboratory strategies were distributed to the participating laboratories. In 2000 and 2001, paper challenges were included in the 16 workshops. Here, we present the results of the 2000 and 2001 Paternity Testing Workshops. The numbers of participating 17 laboratories were 33 (2000) and 36 (2001). A total of 36% (2000) and 31% (2001) of the laboratories submitted typing results of variable number of tandem repeats (VNTRs) investigated with restriction fragment length polymorphism (RFLP) and single 18 locus probes (SLPs). A total of 91% (2000) and 86% (2001) submitted typing results of polymerase chain reaction (PCR) based 19 systems. Typing errors occurred in 0.3% of the submitted PCR-based results in 2000 and in 0.1% in 2001. The results of the 20 paper challenges showed a high degree of variation in the formulas used for calculation of the weight of evidence of rare events 21 22 such as inconsistencies or possible silent alleles. The majority of the laboratories used the same formulas for calculations of 23 frequently occurring events. © 2002 Published by Elsevier Science Ireland Ltd. 24

25 *Keywords:* Paternity testing; DNA profiling; Collaborative exercise; Paternity Index; STR; VNTR; Single locus probes 26

### 27 1. Introduction

28 The English Speaking Working Group (ESWG) of the 29 International Society for Forensic Genetics (ISFG) offers once a year a Paternity Testing Workshop involving genetic 30 31 analysis in a paternity case [1-3]. The purpose of the exercise is to compare typing results as well as laboratory 32 33 strategies among the participating laboratories. As the 34 laboratories do the analyses and statistical calculations using 35 there own methods and frequency data, it is not possible to compare statistical calculations. In order to investigate how 36 37 the laboratories do statistics, a paper challenge has been 38 included in the Paternity Testing Workshop since the year 39 2000. The paper challenges were designed to include both

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routine combinations and rare events such as inconsisten- 40 cies, possible silent alleles and rare alleles. 41

When performing paternity testing, there is a need for obtaining knowledge about mutation rates for the systems used. As the information obtained in each laboratory is often limited, the laboratories were encouraged to provide information about inconsistencies observed in their own laboratory. 47

Here, we present the results of the Paternity Testing Workshops 2000 and 2001 including evaluation of laboratory strategies and typing results. Also presented are the results of the paper challenges and the calculated mutation rates based on the information given by the participating laboratories. 52

### 2. Materials and methods

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Blood samples were sent to the participating laboratories 54 together with a questionnaire and a paper challenge. The 55

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numbers of participating laboratories were 33 (2000) and 36
(2001). A list of participating laboratories is shown in
Appendix A.

In 2000, the blood samples were drawn from three children and their biological mother. The issue in question was whether child 3 was child of the same non-investigated man as children 1 and 2.

In 2001, the blood samples were drawn from two children and an alleged father. The children were known to be children of the same non-investigated mother and the issue in question was whether the children were children of the alleged father.

68 Laboratories that do immigration cases were encouraged 69 to treat the cases as they would do in immigration cases in 70 addition to the paternity cases.

The collated results were presented and discussed by the members of the ESWG at meetings held in 2000 (Söderköp-

73 ing, Sweden) and in 2001 (Münster, Germany).

#### 74 3. Results

### 75 3.1. Methods used for genetic investigations

76 Table 1 shows methods available for genetic investiga-77 tions in the participating laboratories. A total of 94% of the laboratories used polymerase chain reaction (PCR) based 78 79 systems for paternity testing. The numbers of laboratories 80 offering typing of variable number of tandem repeats 81 (VNTRs) with restriction fragment length polymorphism (RFLP) and single locus probes (SLPs) decreased from 82 83 2000 to 2001. In 2001, less than 50% of the laboratories 84 had SLPs available for paternity testing. The percentage of laboratories using conventional systems decreased from 85 18% in 2000 to 11% in 2001. There was an increase in the 86 number of laboratories having mitochondrial DNA 87 (mtDNA) and/or Y-chromosomal short tandem repeats 88 89 (Y-STRs) available for typing. In 2001, these systems 90 were available in a larger number of laboratories (44%) 91 than the SLPs (42%).

