ELSEVIER

Forensic Science International 129 (2002) 72–74

Forensic Science International

www.elsevier.com/locate/forsciint

Announcement of population data

Population data of five STRs in three regions from Portugal

Ana Teresa Fernandes, António Brehm[*]

Human Genetics Laboratory, Centre of Biological Sciences, University of Madeira, Penteada, 9000 Funchal, Portugal

Received 25 March 2002; accepted 31 May 2002

Abstract

Allele and haplotype frequencies of five chromosome STR loci (CD4, TPO, FES, TH01 and VWA) were determined for unrelated males throughout Portugal. This report presents STR data for three separate regions of Portugal, being the first time that data on the south of the country is presented. This study reveals that the three regions from Portugal are not genetically homogeneous. The north of Portugal presents significant differences in the CD4 locus, when compared with the other two populations. When compared with Madeira and Açores, the three regions show a different behavior at TPO and VWA loci.

Keywords: STRs; Population data; Portugal; TPO; VWA; FES; CD4; TH01

Population: In this study we used samples from 120 healthy unrelated males living in the region north of river Douro (north Portugal (NP)), 127 living between river Douro and river Tagus (center of Portugal (CP)), and 151 living in the region south of river Tagus (Alentejo and Algarve, south of Portugal (SP)). In this survey we deliberately avoided the inclusion of individuals from Lisbon. Also, all the individuals as well as both their ancestors were living in the same region at least back to three generations. This way we got a fairly good coverage of the whole territory.

DNA extraction: From the leukocyte fraction using a Chelex based method.

PCR: Conditions according to: TPO [1], CD4, TH01 and VWA [2], FES [3].

Typing: By polyacrylamide gel electrophoresis with visualization by silver staining.

Results: Presented in Table 1 as allele frequencies.

Analyses of data: GENEPOP [4], haplotype and gene diversity according to Nei [5]. Population comparisons were done using Arlequin [6].

Access to the data: http://www.uma.pt/ccbg/LGH/STRPortugal.

Observations: The three populations studied are in Hardy–Weinberg equilibrium. Overall the population from the center of Portugal has less variability than the other two regions (CD4, TPO, FES). At the CD4 locus, the north of Portugal is statistically different from the center and south (exact genotypic differentiation test, $P < 0.05$). When these three regions are compared to populations from Madeira [7], the TPO locus shows significant differences with the center and south (exact test, $P < 0.005$), and the VWA with the north and south (exact test, $P < 0.03$). If the regions are compared to the Açores population [8], there is a difference at the VWA locus with the south (exact test, $P < 0.03$).

---

[*] Corresponding author. Tel.: +351-1291-705383; fax: +351-1291-705399.
E-mail address: brehm@uma.pt (A. Brehm).

0379-0738/02/$ – see front matter
PII: S0379-0738(02)00202-5

Table 1
Allele frequencies and gene diversity values at five STR loci in (a) NP, (b) CP and (c) SP

	CD4 ($N = 120$)	TPO ($N = 111$)	FES ($N = 93$)	TH01 ($N = 96$)	VWA ($N = 98$)
(a) NP					
5	0.454				
6	0.225			0.224	
7	0.013	0.005		0.130	
8	0.007	0.554		0.104	
9	0.004	0.131	0.011	0.271	
9.3				0.260	
10	0.287	0.063	0.323	0.010	
11	0.013	0.207	0.387		
12		0.041	0.242		
13	0.004		0.038		
14					0.143
15					0.102
16					0.235
17					0.291
18					0.173
19					0.046
20					0.010
Ho	0.642	0.640	0.732	0.802	0.867
He	0.663	0.630	0.690	0.785	0.801
P	0.278	0.253	0.108	0.859	0.236
CE	0.373	0.387	0.398	0.540	0.580
PD	0.821	0.828	0.839	0.917	0.929
	CD4 ($N = 127$)	TPO ($N = 125$)	FES ($N = 121$)	TH01 ($N = 111$)	VWA ($N = 122$)
(b) CP					
5	0.374				
6	0.283			0.214	
7				0.150	
8		0.576	0.017	0.150	
9		0.100		0.173	
9.3				0.300	
10	0.299	0.044	0.293	0.014	
11	0.031	0.252	0.335		
12	0.012	0.028	0.322		
13			0.033		
14					0.082
15					0.143
16					0.258
17					0.270
18					0.164
19					0.066
20					0.016
Ho	0.669	0.576	0.702	0.757	0.861
He	0.692	0.594	0.700	0.786	0.805
P	0.476	0.843	0.171	0.535	0.372
CE	0.389	0.339	0.407	0.553	0.583
PD	0.841	0.781	0.847	0.923	0.933
	CD4 ($N = 151$)	TPO ($N = 149$)	FES ($N = 145$)	TH01 ($N = 148$)	VWA ($N = 141$)
(c) SP					
5	0.364				
6	0.272			0.230	
7		0.007		0.162	
8	0.003	0.517	0.003	0.162	
9	0.007	0.094	0.003	0.203	
9.3				0.233	

Table 1 (*Continued*)

	CD4 ($N = 120$)	TPO ($N = 111$)	FES ($N = 93$)	TH01 ($N = 96$)	VWA ($N = 98$)
10	0.318	0.050	0.293	0.010	
11	0.030	0.282	0.324		
12	0.007	0.050	0.328		
13			0.048		
14					0.096
15					0.106
16					0.213
17					0.255
18					0.202
19					0.103
20					0.025
Ho	0.609	0.664	0.710	0.818	0.816
He	0.694	0.642	0.702	0.818	0.820
P	0.122	0.280	0.334	0.970	0.224
CE	0.402	0.382	0.409	0.568	0.616
PD	0.843	0.818	0.849	0.928	0.941

N : number of samples; Ho: observed heterozygosity; He: expected heterozygosity; PD: power of discrimination; CE: a priori chance of exclusion; PD: power of exclusion; P: Hardy–Weinberg equilibrium, exact test based on more than 2000 shufflings, for S.E. < 0.01.

References

[1] L. Gusmão, M.J. Prata, A. Amorim, The STR systems hTPO: population, Int. J. Legal Med. 108 (1995) 167–169.

[2] L. Gusmão, M.J. Prata, A. Amorim, F. Silva, I. Bessa, Characterization of four tandem repeat loci (TH01, VWA31/A, CD4 and TP53) in north Portugal, Hum. Biol. 69 (1997) 31–41.

[3] M.H. Polymeropoulos, D.S. Rath, H. Xiao, C.R. Merril, Tetranucleotide repeat polymorphism at the human c-*fes/fps* proto-oncogene (FES), Nucleic Acids Res. 19 (1991) 4018.

[4] M. Raymond, F. Rousset, GENEPOP (version 1.2): populations genetics software for exact tests and ecumenicism, J. Hered. 86 (1995) 248–249.

[5] M. Nei, Estimation of average heterozygosity and genetic distance from a small number of individuals, Genetics 89 (1978) 583–590.

[6] S. Schneider, J.-M. Kueffer, D. Roessli, L. Excoffier, Arlequin (version 1.1): A Software for Population Genetic Data Analysis, Genetics and Biometry Laboratory, University of Geneva, Switzerland, 1997.

[7] A.T. Fernandes, A. Brehm, C. Alves, L. Gusmão, A. Amorim, Genetic Profile of the Madeira Archipelago Population Using the New PowerPlex® 16 System Kit, Forensic Sci. Int., in press.

[8] R. Velosa, A.T. Fernandes, A. Brehm, Genetic Profile of the Açores Archipelago Population Using the New PowerPlex® 16 System Kit, Forensic Sci. Int., submitted for publication.