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Population genetics of Y-chromosomal STRs in Amharic males from Ethiopia

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Abstract. The Y-STRs are well established in the forensic routine casework. We analysed buccal swab samples from 173 unrelated males of an African population from Ethiopia—the Amharics. The allele and haplotype frequency data, the exclusion power of the STRs and the haplotype diversity were calculated. The most frequent haplotypes are obtained with a frequency of 0.023. The haplotype diversity using these 12 loci was estimated with 0.9974 and the power of discrimination with 0.9917. The allele diversity differs between 0.068 and 0.747. © 2006 Published by Elsevier B.V.

Keywords: Population data; Short tandem repeat; Y-chromosome; Ethiopia

1. Introduction

Y-chromosome-specific short tandem repeat (Y-STRs) markers are well established in the forensic routine casework [1–4]. Y-STRs have proven to be valuable tools for criminal investigations and paternity testing. In this work, we characterise an African population from Ethiopia—the Amharics. The Amharics are the main population in the region of Gondar (Ethiopia).

2. Material and methods

Buccal swab samples from 173 unrelated Amharic males were collected. The DNA extraction was carried out using the Chelex 100 method according to Walsh et al. [5].

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The samples were typed using the PowerPlex[®] Y System (Promega, USA) containing the European minimal haplotype (EMH) loci [6,7] and the loci DYS438 and DYS439 (recommended by SWGDAM) plus the DYS437 locus [8]. Y-STR amplification was performed according to the manufacturer's recommendations [9].

For electrophoretic separation and sample detection, a 4.5% denaturing PAG and the ABI PRISM[®] 377 DNA Sequencer were used (Applied Biosystems, USA), according to manufacturer's instructions. The nomenclature for all loci follows the recommendations of the Y-STR Haplotype Reference Database [7].

Allele/haplotype frequencies were estimated by direct allele/haplotype counting. Allele and haplotype diversities and the exclusion power of the STRs were estimated according to Nei and Tajima [10,11].

The laboratory typed successfully the samples received from the Y-STR Haplotype Reference Database by the Institute of Legal Medicine, Humboldt-University, Berlin.

3. Other remarks

A total of 148 different haplotypes were found from 173 unrelated Amharic males using the present set of Y-STR markers. 131 haplotypes of them were found to be unique,

Table 1 Allele/genotype frequencies of 12 Y-STRs in 173 Amharic male samples

Allele	DYS19	DYS389I	DYS391	DYS392	DYS393	DYS437	DYS438	DYS439
8							0.006	
9			0.289	0.006			0.029	0.012
10			0.619	0.017			0.844	0.040
11	0.093		0.092	0.856	0.011		0.023	0.335
12	0.006	0.190		0.121	0.220		0.092	0.393
13	0.202	0.642			0.613		0.006	0.208
14	0.416	0.168			0.150	0.682		0.006
15	0.237				0.006	0.064		0.006
16	0.040					0.254		
17	0.006							
PD	0.719	0.524	0.525	0.253	0.554	0.466	0.278	0.688
Allele	DYS389II	DYS390		DYS385				
			Gt	Fq	Gt	Fq	Gt	Fq
20		0.023	11-12	0.012	13-18	0.115	16-17.2	0.006
21		0.202	11-13	0.006	13-19	0.012	16-18	0.057
22		0.041	11.2-18	0.006	13-20	0.006	16-19	0.012
23		0.306	12-12	0.123	14-16	0.006	16-20	0.023
24		0.358	12-13	0.098	14-18	0.006	16-21	0.006
25		0.058	12-16	0.006	15-15	0.006	17-17	0.052
26		0.006	12-18	0.006	15-16	0.052	17-18	0.017
28	0.035		12-19	0.034	15-17	0.040	17-19	0.017
29	0.230	0.006	12-20	0.012	15-18	0.012	17-20	0.006
30	0.353		12-21	0.006	15-19	0.012	18-18	0.017
31	0.243		13-13	0.012	16-16	0.069	18-19	0.017
32	0.121		13-14	0.006	16-17	0.089	18-20	0.006
33	0.006							
34	0.012							
PD	0.747	0.731			0.932			

Gt: genotype. Fq: frequency. PD: exclusion power.

2 were shared by four persons, 4 by three persons and 11 were found in two individuals. The total haplotype diversity (HD) using these 12 loci was 0.9974. The power of discrimination was estimated with 0.9917. The DYS392 locus showed the lowest level of polymorphism (gene diversity: 0.747), DYS385a/b the highest (0.068). The other loci varied between 0.253 and 0.722.

When analysing only the nine Y-STR loci from the "minimal haplotype", the most frequent haplotypes for the Amharic population sample (DYS19-DYS389I-DYS389I-DYS390-DYS391-DYS392-DYS393-DYS385a/b: 15-12-29-21-9-11-13-12,13) were searched in the database of YHRD (www.yhrd.org) and were not found in any other population.

There were found low levels of polymorphism for DYS392 (about 86% of the individuals sampled shared allele 11) and DYS438 (about 84% shared allele 10), respectively. For DYS392, similar results were observed for an indigenous population sampled in Maputo (Mozambique [12]), for an Xhosa population sample from Cape Town (South Africa [13]), for a population sample of Central Africa pygmies [14] and in four populations from northwest Africa [15]. These suggest that DYS392 may not be ideally suited to forensic casework in several African populations.

4. Results and discussion

Results are shown in Table 1.

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