Table 2 shows methods used in all cases, methods used in addition when necessary and methods available for use in

 Table 1

 Methods available for genetic investigations in paternity testing

Methods $2000$ $(N = 33)$ (%) $2001$ $(N = 36)$ (%)VNTR/STR systems (PCR)9494mtDNA, Y-STR3944SLP systems5842HLA systems3325MLP systems1514PolyMarker (PCR)1811Conventional systems1811	-		
VNTR/STR systems (PCR)         94         94           mtDNA, Y-STR         39         44           SLP systems         58         42           HLA systems         33         25           MLP systems         15         14           PolyMarker (PCR)         18         11           Conventional systems         18         11	Methods	2000 ( <i>N</i> = 33) (%)	2001 ( <i>N</i> = 36) (%)
mtDNA, Y-STR     39     44       SLP systems     58     42       HLA systems     33     25       MLP systems     15     14       PolyMarker (PCR)     18     11       Conventional systems     18     11	VNTR/STR systems (PCR)	94	94
SLP systems5842HLA systems3325MLP systems1514PolyMarker (PCR)1811Conventional systems1811	mtDNA, Y-STR	39	44
HLA systems3325MLP systems1514PolyMarker (PCR)1811Conventional systems1811	SLP systems	58	42
MLP systems1514PolyMarker (PCR)1811Conventional systems1811	HLA systems	33	25
PolyMarker (PCR)1811Conventional systems1811	MLP systems	15	14
Conventional systems 18 11	PolyMarker (PCR)	18	11
	Conventional systems	18	11

#### Table 2

Methods used always, additional if necessary, and available for special cases

Methods	2000 ( $N = 33$ )	2001 ( $N = 36$ )
Always (%)		
STR (PCR)	76	86
SLP	33	22
Conventional	15	11
VNTR (PCR)	6	6
MLP	6	6
HLA	3	6
PolyMarker (PCR)	0	3
mtDNA, Y-STR	3	0
Additional (%)		
STR (PCR)	76	78
SLP	45	36
HLA	24	14
mtDNA, Y-STR	12	14
VNTR (PCR)	6	8
MLP	6	8
Conventional	6	3
PolyMarker (PCR)	12	0
Available (%)		
mtDNA, Y-STR	24	33
STR (PCR)	15	14
HLA	18	11
PolyMarker (PCR)	6	8
SLP	9	6
Conventional	6	3
MLP	6	3
VNTR (PCR)	6	3

very special cases. Only 3% (2000) or 0% (2001) of the 94 laboratories used mtDNA and/or Y-STRs for typing in all 95 cases whereas 12% (2000) to 14% (2001) used these 96 systems additionally when necessary, and 24% (2000) to 97 33% (2001) had mtDNA and/or Y-STRs available for very 98 special cases. 99

# 3.2. Inter-laboratory comparisons of RFLP typing with 100 SLPs 101

Typing results of systems investigated with RFLP were 102 submitted by 12 laboratories in 2000 and by 11 laboratories 103 in 2001. Typing results from 14 SLPs were reported in 2000 104 and of these, results from nine systems were reported by 105 more than one laboratory. In 2001, typing results from 10 106 SLPs were reported and of these, results from nine systems 107 were reported by more than one laboratory. In 2001, all 108 laboratories used HinfI as a restriction enzyme. Table 3 109 shows the most commonly used systems in 2000 and 2001. 110 The mean coefficient of inter-laboratory variation of DNA-111 fragment sizes of the VNTR systems D2S44, D7S21, D7S22 112 and D12S11 was 1.03% in 2000 and 1.38% in 2001. 113

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Table 3 The most frequently used VNTR systems for RFLP typing with SLPs in paternity testing

