ANNUAL REPORT SUMMARY FOR TESTING IN 2001

Prepared by the Parentage Testing Program Unit
October 2002

PREFACE

This year's annual report continues the past precedent of providing basic summary statistics for testing that took place in the previous year, 2001. The emphasis of the survey questions this year, however, was on apparent mutations and null alleles. This included asking how laboratories were incorporating mutations into the final report and how laboratories were handling situations were there were two or three inconsistencies. As in the past mutations observed for 2001 are provided in table form.

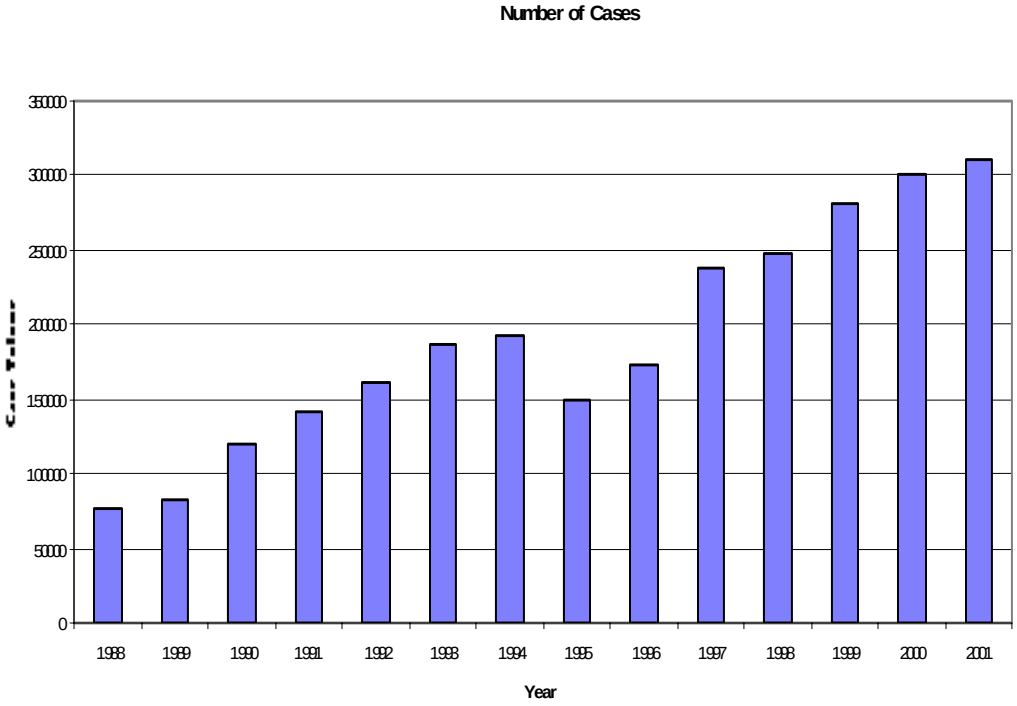
ANNUAL VOLUME OF TESTING

2001 saw another increase in the number of relatedness cases reported. The volume reported was 310,490, an increase of about 3% over last year's volume. A summary of the totals of all years since 1988 is shown in Table 1.

Table 1. The number of parentage cases reported for various years.

Year	No. of Cases
1988	77000
1989	83000
1990	120000
1991	142000
1992	161000
1993	187000
1994	193000
1995	149100
1996	172316
1997	237981
1998	247317
1999	280510
2000	300626
2001	310490

Figure 1. Graph of the case volume for various years.



The data includes totals for the first AABB accredited European laboratory as well as data from one other European laboratory. A total of 46 laboratories responded to the survey. Approximately 79 requests for information were made, with 46 (58%) laboratories responding. Some of the laboratories had closed.

Table 2. Laboratories by the volume of cases reported.

Case Volumes	1994	1995	1996	1997	1998	1999	2000	2001
1-500	40	26	25	20	19	19	13	17
501-1000	6	4	8	7	6	5	6	6
1001-5000	7	9	6	10	11	9	11	11
5001-10000	6	4	3	5	0	3	3	5
10001-50000	1	2	3	5	5	7	8	6
>50001	2	1	1	1	2	1	1	1
Total Laboratories	62	46	46	48	43	44	42	46

Of the cases reported 90,227 were reported as exclusions or a rate of 29.06% exclusions. The average exclusion rate for the laboratories is 28.10% with a standard deviation of 7.17. The median exclusion rate is 29.25% and the mode is 27.87% with a range of 11.03 – 40.86%.

