South Portugal population genetic analysis with 17 loci STRs

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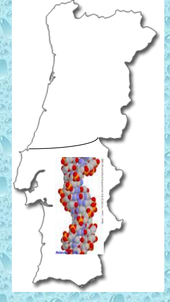

Introduction

- In our laboratory it was necessary to establish a statistically significant allele frequencies population database, from south Continental Portugal residents for further reliable statistical analysis in forensic cases.
- Genetic analysis of 17 loci, CSF1PO, D3S1358, D5S818, D13S317, D16S539, D18S51, D21S11, vWA, FGA, TH01, TPOX, D2S1338, D19S433, Penta E and Penta D, and the segment of X-Y homologous gene Amelogenin locus was performed with AmpF1STR® Identifiler™ (Applied Biosystems) and Geneprint Powerplex®16 (Promega Corporation, Madison, WI USA) are routinely used in our laboratory.

Material and Methods

- Oral swabs and blood samples were obtained from 2723 unrelated south Continental Portugal residents, 2445 caucasians, 102 Angolans and 176 individuals from Cabo Verde, after informed consent used in forensic and paternity testing studies.
- DNA was extracted using Chelex™ 100 resin method [1].The amplification conditions were identical to those proposed by the manufacturers. All amplifications were carried out in a 9700 PE® Applied Biosystems thermocycler [2] [3].
- The amplified products were analysed by capillary electrophoresis using the Abi Prism 3100 DNA sequencer (Applied Biosystems). Alleles were typed using Genescan®Analysis v 3.7 and Genotyper® software.
- Quality control and proficiency testing for these systems have been carried out for the GEP-ISFG working group and ESWG.
- Allele frequencies for each locus were calculated with SPSS 12.0 for windows, Hardy-Weinberg equilibrium, expected heterozygosity (He), observed Heterozygosity were obtained involving the Popgene version 1.32 [6], Power of Discrimination (PD) and the Power of Exclusion (PE) were calculated with MicrosoftExcel for Windows.

