

## STUDY OF D1S80 LOCUS POLYMORPHISM IN THE NORTH OF PORTUGAL

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### INTRODUCTION

The Polymerase Chain Reaction (PCR, Saiki et al, 1985) can enhance the detection and offers a viable alternative to RFLP (restriction fragment length polymorphism) analysis of known VNTR loci (Budowle et al, 1991) particularly in forensic situations where limited amounts of DNA are available.

The Locus D1S80, a VNTR locus, is a very useful genetic marker system for forensic DNA analysis. The polymorphism has been localized in the short arm of chromosome 1 and consists of VNTR repeats each of them about 16 bp long (Nakamura et al, 1988; Kasai et al, 1990). This work presents data for the locus D1S80 from the North of Portugal.

### MATERIAL AND METHODS

Blood samples were collected from 227 unrelated individuals from paternity tests. DNA was extracted by a method previously described by Singer-Sam and col. (1989) using a chelating resin. Some of the samples were extracted using a phenol - chloroform procedure.

Amplification of D1S80 (pMC118) was achieved using the primers described by Kasai et al (1990). The primers were obtained from Cetus and used under conditions recommended by the manufacturer. Amplification was done by the method described by Budowle et al (1990, 1991) with slight modifications, according to Cetus recommendations.

Typing was performed using Phast Gels 10-15 (Pharmacia LKB) and silver staining with the conditions described in Barros et al (1992) or running an 2.5% agarose (FMC) gel in TBE buffer at 4V/cm for 6-7 hours and visualized by staining with ethidiumbromide using the same ladder (Cetus).

### RESULTS AND DISCUSSION

In this study we have found 20 D1S80 alleles (Table 1).

There were no differences in results obtained by the two typing methods used in this work.

Table 1. Frequencies of the D1S80 alleles found in the population of the North of Portugal

Allele	Frequency	Allele	Frequency
14	0.0022	26	0.0088
17	0.0066	27	0.0088
18	0.3084	28	0.0374
19	0.0088	29	0.0881
20	0.0176	30	0.0066
21	0.0264	31	0.0573
22	0.0352	32	0.0110
23	0.0066	34	0.0066
24	0.3084	36	0.0154
25	0.0308	37	0.0088

Total 20

As there are many alleles and thus a great number of genotypes, for chi-square calculation the alleles were pooled into five groups (Budowle et al, 1991). We found that there was no significant deviation from the Hardy-Weinberg equilibrium ( $\chi^2 = 4.4736$ ;  $df = 10$ ;  $0.90 < P < 0.95$ ).

Table 2. Distribution of observed and expected D1S80 genotypes from 227 individuals

Genotype	Observed	Expected	Genotype	Observed	Expected	Genotype	Observed	Expected
14-18	1	0.3084	19-19	1	0.1762	24-30	1	0.9251
17-20	1	0.0528	20-21	1	0.2114	24-31	7	8.0176
17-24	1	0.9251	20-24	3	2.4670	24-32	1	1.5418
17-34	1	0.0198	20-29	2	0.7048	24-34	1	0.9251
18-18	21	21.5859	21-21	1	0.1586	24-36	1	2.1586
18-19	2	1.2335	21-24	3	3.7004	24-37	1	1.2335
18-20	1	2.4669	21-29	1	1.0573	25-28	2	0.5242
18-21	4	3.7004	21-31	1	0.6872	25-29	2	1.2335
18-22	4	4.9339	22-22	1	0.2819	25-31	3	0.8018
18-23	1	0.9251	22-24	6	4.9339	26-31	1	0.2291
18-24	48	43.1718	22-28	1	0.5991	26-37	1	0.0352
18-25	1	4.3172	22-31	1	0.9163	27-29	1	0.3524
18-27	1	1.2335	22-36	2	0.2467	28-29	3	1.4978
18-28	8	5.2423	23-31	2	0.1718	28-31	1	0.9736
18-29	13	12.3348	24-24	21	21.5859	29-31	1	2.2907
18-30	1	0.9251	24-25	6	4.3172	29-32	1	0.4405
18-31	8	8.0176	24-26	2	1.2335	29-36	2	0.6167
18-32	3	1.5412	24-27	2	1.2335	29-37	1	0.3524
18-34	1	0.9251	24-28	2	5.2423	30-36	1	0.0625
18-37	1	1.2335	24-29	13	12.3348	31-36	1	0.4009

The most frequent alleles are the 18 and the 24 repeats with the same frequency (0.3084). All other alleles have frequencies less than 0.1. Table 2 gives the distribution of the observed and expected genotypes found in our population. In total 60 genotypes were observed. The frequency distribution of the alleles in the North of Portugal is slightly different to that found in other Caucasian populations (Budowle et al, 1991; Thyman et al, 1993; Schnee-Griese et al, 1993), even when compared with very close geographical populations such as NW Spain and Central Portugal (Lareu et al, in press). In our population the allelic frequencies of the two most common alleles are equal, while in those there is a prevalence of allele 24. The theoretical a priori chance exclusion value (C.E.) is 0.6174. From genotype data, the power of discrimination (P.D.) according to the equation suggested by Fisher (1951) is 0.9327.

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