COMBINED PATERNITY INDEX

This year the laboratories were asked to indicate what combined paternity index (CPI) they considered acceptable for cases with a standard trio (mother, child, father), mother not tested, and for reconstruction cases. Some laboratories reported using different CPIs for different classes of clients (private versus public contracts). For these laboratories the higher CPI was used for this report. The results are shown in Table 3. The most common minimum PI for standard trios is 100 with 30 laboratories out of 46 (65.22%) using this value, with a range of 100 to 10,000. With MNT cases the lowest accepted CPI dropped to 50 and for reconstruction cases the lowest CPI reported was 10, with a number of laboratories indicating that for these cases they used "whatever was obtained".

Table 3. The number of laboratories using various combined paternity indices for standard trios, mother not tested (MNT) and reconstruction cases (Note that not all laboratories indicated a CPI for each type of case).

	Type of Case		
CPI	**Trio**	**MNT**	**Reconstruction**
10	0	0	2
50	0	1	1
60	0	1	0
100	30	30	18
101	0	1	0
150	2	2	2
200	1	2	1
300	1	0	0
500	2	1	0
1000	3	2	1
1001	1	0	0
2500	1	1	1
10000	2	1	0

TECHNOLOGY USE

The type of technology used continues to show the trend towards the increased use of PCR technology with a decrease in the use of RFLP methods. PCR STR technology was used in 83.34% of reported cases while RFLP analysis was used in 16.00% of reported cases. All other technologies were used in about 0.66% of reported cases. Table 4 provides a breakdown

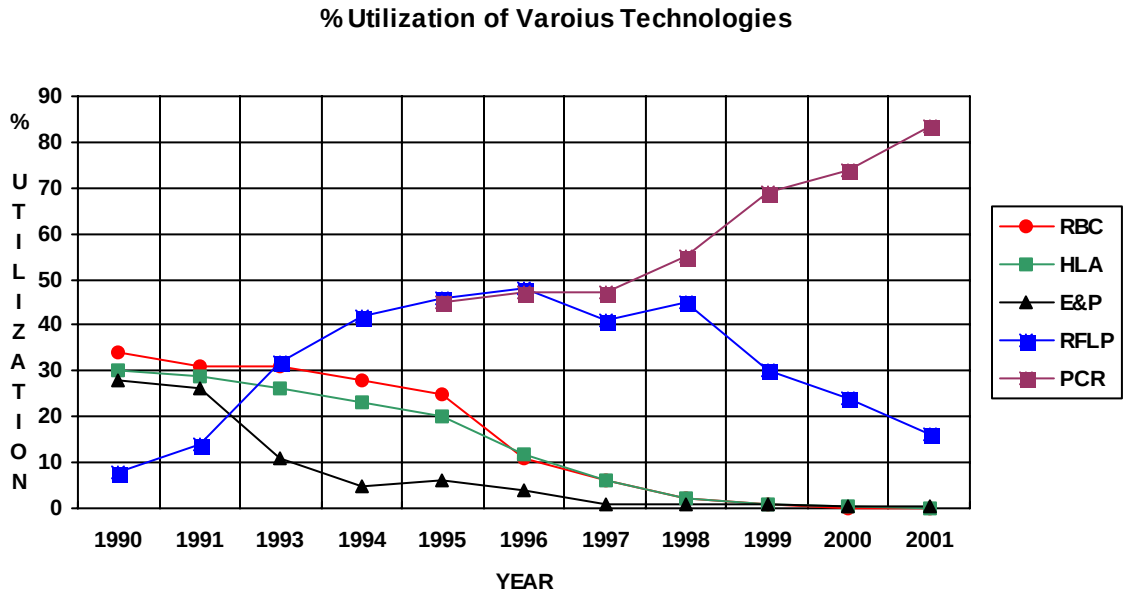
of the technology used to resolve the reported paternity cases. Note that in some cases more than one technology was used so the sum of the number of cases is greater than the numbers given in the volume section above. The question was also asked if the laboratory is using HLA molecular methods what is the source of the frequencies. A number of laboratories that reported not using HLA molecular methods indicated that if they did use these methods they would not use serological tables, while all the laboratories actually using HLA molecular methods reported using serological tables for calculating Class I molecular results. No laboratories reported using SNP technology and a few laboratories are using Y Chromosome analysis in their testing programs.

