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Population genetics of Y-chromosomal haplotypes in Asturias (Northern Spain)

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Abstract

A core set of 10 Y-linked short tandem repeat (STR) polymorphisms has been recently described and validated by the forensic and scientific community: DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385I, DYS385II, DYS388. The goal of this work was the generation of reliable Y-STR haplotype frequency estimates based on the distribution of these Y-linked short tandem repeat polymorphisms from 120 fresh blood samples collected in Asturias (North Spain). One hundred one different haplotypes were found in 120 samples (haplotype diversity value: 0.9952).

Keywords: Y-chromosome; STRs; Population study; Spain

1. Introduction

Y-linked short tandem repeat (Y-STR) haplotyping has proven to be extremely informative in forensic casework, particularly in cases of rape and other sexual assault as well as kinship testing [1–6].

A core set of 10 linked short tandem repeat polymorphisms has been recently described and validated by the forensic scientific community: DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385I, DYS385II and DYS388 [2–7].

To quantify the positive evidence provided by a trace-donor match, or to facilitate likelihood calculations in the case of kinship testing, haplotype frequencies inevitably must be known.

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The goal of this work was the generation of reliable Y-STR haplotype frequency estimates based on the distribution of these 10 linked short tandem repeat polymorphisms in the population of Asturias (North Spain).

2. Materials and methods

One hundred twenty male blood samples were obtained from healthy unrelated individuals born and living in Asturias. DNA was isolated from the samples using a standard phenol–chloroform extraction method. Amplification was carried out according to Nievas [8] in a PE 2400 thermocycler (PE Biosystems). Each Y-STR locus was amplified individually, except DYS389I/II and DYS385 (two loci each with the same set of primers). Ladders for the 10 Y-STR loci: DYS19, DYS385, DYS388, DYS389I, DYS389II, DYS390, DYS391, DYS392 and DYS393 were kindly provided by Dr. Peter de Knijff (Universidad de Leiden, The Netherlands). Detection of the amplified products was carried out using the Automated Laser Fluorescent (ALF Express) DNA sequencer (Pharmacia, Uppsala, Sweden).

Table 1
Y-STR haplotypes among 120 Asturian males (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385I, DYS385II, DYS388)

Haplotype	Frequency	Haplotype	Frequency
13-12-29-24-10-11-13-17-17-12	0.00833	14-12-30-23-9-11-14-11-14-13	0.00833
13-12-30-24-11-12-13-15-19-12	0.00833	14-13-26-24-11-13-13-11-15-12	0.00833
13-13-30-22-10-11-13-17-18-12	0.00833	14-13-28-22-11-13-12-11-14-12	0.00833
13-13-30-23-9-11-14-16-16-12	0.00833	14-13-28-23-10-13-13-9-14-12	0.00833
13-13-30-24-10-11-13-16-18-12	0.00833	14-13-28-23-12-13-13-11-14-12	0.00833
13-13-30-24-10-11-13-16-18-13	0.00833	14-13-28-24-10-14-13-11-14-12	0.00833
13-13-30-24-10-13-13-11-15-12	0.00833	14-13-29-21-10-11-12-13-16-15	0.00833
13-13-31-24-10-11-13-16-17-12	0.00833	14-13-29-22-10-11-12-15-18-14	0.00833
13-13-31-24-10-11-13-16-18-12	0.00833	14-13-29-23-10-11-13-13-16-14	0.00833
13-13-31-24-11-11-12-16-18-12	0.00833	14-13-29-23-10-13-13-12-14-12	0.00833
13-14-29-25-9-11-13-13-14-12	0.00833	14-13-29-23-10-13-15-12-15-12	0.00833
13-14-30-24-9-11-13-13-14-12	0.00833	14-13-29-23-11-13-13-11-14-12	0.025
13-14-31-23-10-11-13-16-19-12	0.00833	14-13-29-23-11-13-13-11-15-12	0.00833
13-14-31-24-9-11-13-13-14-12	0.025	14-13-29-23-11-13-14-11-15-12	0.00833
14-12-27-23-10-11-13-14-15-14	0.00833	14-13-29-24-10-12-13-17-20-12	0.00833
14-12-28-21-10-11-13-13-14-14	0.00833	14-13-29-24-10-13-13-11-12-12	0.00833
14-12-28-22-11-13-13-12-14-12	0.00833	14-13-29-24-10-13-13-11-14-12	0.0333
14-12-28-23-10-11-12-13-14-17	0.00833	14-13-29-24-10-13-13-11-15-12	0.0167
14-12-28-23-11-11-13-11-14-12	0.00833	14-13-29-24-10-13-13-12-14-12	0.0167
14-12-28-23-11-13-13-11-14-12	0.0167	14-13-29-24-10-13-14-11-14-12	0.0167
14-12-28-24-10-11-13-13-14-14	0.00833	14-13-29-24-11-13-13-11-13-12	0.00833
14-12-28-24-10-13-13-11-14-12	0.00833	14-13-29-24-11-13-13-11-14-12	0.05
14-12-28-24-11-13-13-11-14-12	0.00833	14-13-29-24-11-13-13-11-15-12	0.0167
14-12-28-26-11-13-13-11-14-12	0.00833	14-13-29-24-11-13-13-11-16-12	0.00833
14-12-29-23-11-13-14-11-14-12	0.00833	14-13-29-24-11-13-14-11-15-12	0.00833

