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Y-chromosome STR haplotypes in a population from Northeast Spain

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Abstract

The aim of this study is to present the distribution of haplotype frequencies for six Y-chromosome short tandem repeat (STR) loci (DYS19, DYS385, DYS389II, DYS390, DYS391 and DYS393) in a population sample of 100 Caucasoid unrelated individuals living in Northeast Spain. A total of 73 different haplotypes were observed among which 67 were unique and 6 were found at least two times. The most common haplotype occurred 12 times. The overall discrimination capacity of the six loci haplotypes analysed was 73%, and the gene diversity was 96.88%. This database study is an essential prerequisite for using Y-chromosomal STR in routine practice. © 2003 Elsevier Science B.V. All rights reserved.

Keywords: STR; Y chromosome; Haplotypes; Population data

1. Introduction

Short tandem repeat (STR) polymorphisms of the Y chromosome, located on the nonrecombining region of the chromosome [1], have been employed for routine forensic casework [2,3]. Y-linked loci are of special interest, because they are haploid and paternally inherited and these characteristics have meant that Y-STR systems are being used in the analysis of particular cases of paternity and sexual assault.

The goal of this study is to report the distribution of haplotypes for six Ychromosome short tandem repeat (STR) loci (DYS19, DYS385, DYS389II, DYS390,

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Table 1

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Alleles	DYS393	DYS19	DYS389II	DYS390	DYS391				
9					0.05				
10					0.38				
11					0.55				
12	0.16	0.01			0.02				
13	0.72	0.09							
14	0.11	0.64							
15	0.01	0.17							
16		0.05							
17		0.04							
22				0.05					
23				0.26					
24				0.57					
25				0.09					
26				0.03					
27			0.01						
28			0.08						
29			0.51						
30			0.26						
31			0.12						
32			0.02						
Gene diversity	0.44	0.56	0.66	0.65	0.55				

Allele frequencies and gene diversity obtained in this study for DYS19, DYS389II, DYS390, DYS391 and DYS393

DYS391 and DYS393) in a population sample of 100 Caucasoid unrelated individuals living in Northeast Spain, to use these Y-STR loci in forensic genetic diagnosis.

2. Materials and methods

Samples were obtained from 100 unrelated putative fathers from paternity analysis trios, living in Northeast Spain. DNA was extracted from blood samples using the phenol–chloroform–isoamyl alcohol protocol.

 Table 2

 Phenotype frequencies and haplotype diversity of DYS385

Haplotypes	Frequency	Haplotypes	Frequency	Haplotypes	Frequency
10/13	0.01	13/13	0.01	14/17	0.01
11/11	0.01	13/14	0.01	14/18	0.01
11/12	0.01	13/15	0.01	15/16	0.01
11/14	0.47	13/16	0.04	15/17	0.02
11/15	0.10	13/17	0.01	16/17	0.02
11/16	0.01	13/18	0.01	16/18	0.01
12/12	0.01	14/14	0.04	17/19	0.02
12/13	0.01	14/15	0.02	18/18	0.01
12/14	0.05	14/16	0.02	18/19	0.01
12/17	0.02				
Haplotype diversity	0.75				

(i)								
DYS393	DYS19	DYS389II	DYS390	DYS391	DYS385	N		
13	14	30	24	11	11/14	6		
13	14	31	24	11	11/14	6		
13	15	29	24	11	11/14	2		
13	14	29	23	11	11/14	3		
13	14	29	24	10	11/14	4		
13	14	29	24	11	11/14	12		

Table 3 Most frequent haplotypes (N>2)

We used the commercial kit Y-PLEX 6^{TM} developed by Reliagen. The PCR was carried out following the amplification conditions recommended by the manufacturer, in a Perkin Elmer 2400 thermocycler. Fragments were analysed on an ABI310 automated system (Applied Biosystems Division/Perkin Elmer). The gene diversity was calculated according to Nei [4].

3. Results and discussion

Tables 1 and 2 show the allele frequency distributions and the gene diversity for the different loci analysed. The frequencies ranged from 0.01 to 0.72.

A total of 73 different haplotypes were observed among which 67 were unique and 6 were found at least two times. Table 3 lists the most common haplotypes observed. The most common haplotype of all occurred 12 times.

The overall discrimination capacity of the six loci haplotypes analysed was 73% (calculated as the percentage proportion of different haplotypes), and the gene diversity was 96.88%.

No major differences were observed between allele distributions derived from our study and those found in other European and Spanish studies. [5-8].

We consider that the application of these combined six Y-STR loci could be an important tool for forensic identification of male DNA.

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