Table 4. The technology used and number of relationship cases reported in 2001 (in some cases more than one technology was used).

Technology	Number of Cases	Utilization (%)
Red Cell Antigens	5	0.002
HLA Serology	2	0.001
HLA Class 1 Molecular	83	0.026
HLA Class II Molecular	326	0.104
Red Cell Enzymes/Serum Proteins	924	0.294
Allotyping	735	0.233
RFLP	50360	15.998
STR	262344	83.338
SNP	0	0
Y Chromosome	28	0.009
Total of All Technologies	314797	

*Note that some cases used more than one technology therefore this total is higher than the total number of cases reported.

Figure 2 shows the utilization of various technologies since 1990. As indicated above the most commonly used technologies in 1990 (red cell antigens, HLA and red enzymes and serum proteins) now account for less than 1% of all casework. The change in DNA technologies from RFLP to PCR technology is obvious, however prior to 1995 the use of PCR was not tracked in the Annual Reports. Note that in some cases multiple technologies were used in the same case.



SAMPLE SOURCE

There were a total of 741,271 samples used for the casework in 2001. Of these, buccal swabs account for 649,375 (87.60%). The other samples used included 89,503 (12.07%) whole blood samples, 2,238 (0.30%) blood spot cards, and 155 (0.02%) other samples which include various tissues, bone, amniotic fluid, hair and undefined samples.

PROBABILITY OF EXCLUSION

Another new question on this Annual Report was a request for the probability of exclusion for each locus used. A number of laboratories did not respond to this request. The exact reason for not reporting this is not known however a number of laboratories indicate that the PE was "not tracked". This is disturbing as the PE for STR can be calculated from the frequency data or from the heterozygosity of the population data used to obtain the frequencies. This subject will be further addressed in future additions of the guidance document for the parentage testing standards. The original intent was to break this data apart by the source of the frequency tables used in the laboratory, however because some loci were used by only one laboratory and in other cases it was unclear what was the source of the frequency table all the data was pooled. The source of the frequency tables included ABI, Promega, in house, Orchid (LifeCodes), Reliagene and others.

None of the loci/probe/enzyme combinations evaluated with RFLP testing were used (reported) by more than four laboratories. The data was reported for all loci even if a locus was only used by one laboratory. Therefore these data should be viewed with caution.

Table 5. The probability of exclusion reported for various loci evaluated using RFLP methods (Note that no loci had more than four laboratories report results).

LOCUS	PROBE	ENZYME	PE	LOCUS	PROBE	ENZYME	PE	LOCUS	PROBE	ENZYME	PE
D10S28	TBQ7	HAEIII	0.9396	D18S27	SLI604	PST1	0.7050	D6S132	SLI1090	PST1	0.8850
D10S28	SLI917	HAEIII	0.9600	D18S27	SLI605	PST1	0.7800	D6S132	PAC424	HAEII	0.7950
D12S11	SLI737	PST1	0.9033	D1S1339	SLI1335	HAEIII	0.9488	D6S132	SLI1090	HAEIII	0.9300
D12S11	MS43A	PST1	0.9500	D1S339	PAC425	HAEIII	0.8727	D7S21	SLI619	PST1	0.9500
D12S11	MS43	HINF1	0.8779	D1S7	MS1	HINF1	0.9616	D7S21	MS31	HINF1	0.9027
D14S13	CMM101	HAEIII	0.9140	D2S44	YNH24	HAEIII	0.9182	D7S22	G3	HINF1	0.8383
D16S85	SLI779	HAEIII	0.8500	D2S44	SLI106	PST1	0.7933	D7S467	SLI989	PST1	0.8450
D17S26	EFD52	HAEIII	0.8979	D2S44	SLI106	HAEIII	0.9500	D7S467	PAC415	HAEIII	0.8273
D17S26	SLI936	HAEIII	0.9000	D2S44	YNH24	HINF1	0.8831	D7S467	SLI989	HAEIII	0.9300
D17S79	SLI986	PST1	0.7133	D2S92	SLI874	HAEIII	0.9150				
D17S79	V1	HAEIII	0.7200	D4S139	PH30	HAEIII	0.8938				
D17S79	SLI441	HAEIII	0.7500	D4S163	SLI604	PST1	0.8350				
D5S110	PLH1	HAEIII	0.9499	D4S163	SLI604	HAEIII	0.8855				
D5S110	LH1	HAEIII	0.9500								

