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Haplotype analysis of the PowerPlex® Y System in Northeast population from Italy

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Abstract. Recently, a new multiplex set of 12 Y-STRs loci (PowerPelx® Y System, Promega) that includes the 9 Y-chromosome loci of the European minimal haplotype (DYS19, DYS385 loci, DYS389I, DYS389II, DYS390, DYS391, DYS392 and DYS393) plus two loci recommended by SWGDAM (DYS438 and DYS439) and DYS437 locus was commercially released. In the present study we evaluated allele frequencies and other statistical parameters of the PowerPlex® Y System in a population sample of 155 unrelated autochthonous healthy males from northeast Italy. The haplotype diversity (HD) of 12 Y-STR multiplex was 0.9987 and discrimination capacity (DC) was 0.9226. © 2005 Elsevier B.V. All rights reserved.

Keywords: Y-STR; Haplotype diversity; Italian population database; PowerPlex Y

1. Introduction

Y-chromosome analysis is a useful tool in male specific linkage evolutionary study, paternity testing and personal identification [1,2]. Every year an increased number of Y markers are being reported in literature, nevertheless to evaluate their efficiency in forensic science it is necessary to investigate a large number of different populations.

The aim of this study is to evaluate the haplotype distribution of 12 Y-STR loci in a northeast Italy population using PowerPelx® Y System (Promega).

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Table 1
Allele and genotype frequencies (DYS385 loci) and gene diversity (GD) for 12 Y-STRs in a northeast Italy population ($n=155$)

Allele	DYS391	DYS3891	DYS439	DYS389II	DYS438	DYS437	DYS19	DYS392	DYS393	DYS390	Genotype	DYS385 loci
9					0.142						10–14	0.006
10	0.606		0.019		0.316			0.006			11–11	0.006
11	0.361		0.355		0.110			0.452	0.039		11–13	0.006
12	0.026	0.252	0.510		0.406			0.026	0.123		11–14	0.290
13		0.619	0.110		0.026		0.103	0.419	0.690		11–15	0.065
14		0.129	0.006			0.252	0.477	0.090	0.135		11–16	0.026
15						0.484	0.335	0.006	0.013		12–12	0.013
16						0.265	0.052				12–13	0.006
17							0.026				12–14	0.032
18							0.006				12–15	0.026
19											12–16	0.006
20											12–18	0.032
21										0.019	13–13	0.032
22										0.181	13–14	0.039
23										0.174	13–15	0.058
24										0.477	13–16	0.013
25										0.142	13–17	0.013
26										0.006	13–18	0.006
27				0.019							14–14	0.077
28				0.123							14–15	0.052
29				0.452							14–16	0.039
30				0.316							14–17	0.013
31				0.090							14–18	0.006
											15–15	0.026
											15–17	0.013
											15–18	0.006
											16–16	0.013
											16–17	0.006
											16–18	0.039
											16–21	0.006
											17–17	0.006
											17–18	0.006
											18–18	0.006
GD	0.504	0.540	0.606	0.677	0.707	0.637	0.650	0.615	0.492	0.693		0.825

2. Materials and methods

Blood or saliva samples were collected from 105 unrelated, autochthonous healthy males and 50 father–son pairs (confirmed by autosomal STR analysis using AmpF*l*ST Identifiler PCR amplification kit (Applied Biosystems), with paternity probability >99.9%) living in northeast Italy.

Genomic DNA was isolated by using the GenomicPrep Blood DNA Isolation Kit (APB).

PCR amplification was performed using PowerPlex® Y System amplification kit (Promega) according to the user's manual provided by the manufacturer. PCR products were accomplished with the ABI Prism 3100 *Avant* Genetic Analyzer 4-capillary array system (Applied Biosystems) following the manufacturer's protocols. Genotyping of each sample was carried out automatically using the allelic ladders provided with the Y-12plex amplification kit and PowerTyper Y Macro software (Promega). Allele and haplotype frequencies were estimated by gene/haplotype counting. DYS385 loci are analyzed as a "genotype", as recommended by DNA Commission of ISFG [3]. Gene diversity (GD), haplotype diversity (HD) and discrimination capacity (DC) were calculated as described by Kayser et al.

3. Results and discussion

Allele and genotype frequencies (DYS385 loci) and gene diversity for each Y-STR loci analyzed in this population sample are shown in Table 1. The most informative Y-STR was DYS385 loci (0.825) and the least informative was DYS393 (0.492) when considering the gene diversity. A total of 155 unrelated males were analyzed and 143 different haplotypes were defined, among which 134 were unique and 9 occurred more than one time. For the 12 Y-STR loci, haplotype diversity was 0.9987 and discrimination capacity was 0.9226. In one father–son pair out of 50 father–child pairs analyzed, a genetic incompatibility was found at the DYS389I/II system: allele 12 to 11 at DYS389I and allele 30 to 29 at DYS398II. In this case there exists no additional mutation in DY389II, because both loci are combined and mutations in DY389I would automatically lead to a lengthening of the whole locus.

Comparing the number of haplotypes produced and correlated haplotype diversity between the 9 Y-STR loci of the European minimal haplotype and 12 Y-plex, we can observed that PowerPlex Y System has an increased discrimination power and is more informative than the classic set. This result highlights that it is crucial to increase the number of Y-microsatellites for individual identification and paternity testing in forensic genetics in order to improve the discrimination capacity.

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