THE DISTRIBUTION OF HUMTH01 POLYMORPHISM IN NORTHERN AND CENTRAL ITALY

Domenici R, Nardone M, Spinetti I, Venturi M, Bargagna M [*]
Cucurachi N, Buscemi L, Regazzi E, Ferrara SD [°]
Previderè C, Peloso G [**]
Tagliabracci A, Mencarelli R [°°]

Universities of Ancona°°, Parma°, Pavia**, Pisa*

INTRODUCTION

HUMTH01 polymorphism, first described by Edwards *et al* (1991), is due to a variation in the length of a tetrameric short tandem repeat on locus 11p15.5. Seven alleles were identified in the range 183-207 bp (Edwards *et al* 1992).
The aim of this study is to give a contribution to the knowledge of the allelic frequency distribution of this locus in Northern and Central Italy. Population samples were obtained from four Regions: Lombardy, Emilia, Tuscany and Marches. This study was made possible by the collaboration of the Legal Medicine Institutes of four Universities: Pavia (Lombardy), Parma (Emilia), Pisa (Tuscany) and Ancona (Marches).

MATERIALS AND METHODS

The population sample consisted of 588 healthy unrelated subjects: 150 resident in Pavia (45.10 N / 9.10 E), 146 resident in Parma (44.48 N / 10.20 E), 142 resident in Ancona (43.38 N / 13.30 E) and 150 all born in Pisa (43.43 N /10.23 E).
DNA typing was carried out in the laboratories of Ancona, Pisa and Parma according to the method described by Brinkmann *et al* (1992) with minor modifications. In the laboratory of Pavia, amplification products were radiolabeled by dATP^{35}S and fractionated on denaturating sequencing gels (6% PAA, 7M urea).
Alleles were assigned by means of home-made ladders consisting of human alleles (fig 1); the fragment sizes were measured on denaturating sequencing gel by comparison with a DNA sequence ladder derived from known sequences (fig 2).
An adequate number of samples were exchanged between the laboratories to test the reproducibility of the typing results.
The subsamples belonging to the four regions were tested for heterogeneity using G^2 statistics as described by Piazza *et al* (1989). They did not differ significantly from each other and were thus pooled to form a single population for further analysis.

RESULTS AND DISCUSSION

Table 1 shows the observed and expected frequencies in the four sub-samples; the Hardy-Weinberg equilibrium was satisfied. The 5 common alleles found were designated according to Edwards *et al* (1992). Rare alleles ($p < .025$) were pooled.
The heterogeneity test for allele frequency was not significant ($G^2 = 15.25$, df = 12, $P > .20$); the allelic frequencies in the sub-samples and in the pooled sample are shown in table 2 together with the standard error. These values are representative of the Northern and Central Italy, and may be used in

forensic practice. A simple comparison of these frequencies with those obtained by Edwards *et al* (1992) and Wiegand *et al* (1992) showed no apparent differences (fig. 3).
This study confirms that the HUMTH01 polymorphism is particularly suitable for forensic medicine applications, because of its information content. The exclusion chance is 0.583, the discrimination index is 0.923 and the expected heterozigosity is 0.790.