Table 6. The probability of exclusion for various loci evaluated using PCR (Note that for some loci only a single laboratory reported results).

LOCUS	PE	LOCUS	PE	LOCUS	PE
D3S1358	0.5646	D2S1338	0.7260	D3S1744	0.6823
VWA	0.6309	D19S433	0.5851	D18S849	0.5077
FGA	0.7182	F13A01	0.4934	D1S533	0.5270
D5S818	0.4768	FESFPS	0.4605	D9S304	0.5270
D13S317	0.5226	F13B	0.5123	D9S302	0.8200
D7S820	0.5991	LPL	0.5137	D22S683	0.8300
D8S1179	0.6096	PENTA E	0.7390	D18S535	0.5800
D21S11	0.6980	PENTA D	0.7000	D7S1804	0.8000
D18S51	0.7458	D1S80	0.6400	D3S2387	0.7700
TH	0.5468	D17S5	0.7000	D4S2366	0.5600
TPOX	0.3950	HPRTB	0.4520	D5S1719	0.7100
CSF1P0	0.5170	D13S308	0.6200		
D16S539	0.5581	D12S1090	0.8287		

For the CODIS loci a sufficient sample was available to make statistical analysis for some loci, although without regard to the database source as this was not always clear from the information provided. Table 7 shows a basic statistical analysis of the probabilities of exclusion provided for the CODIS loci. The range of probabilities of exclusion was large for some loci. For example in TPOX the range is 27.8% to 61%. It was interesting that several laboratories were found at or near both extremes. This variation may have been caused be using different frequency tables (population sampling differences) or by varying methods of determining the probability of exclusion. In next years survey this will be explored.

Table 7. The mean, standard deviation, mode, median, range and number of laboratories reporting results for the CODIS loci.

Locus	Mean	StDev	Mode	Median	Range	# Labs
D3S1358	0.5646	0.0275	0.5746	0.5556	0.53 - 0.63	19
VWA	0.6309	0.0217	0.6250	0.6280	0.603 - 0.68	23
FGA	0.7182	0.0775	0.7200	0.7210	0.419 - 0.8159	20
D5S818	0.4768	0.0246	0.4554	0.4691	0.44 - 0.516	19
D13S317	0.5226	0.0638	0.4430	0.5334	0.442 - 0.64	20
D7S820	0.5991	0.0307	0.5700	0.6000	0.5123 - 0.6307	21
D8S1179	0.6096	0.0382	0.6128	0.6100	0.53 - 0.68	19
D21S11	0.6980	0.0166	0.7230	0.7000	0.668 - 0.723	19
D18S51	0.7458	0.0117	0.7414	0.7450	0.725 - 0.78	19
TH	0.5468	0.0331	0.5360	0.5409	0.475 - 0.6178	24
TPOX	0.3950	0.0773	0.3600	0.3615	0.278 - 0.61	22
CSF1P0	0.5170	0.0594	0.4900	0.5030	0.47 - 0.66	23
D16S539	0.5581	0.0467	0.5700	0.5680	0.474 - 0.609	22

Mutation Calculation

Single inconsistencies are routinely seen in the testing of paternity cases. Following AABB standards if a laboratory comes to the conclusion that the inconsistency is a mutation, then the mutation result must be incorporated into the reported results. Laboratories were asked how they calculated the paternity index (PI) for these loci. The results varied widely with some laboratories using arbitrary numbers (1, 0.002 and 0.003 were reported) (6% of laboratories), some use the mutation rate as the PI (13% of laboratories), 54% of laboratories used the mutation rate divided by the average probability of exclusion. Some laboratories (27%) utilize methods that used the mutation rate as a transmission frequency with 21% of the laboratories using Brenner's method of using the mutation rate and looking at the repeat length difference between STR alleles.