VNTR systems	Probe	2000 ( <i>N</i> = 19) (%)	2001 ( <i>N</i> = 15) (%)
D2S44	YNH24	89	93
D12S11	MS43a	84	93
D7S21	MS31	84	87
D5S110	MS621	68	80
D7S22	g3	53	73
D16S309	MS205	68	53
D1S7	MS1	37	47
D5S43	MS8	37	47
D4S139	pH30	26	33

#### 114 3.3. PCR typing

115 In 2000, a total of 31 participating laboratories performed 116 STR analysis. Of these, 27 laboratories used commercially 117 available kits. In 2001, a total of 34 participating laboratories performed STR analysis and of these, 31 laboratories used 118 119 commercially available kits. Table 4 shows the most com-120 monly used kits for STR analysis. The use of allelic ladders 121 for assigning the alleles increased from 74% in 2000 to 94% in 2001 (data not shown). Repeat units as nomenclature for 122 the alleles was used, either alone or in combination with 123 124 other kinds of nomenclature, by approximately 80-90% of the laboratories (Table 5). Among the PCR-based typing 125 results submitted in the paternity testing exercises in 2000 126 and 2001, inconsistent nomenclature was only reported in 127 128 the two systems DYS389 and D19S253.

### 129 3.4. Inter-laboratory comparisons of results of PCR typing

Typing results from a total of 64 (2000) and 58 (2001) PCR-based systems were submitted in the paternity testing exercises. Results from 43 (2000) and from 36 (2001) systems were reported by more than one laboratory.

Table 5				
Nomenclature	used	for	PCR-based	systems

Table 4

Frequently used STR-kits for PCR-based typing in paternity testing in 2000 and 2001

Kits	2000 ( $N = 27^{a}$ ) (%)	2001 ( $N = 31^{a}$ ) (%)
SGM Plus (Applied Biosystems)	56	68
Profiler Plus (Applied Biosystems)	59	48
PowerPlex 16 (Promega)	-	45
Profiler (Applied Biosystems)	30	19
FFFL (Promega)	22	19
Cofiler (Applied Biosystems)	22	13
Polymarker (Applied Biosystems)	19	13

<sup>a</sup> Number of laboratories using commercially available STRkits for DNA-typing.

Inter-laboratory comparisons of the results showed typing134errors that was not caused by inconsistent nomenclature or by135reporting errors with frequencies of 0.3% in 2000 and 0.1% in1362001. In 2000, no reporting errors were observed. In 2001,137reporting errors counted for 0.1% of the submitted results.138

# 3.5. Mutation rates 139

The participating laboratories were encouraged to submit 140 information about genetic inconsistencies observed in their 141 own laboratory (Tables 6 and 7). In general, the paternal 142 mutation rates were higher than the maternal mutation rates 143 and the overall mutation rates were higher for the VNTR 144 systems than for the STR systems. For the VNTR systems, 145 paternal mutation rates higher than 1% were seen in the 146 systems D1S7, D4S139 and D7S21. For all STR systems, the 147 mutation rates were below 0.5%. 148

### 3.6. Conclusions of the Paternity Testing Workshop and 149 statistics used in general 150

In 2000, 94% of the laboratories correctly concluded that 151 the biological father of children 1 and 2 was excluded from 152

Nomenclature	2000		2001		
	Manual electrophoresis $(N = 9)$ (%)	DNA sequencer $(N = 27)$ (%)	Manual electrophoresis $(N = 9)$ (%)	DNA sequences $(N = 33)$ (%)	
Repeat units	67	78	56	73	
Bp	0	4	0	6	
'Types'	11	0	11	6	
Repeat units and 'types'	0	4	0	3	
Repeat units and bp	11	11	11	3	
Repeat units and bp and 'types'	11	0	11	0	
No information	0	4	11	9	
Sum	100	101	100	100	

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### Table 6 Mutation rates for VNTR systems