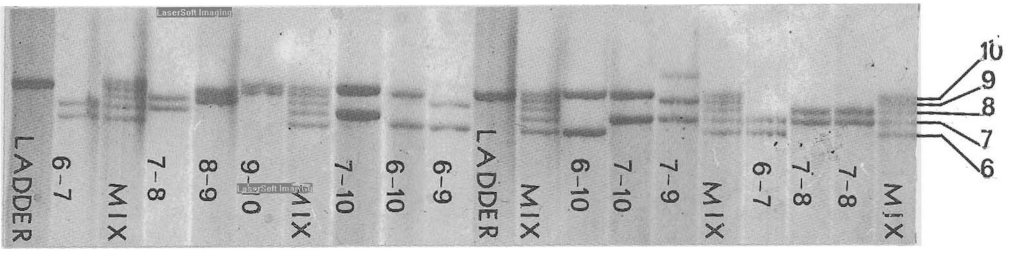

Figure 1: HUMTH01 phenotypes observed by PAA electrophoresis and silver staining

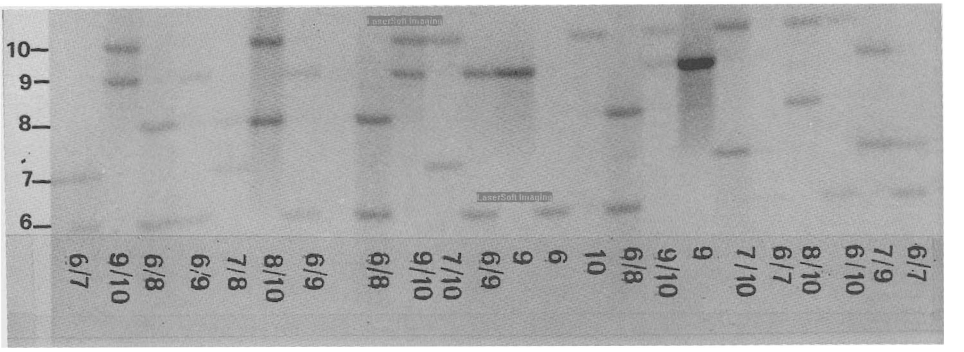

Figure 2: HUMTH01 phenotypes observed by denaturing sequencing gel

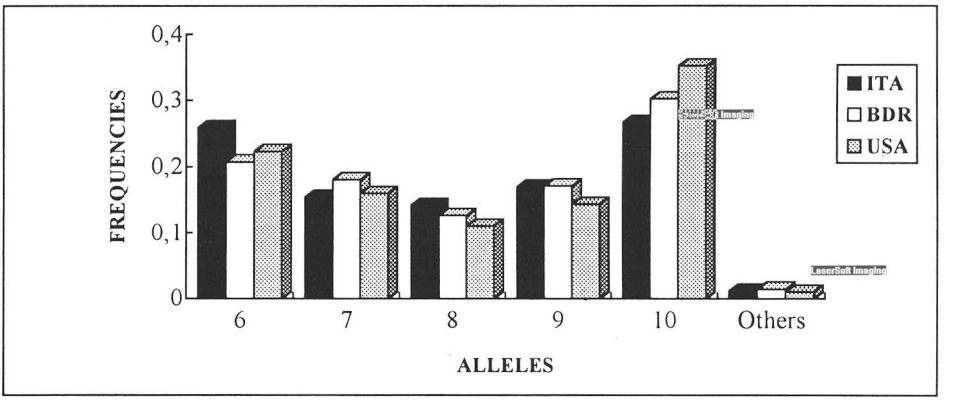

Figure 3: Comparison of our data (ITA) with data from Wiegand (BDR) and from Edwards (USA)

Phenotypes	Ancona		Parma		Pavia		Pisa		Pooled	
	Obs	Exp	Obs	Exp	Obs	Exp	Obs	Exp	Obs	Exp
6	9	8.14	17	15.45	8	7.94	6	8.64	40	39.29
6-7	10	10.77	14	11.71	12	10.35	11	13.20	47	46.79
6-8	10	10.54	11	10.41	11	10.81	15	10.32	47	42.91
6-9	12	12.93	13	15.62	12	12.19	15	10.56	52	51.44
6-10	17	17.00	22	25.38	17	19.55	17	18.96	73	80.91
6-Others	1	0.48	1	0.98	1	0.23	2	1.68	5	3.36
7	3	3.57	1	2.22	2	3.38	4	5.04	10	13.93
7-8	7	6.97	4	3.95	9	7.05	8	7.88	28	25.55
7-9	9	8.56	6	5.92	6	7.95	4	8.07	25	30.63
7-10	13	11.25	10	9.62	14	12.75	22	14.48	59	48.17
7-Others	0	0.32	0	0.37	0	0.15	2	1.28	2	2.00
8	4	3.41	1	1.75	4	3.68	2	3.08	11	11.72
8-9	8	8.37	2	5.26	7	8.31	6	6.31	23	28.09
8-10	11	11.00	12	8.55	12	13.32	10	11.32	45	44.18
8-Others	0	0.31	1	0.33	0	0.15	0	1.00	1	1.84
9	6	5.13	7	3.95	5	4.68	2	3.23	20	16.84
9-10	12	13.50	12	12.82	18	15.02	13	11.59	55	52.97
9-Others	1	0.38	1	0.49	0	0.17	2	1.03	4	2.20
10	9	8.88	11	10.42	12	12.04	8	10.40	40	41.65
10-Others	0	0.50	0	0.80	0	0.28	1	1.84	1	3.46
Others	0	0.01	0	0.02	0	0.00	0	0.08	0	0.07
Total	142	142.00	146	146.00	150	150.00	150	150.00	588	588.00
χ^2		3.875		11.477		6.610		16.130		12.186
df = 15		P>.99		P>.70		P>.95		P>.30		P>.50

Tab.1: HUMTH01 phenotype frequencies in the sub-samples and in the pooled sample

Allele	Ancona	Parma	Pavia	Pisa	Pooled
6	.2394	.3253	.2300	.2400	.2585±.0128
7	.1584	.1233	.1500	.1833	.1539±.0105
8	.1549	.1096	.1567	.1433	.1412±.0102
9	.1901	.1644	.1767	.1467	.1692±.0109
10	.2500	.2671	.2833	.2633	.2662±.0129
Others	.0070	.0103	.0033	.0233	.0111±.0030

Tab.2: HUMTH01 allele frequencies in the sub-samples and in the pooled sample

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