Mutation Reports

One area of concern is the number of inconsistencies necessary to render an opinion of non-paternity. The laboratories were asked if they had seen any case where, in the opinion of the expert, a double or triple "mutation" was not sufficient to render an opinion of non-paternity. The laboratories

reported 47 cases with double mutations (0.015% of all reported cases) and five cases with triple mutations (0.002% of all reported cases) as inclusions. When reporting these cases most laboratories report them with the inconsistencies noted and statistically considered.

Tables 8 & 9 show the mutation rates for genetic markers analyzed by either RFLP or PCR. The data presented reflects data reported for 2001 unless otherwise noted. This was done to correct a perceived error in the previous mutation data. However, there does not appear to be any significant deviation from previous year's data.

Table 10 provides a new approach to reporting the number of length repeats from the obligatory allele for loci analyzed using PCR. This clearly shows that the differences in repeat lengths are plus or minus one repeat length from the obligatory allele. In next years survey an attempt may be made at obtaining actual data for specific changes from one allele to another.

Table 8. Σ Mutation Rates Summarized for Genetic Markers Analyzed by RFLP Mapping.

System	Maternal δ (%)	Paternal δ (%)	Null (%)	Multi-Banded (%)
D1S7*	9/580=1.55	11/721=1.52	1/560=0.17	2/461=<0.43
D1S339	217/91007=0.24	407/107664=0.38	97/96211=0.10	203/74646=0.27
D2S44	356/218066=0.16	263/259256=0.10	622/268976=0.23	458/280521=0.16
D4S139	43/80080=0.05	987/103641=0.95	26/82241=0.03	917/87296=1.05
D4S163	6/31487=0.02	80/72760=0.11	99/87008=0.11	21/70674=0.03
D5S110	141/25348=0.56	443/25321=1.75	11/28297=0.04	520/32790=1.59
D5SS43*	0/525=<0.19	0/536=<0.19	UNK.	UNK.
D6S132	14/66759=0.02	86/115412=0.08	4/139388=0.003	41/163417=0.03
D7S21	20/1073=1.86	41/1398=2.93	UNK.	1/1235=0.081
D7S22	15/2843=0.52	91/3292=2.76	UNK.	UNK.
D7S467	18/102899=0.02	185/172176=0.11	18/197506=0.009	46/189427=0.02
D10S28	354/198227=0.18	207/218283=0.09	106/197102=0.05	224/200039=0.11
D12S11	6/17712=0.034	16/21938=0.07	3/24575=0.001	7/21752=0.03
D14S13*	19/30596=0.06	108/33085=0.33	3/21391=0.01	119/26343=0.45
D16S309	0/286=<0.35	2/2234=0.09	UNK.	UNK.
D16S85	0/565=<0.18	3/614=0.50	4/795=0.5	0/795=<0.13
D17S26	61/63351=0.10	163/66533=0.25	6/22785=0.03	44/57617=0.08
D17S79	7/16621=0.04	25/22545=0.11	15/12255=0.12	26/19662=0.13

Σ cumulative compilation of current and previous AABB mutation data

* data from 2000 AABB Annual Report (no data submitted for these systems)

δ The data under these column headings refers to the number of inconsistencies/number of total meioses expressed as a percentage.

Null alleles are assumed when cases of paternal or maternal exclusion occur due to nonmatching homozygous banding patterns when there is otherwise overwhelming evidence in favor of paternity or maternity.

