

**POPULATION STUDY OF 3 STR LOCI IN THE NORTH OF PORTUGAL**

M. F. Pinheiro<sup>1,2</sup>, M.L.Pontes<sup>1</sup>, M. Gené<sup>3</sup>, E. Huguet<sup>3</sup> and J. Pinto da Costa<sup>1,2</sup>

1 Medico-Legal Institute, 4050 PORTO, PORTUGAL

2 Biomedical Sciences Institute "Abel Salazar", University of Oporto

3 Forensic Genetics Laboratory. School of Medicine University of Barcelona (UB).  
28028 BARCELONA, SPAIN

**SYSTEMS AND LOCI:** HUMVWA31A (12 p 12-12 pter)  
HUMTH01 (11 p 15.5 - p 15)  
HUMFES/FPS (15 q 25-qter)

**POPULATION AND SAMPLE SIZE:**

HUMVWA31A n=263. HUMTH01 n=319. HUMFES/FPS n=307

**METHODS:**Primers :

HUMVWA31A (Kimpton et al. 1992)  
HUMTH01 (Polymeropoulos et al. 1991)  
HUMFES/FPS (Polymeropoulos et al. 1991)

PCR amplification conditions:

Singleplex amplifications in a Perkin Elmer 480 according to EDNAP recommendations (Kimpton et al. 1995).

Electrophoretic methods:

Short 5% Long Ranger gel electrophoresis. The gels were run for 2 h at 1000 V, 35 mA, 25 W, 50°C and laser power at 3 mW on the ALF DNA sequencer. The allelic ladders were constructed with alleles of known size from each locus. STR products were automatically sized by the Fragment Manager software (Pharmacia TM), using internal standard sizers with 100 and 300 bp (Pharmacia TM) and an external ladder with 50-500bp (Pharmacia TM).

**RESULTS:**

Table 1. Observed genotypes in a sample population of the North of Portugal

<b>HUMVWA31A</b>		<b>HUMTH01</b>		<b>HUMFES/FPS</b>	
Gen.	Obs.	Gen.	Obs.	Gen.	Obs.
13-14	1	6-6	9	8-10	4
13-16	1	6-7	25	8-11	1
13-17	1	6-8	20	8-12	2
14-14	2	6-9	30	8-13	1
14-15	13	6-9.3	41	10-10	31
14-16	8	6-10	2	10-11	66
14-17	13	7-7	6	10-12	52
14-18	11	7-8	19	10-13	6
14-19	10	7-9	17	10-14	2
15-15	6	7-9.3	27	11-11	49
15-16	17	8-8	2	11-12	48
15-17	15	8-9	17	11-13	15
15-18	11	8-9.3	24	12-12	26
15-19	2	8-10	2	12-13	4
16-16	8	9-9	12		
16-17	33	9-9.3	32		
16-18	20	9-10	2		
16-19	7	9.3-9.3	32		
16-20	5				
17-17	20				
17-18	23				
17-19	11				
17-21	2				
18-18	9				
18-19	9				
18-20	3				
19-19	2				
$X^2=39.63$		$X^2=15.86$		$X^2=18.23$	
d.f.=36		d.f.=15		d.f.=15	
0.350 > p > 0.300		0.400 > p > 0.350		0.260 > p > 0.250	

Table 2. Allele frequencies in a sample population of the North of Portugal

HUMVWA31A		HUMTH01		HUMFES/FPS	
Allele	Frequency	Allele	Frequency	Allele	Frequency
13	0.0057	6	0.2131	8	0.0130
14	0.1141	7	0.1567	10	0.3127
15	0.1331	8	0.1348	11	0.3713
16	0.2034	9	0.1912	12	0.2573
17	0.2623	9.3	0.2947	13	0.0423
18	0.1806	10	0.0094	14	0.0033
19	0.0817				
20	0.0152				
21	0.0038				

Table 3. Statistical parameters

	HUM VWA31A	HUM TH01	HUM FES/FPS
Heterozygosity Index (HI)	0.8213	0.8088	0.6547
Power of discrimination (PD)	0.9429	0.9223	0.8483
Chance Exclusion (CE)	0.6410	0.5810	0.4290
Essen - Möller mean value (EM)	9.49	9.57	9.69

**COMMENTS:**

The high number of alleles and the statistical parameters obtained make these markers good tools for forensic analysis. The described automated typing protocol is presently employed in our routine paternity testing and in some criminal cases.

**REFERENCES:**

- Kimpton C, Walton A, Gill P, (1992) A further tetranucleotide repeat polymorphism in the VWF gene. *Hum Mol Genet* 1:287
- Kimpton C, Gill P, D'Aloja E, Andersen JF, Bar W, Holgersson S, Jacobsen S, Johnsson V, Kloosterman AD, Lareu MV, Nellesmann L, Pfitzinger H, Phillips CP, Rand<sup>m</sup> S, Schmitter H, Schneider PM, Sternersen M, Vide MC (1995) Report on the second EDNAP collaborative STR exercise. *Forensic Sci Int* 71:137-152
- Polymeropoulos MH, Xiao H, Rath DS, Merrill CR (1991) Tetranucleotide repeat polymorphism at the human tyrosine hydroxylase gene (TH). *Nucleic Acids Res* 19:4018