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ALLELE FREQUENCY OF THE HUMVWA31 LOCUS IN ITALY. REPORT OF THE GEFI COLLABORATIVE STUDY "ANCONA 1"

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Introduction

Since 1991 the Italian Group for Forensic Haemogenetics (GEFI) promotes collaborative studies on DNA polymorphisms with the aim of standardizing experimental protocols, ensuring exchange of data and experience among participating forensic haemogenetics laboratories, and setting up controlled databases for individual identification and paternity testing purposes. The last collaborative exercise developed by GEFI and completed in 1995 involved HumFES/FPS and HumVWA31 loci. Results on the HumVWA locus[1], a tetranucleotide marker located in an intron of the von Willebrand factor gene on the short arm of chromosome 12, are described here.

Materials and methods

Twenty-four laboratories participated in this exercise on the basis of a protocol involving the characterization of at least 50 unrelated, locally residing subjects, and two common blind controls.

Each laboratory received a dossier containing the most important information on this marker and suggestions for specific amplification and electrophoresis protocols[2]. An aliquot of a specific ladder containing 8 alleles (11, 14-20) to perform allele typing by side-to-side comparison was also supplied free of charge. Each laboratory was, however, allowed choose alternatively well-known amplification, electrophoresis and detection protocols because they were already used for routine analysis.

Results and discussion

Twenty-two laboratories from 13 Regions throughout Italy sent results (91.66%). Genotype characterization of the two control bloodstains was correctly carried out by all laboratories.

Genotype heterogeneity among samples was assayed by a two-way contingency table of 22 columns (laboratories) by 13 rows (12 individual genotypes, embracing 80% of the total population, plus the cumulative group *others).* The grand total chi square (263.84) was not significant (252 d.f. P=0.292) of heterogeneity, although one sample showed a rather large individual contribution. Hardy-Weinberg equilibrium was tested in each subsample by reducing the system to six allele frequencies (the six most frequent alleles included 98.4% of total sampled

genes). One sample, that already marked by maximum genotype diversity, was very far from equilibrium (P<.001) and was excluded from further computations; a second laboratory had too high an accordance (P>.999). All other samples showed probability values between .05 and .95.

The final database was composed of 2,465 individuals, including 37 different genotypes and 10 alleles. The most frequent genotype (16/17) had a frequency of 13.0%, followed by genotype 17/18 (10.9%). Ninety per cent of the population included 15 genotypes and 6 alleles (14-19). About 4% of the population had rare genotypes (frequency <1%). Table 1 shows the allele counts in the 13 Regions, arrayed in North-South order.

Italian Region	N. Ind.	all. 11	all. 13	all. 14	all. 15	all. 16	all. 17	all. 18	all. 19	all. 20	all. 21	N. genes
Friuli	125	0	0	36	22	61	71	52	7	1	0	250
Veneto	120	0	0	25	33	43	87	36	15	1	0	240
Lombardy	356	0	1	72	88	142	195	156	50	8	0	712
Emilia	244	1	0	40	64	93	141	108	37	3	1	488
Liguria	306	0	2	56	78	140	166	113	50	7	0	612
Tuscany	250	0	2	41	55	111	140	111	34	5	1	500
Marches	178	0	0	32	27	73	102	80	33	8	1	356
Umbria	109	0	0	17	17	51	69	46	14	4	0	218
Latium	374	0	3	70	86	160	212	152	52	13	0	748
Sardinia	86	0	1	19	16	32	55	30	17	2	0	172
Campania	52	0	1	7	12	18	28	29	5	4	0	104
Puglia	52	1	0	17	10	19	29	15	10	3	0	104
Sicily	213	2	2	47	44	95	130	70	33	3	0	426
TOTAL	2465	4	12	479	552	1038	1425	998	357	62	3	4930

Seven alleles (14-20) were common to all Regions and showed an overall frequency higher than 1%; three other rare alleles (11, 13, 21) were identified in some Regions. The expected heterozygosity of the locus was 80.4%. The heterogeneity of gene frequency among Regions was tested by a contingency table applied to 7 allele classes. Counts of alleles 11 and 13 were summed to allele 14, and counts of allele 21 was summed to allele 20. There was a modest level of genetic differentiation (chi square = 94.59, 72 d.f., P = .038). This was confirmed by computing the variance of gene frequency (F_{St}), based on observed and expected heterozygosity in each Region and in the total sample. Its value was 0.0045.

We computed the Italian allele frequencies (excluding Sardinia) by weighting the regional frequencies by total resident population (1993 ISTAT census data). Figure 1 shows the results in the form of a column chart.

The Italian allele frequencies of the HumVWA31 locus found in this study are similar to those reported for other Caucasian populations[3, 4].

This work validates the use of the HumVWA31 marker in forensic and population analyses and

contributes to standardizing analytical methods in our forensic haemogenetic laboratories.

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Figure 1. HumVWA31 allele frequencies in Italians