Table 9. Σ Apparent Mutations Summarized for Genetic Systems Analyzed by PCR

System	Maternal δ (%)	Maternal Null (%)	Paternal δ (%)	Paternal Null (%)	Paternal or Maternal
D1S80*	4/14052=.03	UNK.	75/199543=.04	2/60372=.01	UNK.
D1S2131*	0/1212=.08	UNK.	3/1240=.24	UNK.	UNK.
D1S533*	UNK.	UNK.	6/3830=.16	UNK.	UNK.
D2S1338	0/1025=<.1	0/1025=<.1	10/46195=.02	0/1630=0	3/2050=.15
D2S548*	1/1212=.08	UNK.	0/1240=<.08	UNK.	UNK.
D3S1358	14/94449=.02	4/209197=.002	193/147483=.13	11/113424=.01	132/171597=.08
D3S1744	16/10141=.16	0/5707=0	84/20290=.41	0/9197=0	UNK.
D3S2386*	0/1212=<.08	UNK.	1/1240=.08	UNK.	UNK.
D5S818	51/216242=.02	6/139968=.004	451/325299=.14	33/178346=.02	206/199941=.10
D7S820	30/206714=.02	3/131816=.002	379/303447=.13	13/155770=.008	136/168370=.08
D8S306*	1/1212=.08	UNK.	3/1240=.24	UNK.	UNK.
D8S1179	15/77866=.02	15/77866=.02	205/126616=.20	13/73502=.02	74/151368=.05
D9S302*	19/8332=.22	0/5669=<.02	49/11179=.44	0/8568=<.02	UNK.
D10S1214*	28/2903=.97	UNK.	114/2938=3.88	UNK.	UNK.
D12S1090	9/4894=.18	UNK.	111/12801=.87	0/3395=<.02	UNK.
D13S317	90/218730=.04	241/157391=.15	373/270701=.14	177/245806=.07	328/489431=.07
D14S297*	0/1212=<.08	UNK.	0/1240=<.08	UNK.	UNK.
D16S539	38/169351=.02	11/125403=.009	203/180286=.11	23/125493=.02	127/349637=.04
D17S5*	0/228=<.44	UNK.	7/6568=.11	UNK.	UNK.
D17S1185*	0/1212=<.08	UNK.	0/1240=<.08	UNK.	UNK.
D18S51	47/86851=.05	7/72410=.009	229/110748=.21	10/75782=.013	160/115433=.14
D18S535*	1/2676=.04	UNK.	2/2624=.08	0/5300=<.02	UNK.
D18S849	0/4291=<.03	UNK.	18/10440=.17	0/6750=<.02	UNK.
D19S253*	8/2997=.27	1/1785=.06	17/3247=.52	7/2007=.35	UNK.
D19S433‡	3/1025=.3	0/1025=0	1/1025=.1	0/1025=0	1/2050=.05
D21S11	107/102238=.1	13/90177=.01	182/118384=.15	11/85972=.01	221/220622=.1
D21S1437*	0/1212=<.08	UNK.	1/1240=.08	UNK.	UNK.
D22S445*	2/1212=.17	UNK.	1/1240=.08	UNK.	UNK.
D22S683*	2/2670=.08	UNK.	9/2625=.34	0/5295=<.02	UNK.
ACTBP2*	0/330=<.3	UNK.	330/51610=.64	UNK.	UNK.
CYP19*	6/343=1.75	UNK.	205/177210=.12	321/47259=.68	UNK.
CYAR04*	2/3539=.06	UNK.	UNK.	UNK.	UNK.
FGA	56/94290=.06	4/83342=.005	893/298824=.3	17/86854=.02	271/130332=.21
CSF1PO	43/179353=.02	2/129721=.002	573/394570=.14	12/148441=.008	190/149314=.13
FESFPS	3/18572=.02	1/9914=.01	79/148682=.05	0/17146=0	0/35718=0
F13A01	1/10166=.01	0/2297=0	37/65039=.06	0/4943=0	3/4233=.07
F13B	1/12324=.008	0/6902=0	8/26785=.03	0/10146=0	1/4938=.02
LPL	0/8470=0	0/4581=0	9/16592=.05	1/10635=.009	4/9944=.04
THO1	17/189478=.008	10/145630=.007	25/242231=.01	8/161892=.005	14/176805=.008
TPOX	7/169002=.004	0/140236=0	27/194792=.01	3/148086=.002	15/171420=.009
Penta E	10/19982=.05	1/19982=.005	30/21703=.14	1/21703=.005	34/27122=.13
Penta D‡	5/2297=.22	0/2297=0	2/2706=.07	0/2706=0	7/5003=.14
VWA	74/243918=.03	11/175088=.006	1388/454771=.30	45/223277=.02	368/211176=.17

Σ cumulative compilation of current and previous AABB mutation data

*data from 2000 AABB Annual Report (no data submitted for these systems)

‡ systems added since the 2000 AABB Annual Report

δ The data under these column headings refers to the number of inconsistencies/number of total meioses expressed as a percentage.

