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Extended Northern Portuguese database on 21 autosomal STRs used in genetic identification

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Abstract. 21 autosomal STRs are used in our laboratory in routine casework by using two commercially available STR kits (Applied Biosystems' Identifiler and Promega's Powerplex 16) and also an in-house multiplex system, used especially in deficient paternity cases, which amplifies four more loci (CD4, F13A01, FES and MBPB). Northern Portuguese population data are presented here extensively, including D2S1338 and D19S433 for the first time. Deviations from Hardy–Weinberg equilibrium were detected in D8S1179 and Penta D loci, but applying the Bonferroni correction for the number of loci analysed, the departure in both loci was not significant ($0.05/21 = 0.0024$). Both commercial STR kits share 13 loci but use different primer pairs, and so genotype inconsistencies may occur. For individuals genotyped as homozygotes with one kit and as heterozygotes with the other, the latter genotype was the one considered. The overall matching probability for the 21 STRs in our population sample is of 1 in 1.56×10^{24} individuals and combined power of exclusion of 0.9999999914.

Keywords: STR; Population data; Northern Portugal

1. Introduction

Routine casework in our genetic identification laboratory is carried out with two commercially available autosomal STR kits, namely, Identifiler (Applied Biosystems) and Powerplex 16 (Promega), which together amplify a total of 17 STR loci. In some instances, namely, in deficient paternity cases, we can also count on an in-house multiplex system which amplifies four more loci, totalling 21 STRs. Along the years, we have accumulated population frequency data in our database, namely, from individuals residing

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Table 1
Population data for 21 STRs in Northern Portugal

Allele	CD4	CSF1PO	D2S1338	D3S1358	D5S818	D7S820	D8S1179	D13S317	D16S539	D18S51	D19S433	D21S11	F13A01	FES	FGA	MBPB	Penta D	Penta E	TH01	TPO	VWA
	$N=382$	$N=1825$	$N=760$	$N=1816$	$N=1816$	$N=1809$	$N=1822$	$N=1818$	$N=1283$	$N=1810$	$N=761$	$N=1817$	$N=484$	$N=487$	$N=1833$	$N=371$	$N=1280$	$N=1281$	$N=2403$	$N=2402$	$N=2303$.
Ho	0.696	0.731	0.861	0.784	0.702	0.794	0.808	0.773	0.760	0.891	0.806	0.843	0.711	0.700	0.857	0.741	0.837	0.899	0.783	0.639	0.804
He	0.705	0.724	0.862	0.787	0.702	0.810	0.811	0.782	0.776	0.876	0.794	0.843	0.754	0.700	0.866	0.728	0.839	0.885	0.796	0.648	0.810
PD	0.855	0.872	0.967	0.921	0.859	0.937	0.941	0.922	0.915	0.972	0.930	0.957	0.899	0.852	0.967	0.879	0.953	0.976	0.927	0.823	0.937
CE	0.422	0.452	0.711	0.556	0.436	0.601	0.616	0.566	0.546	0.733	0.587	0.673	0.509	0.417	0.712	0.469	0.657	0.755	0.570	0.386	0.602
P	0.929	0.496	0.801	0.754	0.597	0.112	0.034	0.495	0.760	0.919	0.820	0.552	0.098	0.818	0.316	0.360	0.015	0.786	0.658	0.691	0.915

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

in Northern Portugal, which we are now presenting extensively, together with parameters of forensic interest. This is also the first time we are presenting data on both D2S1338 and D19S433.

2. Materials and methods

DNA was extracted from blood or buccal swab samples from routine paternity cases using the Chelex method [1]. PCR amplification was done according to manufacturer's instructions (AB Applied Biosystems and Promega) and according to Alves et al. [2]. Genotyping was carried out on an ABI 310 Genetic Analyser (AB Applied Biosystems) with the appropriate software. Hardy–Weinberg equilibrium was assayed by an exact test [3], using GENEPOP software [4]. Expected heterozygosity (He) was calculated according to Nei [5].

3. Results

Population data for the 21 STRs are displayed in Table 1. Allele frequencies can be obtained from the corresponding author upon request.

Deviations from Hardy–Weinberg equilibrium were detected in D8S1179 and Penta D loci, but applying the Bonferroni correction for the number of loci analysed, the departure in both loci was not significant ($0.05/21 = 0.0024$).

Both commercial STR kits share 13 loci but use different primer pairs, and so genotype inconsistencies may occur. When individuals were genotyped as homozygotes with one kit and as heterozygotes with the other, the latter genotype was the one considered.

The overall matching probability for the 21 STRs in our population sample is of 1 in 1.56×10^{24} individuals and combined power of exclusion of 0.9999999914.

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