VNTR system	Probe	Paternal	Paternal			Maternal			
		Meioses N	Mutation rate (%)	No. of labs	Meioses N	Mutation rate (%)	No. of labs		
D1S7	MS1	1157	4.24	4	1448	3.80	4		
D1S80	_	1522	0.26	2	1507	0.13	2		
D2S44	YNH24	9843	0.23	5	10319	0.17	6		
D4S139	pH30	2245	1.25	3	2671	0.15	3		
D5S110	MS621	1833	1.71	3	2106	0.52	3		
D7S21	MS31	10704	1.47	7	11293	0.06	7		
D7S22	g3	2604	0.77	4	3071	0.10	4		
D12S11	MS43a	10270	0.10	6	10780	0.01	6		
D16S309	MS205	2452	0.82	4	2804	0.36	4		

Table 7

Mutation rates for STR systems

STR system	Paternal			Maternal		Inconsistency between woman and child	
	Meioses N	Mutation rate (%)	No. of labs	Meioses N	Mutation rate (%)	No. of labs	or man and child ( <i>N</i> )
Amelogenin	5753	0.05	3	5418	0.02	2	,0,
CSF1PO	4596	0.13	4	5038	0.02	3	'0'
D2S1338	1755	0.23	5	2295	0.09	7	'0'
D3S1358	5762	0.21	5	6837	0.04	7	2
D5S818	5113	0.16	5	5634	0.07	4	'0'
D7S820	5031	0.10	5	5933	0.03	6	'0'
D8S1179	2613	0.34	5	3295	0.03	7	'0'
D13S317	5545	0.20	6	5603	0.04	4	'0'
D16S539	1544	0.19	3	2088	0.05	4	'0'
D18S51	3346	0.30	5	3043	0.03	6	'0'
D19S433	2582	0.12	4	2167	0.09	6	1
D21S11	4200	0.19	6	4029	0.15	8	1
F13A01	723	0.14	2	760	<b>'</b> 0'	2	'0'
FIBRA (FGA)	6926	0.46	7	7504	0.01	7	1
Penta D	233	<b>'</b> 0'	2	325	<b>'</b> 0 <b>'</b>	2	<b>'</b> 0 <b>'</b>
Penta E	415	0.24	2	384	<b>'</b> 0 <b>'</b>	2	'0'
TH01	6242	0.02	3	7112	<b>'</b> 0 <b>'</b>	5	<b>'</b> 0 <b>'</b>
TPOX	4579	0.04	3	5098	<b>'</b> 0 <b>'</b>	3	<b>'</b> 0 <b>'</b>
vWA	17717	0.30	9	19116	0.02	10	3

paternity of child 3. A total of 3% of the laboratories
concluded that the results were inconclusive and 3% did
not submit a conclusion. In 2001, all laboratories correctly
concluded that the alleged father could be the biological
father of both investigated children.

The probability of paternity (W) and/or the paternity index (PI) for calculating the weight of evidence in routine paternity cases were used by 73% (2000) and 78% (2001) of the laboratories (Table 8). Only 6% (2000) and 3% (2001) did not use statistics when evaluating the results in routine paternity cases.

164 The requirements for issuing a report with positive weight 165 for paternity varied among the laboratories. In general, the requirements increased from 2000 to 2001 and in 2001, 33% 166 of the laboratories required a paternity index of 10,000 or 167 more for issuing a report (Table 9). 168

# 4. Paper challenge

# 4.1. The paper challenge 170

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As laboratories used different systems for typing as well 171 as different frequencies in calculations, comparison of calculated PI-values in the performed paternity testing was not 173 possible. In order to compare calculations and to compare 174

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Table 8							
Statistics	used	in	reporting	results	of	paternity testing	

Statistics	2000 ( <i>N</i> = 33) (%)	2001 (N = 36) (%)		
W (probability of paternity)	73	78		
PI (paternity index)	73	78		
EM value	18	14		
Probability of exclusion /exclusion chance	15	11		
Other	9	3		
No statistics	6	3		
No information	-	3		

how laboratories deal with inconsistencies, possible silentalleles as well as rare alleles, a paper challenge was included

177 in the exercise.