Null alleles are assumed when cases of paternal or maternal exclusion occur due to nonmatching homozygous banding patterns in cases in which there is overwhelming evidence in favor of paternity or maternity.

Table 10. The distance (repeat lengths) from the obligatory allele.

PCR MUTATIONS: DISTANCE FROM OBLIGATORY ALLELE (Expressed as Percent of Total Number of Mutations)

	Maternal						Paternal					
	STR Distance From Obligatory Allele						STR Distance From Obligatory Allele					
GENETIC SYSTEM	+1	-1	+2	- 2	OTHER	TOTAL #	+1	-1	+2	- 2	OTHER	TOTAL #
D2S1338	0.000	0.000	0.000	0.000	0.000	0	1.000	0.000	0.000	0.000	0.000	1
D3S1744	0.000	0.000	0.000	0.000	0.000	0	0.000	1.000	0.000	0.000	0.000	1
D3S1358	0.440	0.550	0.000	0.000	0.000	10	0.540	0.420	0.025	0.000	0.008	118
D5S818	0.410	0.500	0.090	0.000	0.000	24	0.580	0.380	0.030	0.000	0.013	172
D7S820	0.810	0.180	0.000	0.000	0.000	11	0.620	0.340	0.010	0.020	0.010	106
D8S1179	0.600	0.400	0.000	0.000	0.000	18	0.460	0.520	0.020	0.000	0.000	112
D12S1090	0.000	0.000	0.000	0.000	0.000	0	0.660	0.340	0.000	0.000	0.000	3
D13S317	0.520	0.480	0.000	0.000	0.000	122	0.650	0.330	0.023	0.000	0.000	188
D16S539	0.830	0.110	0.050	0.000	0.000	20	0.680	0.310	0.000	0.000	0.000	86
D18S51	0.400	0.570	0.030	0.000	0.000	32	0.500	0.470	0.009	0.017	0.000	129
D18S849	0.000	0.000	0.000	0.000	0.000	0	1.000	0.000	0.000	0.000	0.000	3
D19S433	0.670	0.330	0.000	0.000	0.000	3	1.000	0.000	0.000	0.000	0.000	1
D21S11	0.630	0.360	0.013	0.000	0.013	84	0.350	0.610	0.020	0.009	0.009	113
CSF1PO	0.550	0.450	0.000	0.000	0.000	26	0.750	0.220	0.030	0.008	0.000	148
FGA	0.380	0.590	0.000	0.023	0.000	49	0.490	0.500	0.005	0.005	0.000	220
F13A	0.000	1.000	0.000	0.000	0.000	1	0.500	0.000	0.000	0.500	0.000	2
F13B	0.000	0.000	0.000	0.000	0.000	0	0.000	0.000	0.000	0.000	0.000	0
FESFPS	0.000	0.000	0.000	0.000	0.000	0	1.000	0.000	0.000	0.000	0.000	1
LPL	0.000	0.000	0.000	0.000	0.000	0	0.330	0.330	0.330	0.000	0.000	4
PENTA D	0.800	0.000	0.000	0.000	0.200	5	0.500	0.000	0.000	0.000	0.500	2
PENTA E	0.550	0.220	0.110	0.000	0.110	9	0.750	0.200	0.000	0.000	0.050	22
THO1	0.860	0.140	0.000	0.000	0.000	7	0.250	0.250	0.000	0.500	0.000	7
TPOX	0.400	0.400	0.200	0.000	0.000	5	0.500	0.330	0.000	0.160	0.000	6
VWA	0.430	0.560	0.000	0.000	0.000	35	0.640	0.340	0.010	0.003	0.003	295