Results

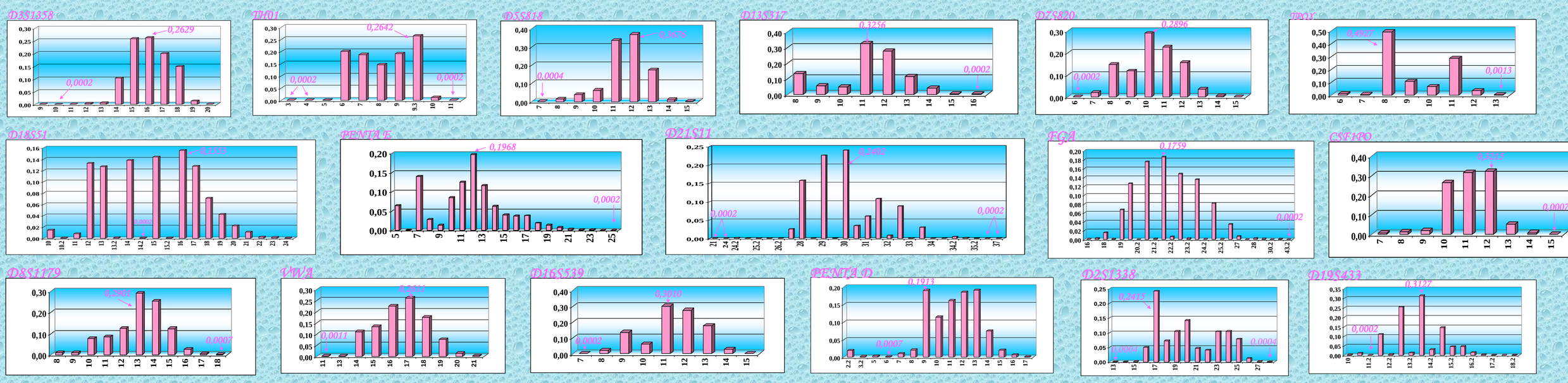

Figure 1- Allele frequencies distribution of the 17 loci analysed

Table 1-Statistical Parameteres

	D3S1358	TH01	D21S11	D18S51	PENTA E	D5S818	D13S317	D7S820	D16S539	CSF1PO	PENTA D	vWA	D8S1179	TPOX	FGA	D2S1338	D19S433
MF	0.0010	0.0010	0.0011	0.0011	0.0012	0.0010	0.0010	0.0010	0.0010	0.0010	0.0011	0.0011	0.0011	0.0009	0.0011	0.0011	0.0011
P	0.8666	0.9273	0.7492	0.3534	0.0776	0.9823	0.8425	0.9211	0.8740	0.7198	0.0007	0.0003	0.0824	0.4949	0.8648	0.0009	0.9979
Hom	0.2185	0.2093	0.1755	0.1285	0.1197	0.2916	0.2303	0.2005	0.2339	0.2629	0.1583	0.1891	0.1972	0.3448	0.1351	0.1432	0.1829
H0	0.7815	0.7907	0.8245	0.8715	0.8803	0.7084	0.7697	0.7995	0.7661	0.7371	0.8417	0.8109	0.8028	0.6552	0.8649	0.8568	0.8171
HE	0.7905	0.7979	0.8400	0.8786	0.8910	0.7176	0.7780	0.8031	0.7794	0.7233	0.8467	0.8148	0.8080	0.6582	0.8652	0.8715	0.7989
PD	0.9236	0.9276	0.9555	0.9731	0.9773	0.8720	0.9191	0.9332	0.9177	0.8723	0.9576	0.9399	0.9378	0.8320	0.9668	0.9711	0.9332
PE	0.5856	0.5946	0.6844	0.7531	0.8422	0.4766	0.5772	0.6126	0.5723	0.4735	0.6916	0.6311	0.6272	0.4143	0.7276	0.7449	0.6135

MF:Minimal Frequencies; P: -chi square test, for Hardy-Weinberg equilibrium Hom Homozigosity; HO.Observed Heterozigosity; HE: Expected Heterozigosity; PD:Power of Discrimination; PE: Power of exclusion

Table 2- Statistical tests for comparision between the subpopulations of our sample

	D3S1358	TH01	D21S11	D18S51	PENTA E	D5S818	D13S317	D7S820	D16S539	CSF1PO	PENTA D
χ^2	0.0272	0.0001	0.0000	0.0000	0.0000	0.0002	0.9145	0.0012	0.7171	0.7128	0.0000
G test	0.1844	0.0002	0.0000	0.0000	0.0727	0.0018	0.9517	0.0161	0.7297	0.4268	0.04286
	vWA	D8S1179	TPOX	FGA	D2S1338	D19S433					
χ^2	0.0000	0.0935	0.3841	0.0000	0.0000	0.0000					
G test	0.0009	0.6740	0.8134	0.0000	0.0327	0.0000					

Discussion and Conclusions:

- The combined Power of Discrimination for the 17 loci was > 0.9999999999999999, and the combined probability of exclusion was 0.99999999973 [4]
- In our sample were detected smaller alleles and microvariants tha are not present in Northern Portugal [5].
- Concerning Hardy-Weinberg equilibrium genotipe deviations ($p>0.01$) were observed in the systems PENTA D, vWA, and D2S1338. This is probably due to the heterogeneity of this population with the presence of alleles only found in individuals of African Origin. The southern Portugal since long time is a region subjected to immigrations that could affect allele frequencies in the population wich do not remain unchanged over time. .
- By χ^2 and G statistics (table 2) significant differences were found between the 3 subpopulations of our sample in most of the systems. This fact make us to considerer the use of subpopulations frequencies in our forensic casework [4].

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[4] A. Carracedo, F. Barros, Problemas bioestatísticos en Genetica Forense, 1996, Universidade de Santiago de Compostela, 41-67
[5] M.F. Pinheiro, I.Cainé, L. Pontes, D.Abrantes, G. Lima, M.J.Pereira, P.Rezende, Allele frequencies of sixteen STRs in the population of Northern Portugal, For. Sci.Int. (148) (2005) 221-223
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