SPANISH POPULATION DATA ON SEVEN LOCI (D1S80, D17S5, HUMTH01, HUMVWA, ACTBP2, D21S11 and DQA1): EQUILIBRIUM AND INDEPENDENCE

M. Lorente[1], J.A. Lorente[1], J.C. Alvarez[1], B. Budowle[2] &
E. Villanueva[2]

[1] University of Granada, Dept. of Forensic Medicine, Granada, Spain

[2] FSRTC - Research Unit. Laboratory Division. FBI Academy, Quantico, Va, USA.

INTRODUCTION

Amplification by the Polymerase Chain Reaction (PCR) and subsequent electrophoresis of the amplified products have become a useful approach for typing variable number of tandem repeats (VNTR) loci. Currently there is an increasingly number of data on VNTR and especially on the STR loci (i.e., Edwards et al, 1992), regarding their allele frequencies and genotype distribution in various population.

For the genetics markers, such as VNTRs & STRs, in identity testing, it is desirable to collect allele/genotype data from relevant populations, so that the forensic scientist can provide a guideline to estimate of the rarity of a genetic profile.

MATERIALS AND METHODS

Population: 120 unrelated Spanish Caucasians from Andalucia (South of Spain)
Statistical tests:
1. Hardy-Weinberg equilibrium:
1.1. Goodness-of-fit chi-square test, based on the total number of homozygotes and heterozygotes
1.2. Likelihood ratio test, comparing the frequencies of each specific genotype with their expectations under the Hardy-Weinberg assumptions.
1.3. Guo-Thompson's exact test, 1000 replications.
2. Gametic phase equilibrium:
2.1. Interclass correlation test (Karlin et al, 1991)
2.2. Test of the observed variance (Sk^2) of the number of heterozygotes classes.

RESULTS AND DISCUSSION

Interclass correlation test (Karlin et al, 1981)

	RHO	P
D1S80-HUMTH01	0.045760	0.323
D1S80-D17S5	-0.214510	0.644
D1S80-ACTBP2	-0.037771	0.436
D1S80-HUMVWA	-0.023085	0.605
D1S80-D21S11	0.011053	0.786
HUMTH01-D21S11	-0.029091	0.527
HUMTH01-ACTBP2:	-0.040358	0.363

Interclass correlation test (continuation)

HUMTH01-VWA:	-0.012521	0.784
HUMTH01-D17S5:	0.014939	0.764
D17S5-D21S11:	0.025888	0.558
D17S5-ACTBP2:	-0.183110	0.704
D17S5-HUMVWA:	-0.051937	0.256
DQA1-D1S80:	-0.022010	0.615
DQA1-D17S5:	-0.017704	0.692
DQA1-D21S11:	0.010995	0.835
DQA1-ACTBP2:	-0.002433	0.958
DQA1-HUMTH01:	-0.014330	0.753
DQA1-HUMVWA:	0.007176	0.881
ACTBP2-HUMVWA:	0.061216	0.183
ACTBP2-VWA:	0.061216	0.183
ACTBP2-D21S11:	-0.008573	0.856
HUMVWA-D21S11:	-0.005315	0.906

Results of tests used for Hardy-Weinberg equilibrium

	Chi-square	Likelihood	Guo-Thompson
HUMTH01	0.225	0.097	0.070
HUMVWA	0.005	0.434	0.349
ACTBP2	0.017	0.840	0.008
D21S11	0.547	0.008	0.115
HLA-DQA1	0.554	0.458	0.597
D1S80	0.708	0.999	0.508
D17S5	0.249	0.394	0.352

Parameters of Forensic interest:

	Discrim.Pow.	# combinat.	Pater. Ind.
HUMTH01	0.9333	2380	74.28%
HUMVWA	0.9415	1281	75.03%
ACTBP2	0.9928	279577	90.48%
D21S11	0.9307	4068	74.47%
HLA-DQA1	0.9343	1281	74.46%
D1S80	0.9208	83385	79.44%
D17S5	0.9676	4068	82.89%
Total	0.999999	-	99.90

According to these results, all but one loci (ACTBP2) are in equilibrium and independent in all cases. A test for independence for ACTBP2 can be problematic because of the allele designation strategy studied. The even numbered alleles generally represent discrete data, while the odd numbered alleles are drived from binned data.

Classical tests are more prone to be affected by the sample characteristics. In this study, chi-square test is significative for the HUMVWA locus because the ratio homozygotes/heterozygotes is higher than expected. Nevertheless, both the likelihood ratio test amd the Guo-Thompson's one are not significative. Therefore, we strongly recommend to use more than one test to check the Hardy-Weinberg equilibrium and the gametic phase equilibrium.

In conclussion, these seven loci are useful for identity testing in criminalistic and paternity cases.

REFERENCES

Chakraborty R, Fornage M, Guegue R. Boerwinkle E (1991) Population genetics of hypervariable loci: analysis of PCR-based VNTR polymorphism within a population. In: T. Burke, G. Dolf, A.J. Jeffreys, and R. Wolff (eds.), DNA fingerprinting: approaches and applicatios, Birkhauser Verlag, Berlin, pp. 127-143

Edwards A, Hammond H, Jin L, Caskey CT, Chakraborty R (1992) Genetic variation at five trimeric and tetrameric loci in four human population groups. Genomics 12: 241-253

Guo SW, Thompson EA (1992) Performing the exact test of Hardy-Weinberg proportion for multiple alleles. Biometrics 48: 361-372

Karlin S, Cameron EC, Williams PT (1981) Sibling and parent-offspring correlation estimation with variable family size. Proc. Natl. Acad. Sci. U.S.A. 78: 2664-2668

Nei M, Roychoudhury AK (1974) Sampling variances of heterozygosity and genetic distance. Genetics 76: 379-390

Nei M (1978) Estimation of average and genetic distance from a small number of individuals. Genetics 89: 583-590

Weir BS (1992) Independence of VNTR alleles defined by fixed bins. Genetics 130: 873-887

This work was supported by a grant of the Spanish Ministry of Education and Science (DGICYT) PB93-1155