Allele Frequencies in 4 Str's in a Population of Portugal (Central Area)

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Systems and loci: HUMTHO1 (11 pl. 5.5 - p 15); HUMVWA31 (12 p 12 - pter.); HUMF13A1 (6 p 25 - p 24); HUMFESFPS (15 q25 - q ter).

Population and sample size: Portugal (Central Area). N: 344.

Methods: Primers (Kimpton et al. 1995)

PCR amplification conditions: Kimpton et al. 1995 (quadruplex).

Electrophoretic methods: 6% polyacrylamide denaturing gel electrophoresis. The gels were run for 6 h at constant power (30w), 2500 V and 40 mA on the ABI 373 A DNA Sequencer. Typing was performed using the internal standard Genescan Rox 2500 and allelic ladders from the 2nd EDNAP Collaborative STR Exercise (1995).

Results:

TH01

Observed genotypes

Gen.	Obs.	Gen.	Obs.	Gen.	Obs.	Gen.	Obs.
6-6	14	**7-7**	4	**8-8**	5	**9-9.3**	37
6-7	19	**7-8**	11	**8-9**	17	**9.3-9.3**	34
6-8	28	**7-9**	23	**8-9.3**	25	**9.3-10**	2
6-9	30	**7-9.3**	24	**8-10**	2		
6-9.3	54	**7-10**	2	**9-9**	13		

Allele frequencies

Allele	Frequency	Allele	Frequency	Allele	Frequency
6	0.2311	**8**	0.1352	**9.3**	0.3052
7	0.1265	**9**	0.1933	**10**	0.0087

Exact Test P=0.86

FES

Observed genotypes

Gen.	Obs.	Gen.	Obs.	Gen.	Obs.	Gen.	Obs.
8-10	3	**10-11**	86	**11-12**	61	**13-13**	1
8-11	7	**10-12**	55	**11-13**	7		
8-12	3	**10-13**	6	**12-12**	21		
10-10	32	**11-11**	53	**12-13**	9		

Allele frequencies

Allele	Frequency	Allele	Frequency	Allele	Frequency
8	0.0189	**11**	0.3881	**13**	0.0349
10	0.3110	**12**	0.2471		

Exact Test P=0.82

VWA

Observed genotypes

Gen.	Obs.	Gen.	Obs.	Gen.	Obs.	Gen.	Obs.
13-16	1	**15-15**	1	**16-19**	11	**18-19**	8
14-14	4	**15-16**	21	**16-20**	1	**18-20**	1
14-15	13	**15-17**	20	**17-17**	24	**18-21**	1
14-16	17	**15-18**	20	**17-18**	39	**19-19**	3
14-17	26	**15-19**	3	**17-19**	12	**19-20**	1
14-18	14	**16-16**	19	**17-20**	3		
14-19	5	**16-17**	40	**17-21**	2		
14-20	1	**16-18**	24	**18-18**	9		

Allele frequencies

Allele	Frequency	Allele	Frequency	Allele	Frequency
13	0.0014	**16**	0.2224	**19**	0.0669
14	0.1221	**17**	0.2762	**20**	0.0102
15	0.1148	**18**	0.1817	**21**	0.0043

Exact Test P=0.84

F13

Observed genotypes

Gen.	Obs.	Gen.	Obs.	Gen.	Obs.	Gen.	Obs.
3.2-3.2	6	**4-5**	2	**5-7**	40	**6-16**	2
3.2-4	1	**4-6**	5	**5-8**	2	**7-7**	51
3.2-5	8	**4-7**	6	**5-14**	4	**7-8**	5
3.2-6	19	**4-16**	1	**5-16**	3	**7-14**	3
3.2-7	17	**4-17**	1	**6-6**	35	**7-16**	4
3.2-16	1	**5-5**	10	**6-7**	67	**7-17**	2
4-4	2	**5-6**	46	**6-8**	1		

Allele frequencies

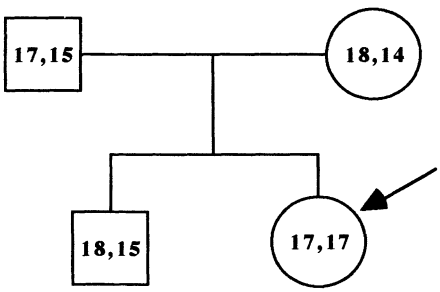
Allele	Frequency	Allele	Frequency	Allele	Frequency
3.2	0.0843	**6**	0.3052	**14**	0.0102
4	0.0291	**7**	0.3575	**16**	0.0160
5	0.1817	**8**	0.0116	**17**	0.0044

Exact Test P=0.24

Statistical parameters (4 STRs)

	TH01	FES	VWA	F13	Comb
CE	0.5616	0.4216	0.6219	0.5078	0.9528
PD	0.9145	0.8436	0.9365	0.8885	0.9999
h	0.7764	0.6900	0.8086	0.7375	

CE, chance of exclusion; PD, power of discrimination; h, allelic diversity values



An odd exclusion of mother, in the VWA system, found in a family, under study.

Comments: Our sample was found to be in Hardy - Weinberg equilibrium. The other statistical parameters show that this 4-plex is a robust and discriminating combined system, very useful to routine forensic casework, namely in parentage analysis.

Family studies involving 120 families (data to be published) show no disagreement with Mendelian inheritance. However in one particulary family, we detected an exclusion of the mother, in the VWA system. Such an odd exclusion is in contradiction with data from 26 other ("classical" and PCR) systems, yet confirmed by singleplex amplification of the VWA system alone and by repeated analysis with different samples from the same individuals. This problem requires further and extensive investigation, still in progress at our laboratory.

Some modifications of Primer concentrations had to be done in order to get balanced allele signals from the different PCR systems. Each new batch of Primers must be tested prior to use in routine, and concentrations adjusted, if necessary.

References: Kimpton C et al.(1995). Report on the second EDNAP collaborative STR exercise. Forensic Sci Int 71 : 137 - 152.