178 In 2000, the paper challenge included a total of eight 179 systems. In one system, a shared silent allele between the 180 man and the child was present. In two systems, inconsis-181 tencies were present. It was not possible to decide whether 182 the inconsistencies were between the woman and the child or 183 between the man and the child. A total of 40% of the 184 laboratories that calculated the paper challenge concluded 185 that 'paternity is excluded'. A total of 50% concluded that 'results are inconclusive' and 10% concluded that 'results 186 187 are in favour of paternity'.

188 A total of 13 laboratories submitted results of PI-values 189 calculated for each system. Due to the inconsistencies and 190 possible silent alleles, a total of five laboratories did not 191 calculate a cumulative paternity index. All the remaining 192 eight laboratories reported different cumulative PI-values.

193 In 2001, the paper challenge included nine systems. In 194 one system, an inconsistency between the woman and the 195 child was present. In two systems, the man and the child shared a rare allele and in three systems possible silent 196 197 alleles were present. A total of 65% of the laboratories 198 concluded that 'results are in favour of paternity' whereas 199 35% concluded that 'paternity cannot be excluded, addi-200 tional testing is recommended'.

 Table 9

 Requirements for issuing a report with positive weight for paternity

A total of 22 laboratories submitted results of PI-values201calculated for each system. A total of 21 different cumula-202tive PI-values were submitted. The main difference in the203reported PI-values were caused by different calculations204when possible silent alleles, inconsistencies and rare alleles205were present. In the remaining systems, 82–91% of the206laboratories reported consistent PI-values.207

### 4.2. Rare alleles

In the 2001 paper challenge, information about the num-209 ber of observations of the allele in the database and the total 210 number of alleles in the database was provided. Approxi-211 mately 50% of the laboratories used a minimum frequency 212 for alleles that were in the database with zero or one 213 observation (Table 10). For alleles that were not present 214 in the database, 32% used a frequency of  $1/N_{tot}$ , where  $N_{tot}$  is 215 the total number of alleles in the database. A total of 5% did 216 not calculate a paternity index and a total of 9% used a 217 frequency in which the alleles observed in the present case 218 were added to the database. 219

If the allele was in the database with only one count, 45% 220 used a frequency of  $1/N_{tot}$  whereas 5% used a frequency in 221 which the observed alleles in the present case were added to 222 the database. 223

# 4.3. Silent alleles 224

In the 2000 paper challenge, one system included a 225 possible silent allele shared between the man and the child. 226 A total of 23% of the laboratories did not consider possible 227 silent alleles. In the 2001 paper challenge, three systems 228 included possible silent alleles shared between the man and 229 the child. 230

Table 11 shows how frequencies of possible silent alleles231were calculated. The constellations shown are from the232paper challenge in 2001. The percentage of laboratories233that did not consider silent alleles as possible alleles varied234from 64 to 82% depending on whether silent alleles was235present in the database and whether the alternative to a236shared silent allele was an inconsistency or not. Among the237

PI-values	W-values <sup>a</sup>	2000 ( <i>N</i> = 33) (%)	2001 ( <i>N</i> = 36) (%)
100-1000	99%-99.9%	24	19
1000-10000	99.9%-99.99%	33	22
10000-100000	99.99%-99.999%	21	25
>100000	> 99.999%	3	8
Less than a certain number of inconsistencies	9	11	
No requirement/other	9	14	

<sup>a</sup> The a posteori probability of paternity,  $W_{\text{post}}$ , is calculated based on the a priori probability of paternity,  $W_{\text{prior}}$ , and the PI, which is a true likelihood ratio.  $W_{\text{post}} = \text{PI} \times W_{\text{prior}}$ . By tradition, the *W*-value is calculated under the assumption that  $W_{\text{prior}} = 0.5$ .

