

POPULATION GENETICS OF D1S80, HUMVWFA31/A AND HUMF13A1 FROM PORTUGAL AND GOA(INDIA)

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INTRODUCTION

Analysis of variable number of tandem repeats (VNTR) loci is widely used for individualization in forensic testing. VNTR locus D1S80 is successfully amplified by PCR. STR loci HUMVWFA31/A and HUMF13A1 are also two informative PCR based identification systems. The aim of this paper is to provide information about estimate allele frequencies for the three loci in **Portuguese and Goa (India) populations**, due to the Portuguese presence in Goa during four centuries.

MATERIAL AND METHODS

Unrelated Portuguese blood samples from paternity cases were studied for population analysis: D1S80-187 samples, HUMVWFA31/A-114 samples and HUMF13A1- 108 samples. These samples mainly from the South of Portugal, include Continental as well as Madeira and Açores Islands. Goa blood samples were obtained from unrelated persons from Portuguese origin living in Goa: D1S80-120 samples, HUMVWFA31/A-102 samples and HUMF13A1-95 samples.

DNA was extracted from blood by **Chelex method**. **D1S80 amplification** was carried out under the following conditions: 95°C 60sec, 65°C 60sec, 70°C 8min, 30 cycles (1). **HUMVWFA31/A and HUMF13A1 amplifications** were performed according to the EDNAP protocol(2), with slight modifications, using 0.5 uM of each primer in a 50 ul reaction volume, of which one primer was labelled with fluorescein. Amplification conditions: 94°C 45sec, 52°C 60sec, 72°C 60sec, 30 cycles.

D1S80 and the two STR systems were simultaneously analysed on an Automatic Laser Fluorescent (A.L.F.) **DNA sequencer** in a 4% hydrolink gel. Allelic ladders were used as external standards and three internal size markers (114bp, 402bp, 821bp) were added into each lane (3).

RESULTS AND DISCUSSION

In the Portuguese Population, we have obtained 27 alleles for **D1S80** locus, ranging from 15-48 number of repeats and in Goa Population 19 alleles, ranging from 14-36. The allele frequency differences in both populations are the allele 14, 16 and 19 presence and a higher 31 allele frequency in Goa Population (Table1, Fig.1).

TABLE 1. D1S80 allele frequencies in Portuguese and Goa populations

Allele	Port	Goa	Allele	Port	Goa	Allele	Port	Goa
14	-	0.0042	22	0.0321	0.0471	30	0.0107	0.0083
15	0.0027	0.0042	23	0.0107	0.0083	31	0.0267	0.1083
16	-	0.0167	24	0.3449	0.3167	32	0.0027	-
17	0.0027	0.0042	25	0.0401	0.0333	33	0.0027	-
18	0.2593	0.3208	26	0.0133	0.0083	34	0.0054	-
19	-	0.0208	27	0.0160	0.0125	35	-	0.0083
20	0.0160	-	28	0.0668	0.0417	36	0.0080	0.0042
21	0.0348	0.0125	29	0.0695	0.0250	37-48	0.0349	-

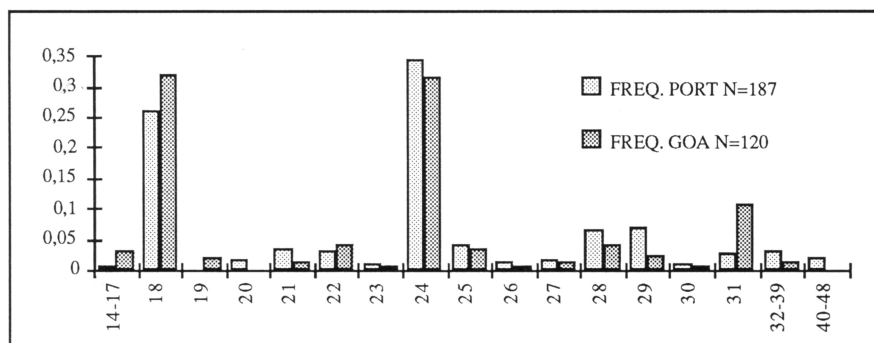


Fig.1. D1S80 allele frequency distribution in Portuguese and Goa Populations

For **HUMVWFA31/A**, Portuguese and Goa Populations are in good agreement withHWE (Port- $X^2=6.766$, d.f.=10, $0.70 < p < 0.80$; Goa- $X^2=13.41$, d.f.=10, $0.20 < p < 0.30$), showing frequency differences in alleles 14 and 19. **HUMF13A1** is also in good agreement for HWE (Port $X^2=11.99$, d.f.=10, $0.20 < p < 0.30$; Goa $X^2=6.039$, d.f.=10, $0.80 < p < 0.90$), with higher frequencies in alleles 14 to 17 and mainly in allele 3.2 in Goa Population (Table2, Fig.2).

TABLE 2. HUMVWFA31/A and HUMF13A1 allele frequencies in Portuguese and Goa Populations

HumVWFA31/A			HumF13A1		
Allele	Port	Goa	Allele	Port	Goa
12	0.0044	-	3,2	0.0741	0.1947
13	-	0.0098	4	0.0417	0.0474
14	0.0746	0.1373	5	0.2222	0.3368
15	0.1272	0.0833	6	0.2870	0.1684
16	0.2149	0.2255	7	0.3056	0.1895
17	0.2632	0.3088	8	0.0185	0.0053
18	0.1667	0.1471	9	0.0370	-
19	0.1140	0.0539			
20	0.0263	0.0343	14	-	0.0158
21	0.0087	-	15	0.0093	0.0263
			16	0.0046	0.0105
			17	-	0.0053

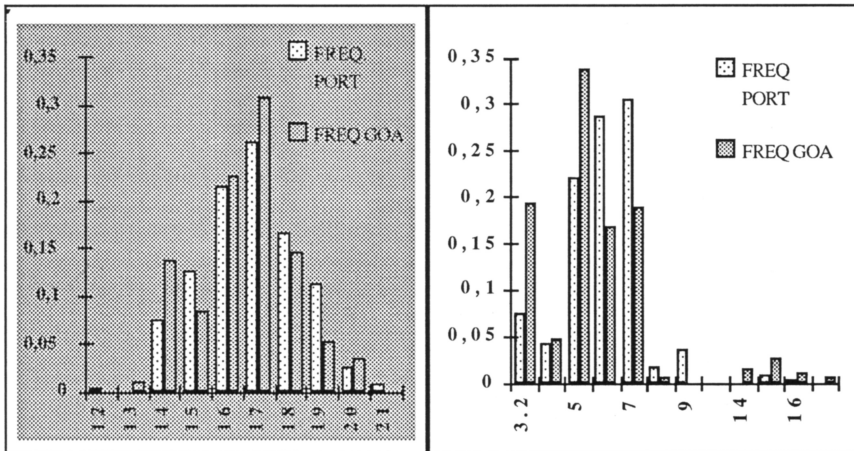


Fig.2. HumVWFA31/A(right) and HumF13A1(left) allele frequency distribution in Portuguese and Goa Populations

In conclusion, Goa population database has been established for the VNTR locus D1S80 and two STR loci HumVWFA31/A and HumF13A1, and although from Portuguese origin, it is in agreement with other Indian populations studied.

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