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Table 10

		e ,						
Constellation No. (		No. ( <i>x</i> ) of observed	Calculation of the frequency of allele c					
Woman	Child	Man	c-alleles in database	A minimum frequency <sup>a</sup> (%)	$1/N_{\rm tot}^{\ b}(\%)$	(x + ac) $(N_{tot} + ac)$	t. obs.)/ act. obs.) <sup>c</sup> (%)	x/N <sub>tot</sub> (%)
ab ab	bc bc	cd cd	0 1	55 50	32 45x	9 5		$5 = 1/N_{tot}$

Frequency calculation of rare alleles in the 2001 paper challenge (N = 22)

<sup>a</sup> Among the 11 and 12 laboratories using a minimum frequency for calculation, a total of 10 different frequencies were used. Four of these were reported as formulas  $(3/N_{tot}, 5/N_{tot}, 5/(2N_{tot}))$  and  $1 - 0.05^{1/N_{tot}}$  whereas seven were reported as a fixed frequency ranging from 0.003 to 0.015.

<sup>b</sup>  $N_{\text{tot}}$  is the total number of alleles in the database.

<sup>c</sup> act. obs. is the number of allele c observed in the present case.

Table 11 Frequency calculation and number of formulas used in constellations with silent alleles (N = 22)

Constellation			No. ( <i>x</i> ) of observed	Calculation of the frequency of the 0-allele			No. of formulas used
Woman	Child	Man	0-alleles in database <sup>a</sup>	0-alleles not considered (%)	$x/N_{\text{tot}}^{b}$ (%)	Minimum frequency (%)	for calculation of PI
ab	b	с	2	64	20	16	12
a	а	а	2	77	9	14	4
ab	a	а	0	82	9	9	5

<sup>a</sup> 0-alleles: silent alleles.

<sup>b</sup>  $N_{\text{tot}}$ : the total number of alleles in the database.

three systems containing possible silent alleles, a total of 4– 12 different formulas were used for calculations.

# 240 4.4. Inconsistencies

In 2000, the paper challenge included two systems with 241 inconsistencies. It was not possible to decide whether the 242 243 inconsistency was between the woman and the child or 244 between the man and the child. A total of 13 laboratories 245 submitted data for these two systems. Of these, five labora-246 tories did not calculate a paternity index. Among the eight remaining laboratories, four different formulas were used for 247 248 calculations.

In 2001, a maternal inconsistency was present in a single
system. Three laboratories did not calculate a paternity index
for that system. Among the remaining laboratories, a total of
nine different formulas were used for calculation of PI.

### 253 5. Discussion

From 1995 to 2001, the percentage of laboratories using RFLP typing with SLPs decreased from 100 to 42% ([2], Table 1). Inter-laboratory comparison of the results for the systems D2S44, D7S21, D7S22 and D12S11 showed a mean coefficient of variation of 1.03% in 2000 and of 1.38% in 2001. The slightly higher variation in 2001 seemed to be due259to a higher variation among laboratories in general.260

In 2001, analysis of mtDNA and/or Y-STRs was available 261 in a larger number of laboratories than RFLP-based analysis 262 with SLPs. As mtDNA and Y-STR testing investigate female 263 and male transmission, respectively, these investigations are 264 especially useful in cases with special circumstances. 265 mtDNA and/or Y-STR typing was mainly used when addi-266 tional systems had to be included in the analysis or when 267 special circumstances were present. 268

In laboratories using DNA sequencers for STR analysis, 269 allelic ladders were used only by 74% in 2000 but this 270 percentage increased to 94% in 2001. Inclusion of 271 sequenced allelic ladders in STR analyses improves the 272 accuracy of allele designation [4] and has been recom-273 mended by the DNA Commission of the ISFG [5-7]. 274 Commercial allelic ladders generally use a nomenclature 275 based on the number of repeat units as recommended by the 276 DNA Commission of the ISFG [6,7]. In 2000 and 2001, 277 almost 80-90% of the laboratories performing STR analysis 278 used a nomenclature based on repeat units. 279

Inconsistent nomenclature was observed in the systems 280 DYS389 and D19S253. A new nomenclature has been 281 recommended for the system DYS389 (http://www.ystr.charite.de) and the inconsistency here was due to the fact that 283 some laboratories used the old nomenclature. 284

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Reported errors were seen in 0.3% of the typing results
investigated by PCR in 2000 and in 0.2% in 2001. In 2000,
all errors seemed to be due to errors in typing. In 2001, 0.1%
reporting errors were observed and 0.1% seemed to be due to
typing errors.

290 As mutations are rare events, the information that each laboratory is able to collect is limited and the Paternity 291 Testing Workshop cumulated information about mutation 292 293 rates (Tables 6 and 7). Different laboratories may, how-294 ever, use different criteria for the definition of a mutation 295 in VNTR systems. The VNTR systems had in general 296 higher mutation rates than the STR systems. Comparison 297 of the mutation rates with those collected by the American 298 Association of Blood Banks (AABB) (http://www.aab-299 b.org/) showed differences mainly in the VNTR systems D1S80, D5S110, D7S21, D7S22, D16S309 and in the STR 300 systems D3S1358, F13A01 and FGA. In the remaining 301 302 systems there was a good concordance between the mutation rates observed in the AABB surveys and in the ESWG 303 304 exercises.

305 In 1999, 55-60% used the PI or/and W-value when 306 evaluating the weight of evidence [3]. This percentage has increased and today, almost 80% of the laboratories 307 308 report the PI or/and the W-value. During the last 3 years, the 309 requirements for issuing a report with positive weight for 310 paternity has increased. In 1999, 21% of the laboratories required a PI-value of 10,000 or more [3]. This percentage 311 increased to 33% in 2001 (Table 9). 312

To analyse how laboratories performed statistical analyses, a paper challenge was included in the workshop. The paper challenge consisted of a fictive paternity case in which constellations included inconsistencies, possible silent alleles and rare alleles.

Some laboratories did not calculate a PI-value if inconsistencies were present. Among the laboratories that did
consider mutations, a large number of formulas were used
for calculation.

Silent alleles have been described for several systems and
 occur, e.g. when the allele size is outside the range of the
 measurement. In PCR-based systems, silent alleles may also
 be caused, e.g. by mutations in a primer binding site [8].

In the paper challenges, 23–82% of the laboratories did
not consider silent alleles. Among the remaining laboratories, silent alleles were treated differently and different
formulas were used for calculations.

For rare alleles the frequencies used for calculations
varied. Almost half of the laboratories used a fixed minimum
frequency when the allele in question was absent or found
only once in the database. The minimum frequency differed
among the laboratories.

In conclusion, the results of the Paternity Testing Workshops 2000 and 2001 showed a high degree of concordance
concerning techniques, systems and nomenclature used.
Typing errors counted for only 0.1–0.3% of the PCR-based
systems and inconsistent nomenclature was observed in only
two systems. Also calculations of PI-values in constellations

with no special events showed a high degree of uniformity.341However, when rare events such as inconsistencies and<br/>possible silent alleles were present, differences in calcula-<br/>tions were observed.343

### Acknowledgements

Our thanks are due to our blood donors and to the staff of 346 the participating laboratories. 347

### Appendix A.

#### A.1. Participating laboratories

• Amtlich benannter Sachverständiger für Abstammungs-	
gutachten, Chefarzt am Zentrallaboratorium, Berlin, Ger-	351
many (2000, 2001).	352
• Antwerp Blood Transfusion Center, Edegem, Belgium	
(2000, 2001).	354
• Arzt für Laboratoriumsmedizin, Heidelberg, Germany	
(2000, 2001).	356
• BJ Diagnostik GmbH, Giessen, Germany (2001).	
• Cellmark Diagnostics, Abingdon Business Park, Oxon,	
UK (2001).	359
Codgene, Institut de Médecine Légale, Strasbourg, France	
(2000, 2001).	361
• Department of Forensic Genetics, Institute of Forensic	
Medicine, Copenhagen, Denmark (2000, 2001).	363
• Department of Forensic Medicine & Science, The Uni-	
versity of Glasgow, Glasgow, UK (2001).	365
• Department of Forensic Medicine, Medical Academy of	
Lodz, Lodz, Poland (2001).	367
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