Freq=frequency.

Haplotype frequencies were determined for all the 10 loci in accordance with Kayser et al. [2] and genetic diversity according to Edwards et al. [9].

3. Results and discussion

Table 1 shows the haplotype distribution in the Asturian population sample that has been typed for these Y-STR loci. A total of 101 haplotypes was detected in 120 individuals. The most common haplotype was DYS19*14-DYS389I*13-DYS389II*29-DYS390*24-DYS391*11-DYS392*13-DYS393*13-DYS385*11-14, DYS388*12 with a frequency of 5%.

For Y-chromosome-specific markers, the allele distribution for each marker is less important than the haplotype diversity for the whole array of loci. The haplotype diversity value of the Y-chromosome corresponds to the value of the power of discrimination (PD) and the chance of exclusion (CE) for unrelated males. The haplotype diversity for all the 10 Y-STR loci in the Asturian population was calculated to be 0.9952 ± 0.0023. This implies that this set of Y-specific STR loci will be valuable additions to our current test

Haplotype	Frequency	Haplotype	Frequency
14-13-29-24-11-13-14-12-15-12	0.00833	15-12-29-22-10-11-13-12-14-12	0.0167
14-13-29-25-10-13-13-11-14-12	0.00833	15-12-29-22-10-11-14-13-13-12	0.00833
14-13-29-25-10-13-14-11-14-12	0.00833	15-12-29-23-10-11-12-14-14-12	0.00833
14-13-29-25-11-13-13-11-14-12	0.00833	15-13-28-24-10-13-13-11-15-12	0.00833
14-13-29-25-11-13-13-11-15-12	0.00833	15-13-29-22-10-11-12-13.15-15	0.00833
14-13-30-23-11-12-13-11-14-12	0.00833	15-13-29-23-10-11-12-13.14-17	0.00833
14-13-30-24-9-13-13-15-17-12	0.00833	15-13-29-23-11-13-13-11.14-12	0.00833
14-13-30-24-10-13-13-12-14-12	0.00833	15-13-29-23-11-13-14-10.15-12	0.00833
14-13-30-24-11-11-13-11-14-14	0.00833	15-13-29-24-11-13-13-11.14-12	0.00833
14-13-30-24-11-14-13-11-14-12	0.00833	15-13-29-24-11-15-13-11.14-12	0.00833
14-14-29-22-11-13-13-11-17-12	0.00833	15-13-29-25-10-12-13-12.16-13	0.00833
14-14-30-23-10-13-12-11-14-12	0.00833	15-13-30-23-9-11-12-13-15-16	0.00833
14-14-30-23-11-13-13-11-15-12	0.00833	15-13-30-23-10-13-12-11-16-12	0.00833
14-14-30-23-11-13-13-12-14-12	0.00833	15-13-30-23-10-13-13-14-18-13	0.00833
14-14-30-24-10-11-12-13-20-16	0.00833	15-13-30-24-9-13-14-12-16-12	0.00833
14-14-30-24-10-13-13-11-14-12	0.00833	15-13-30-24-10-13-13-11-14-12	0.00833
14-14-30-24-10-13-13-11-16-12	0.00833	15-13-30-25-10-13-13-11-13-12	0.00833
14-14-30-24-11-13-13-11-14-12	0.00833	15-13-31-23-10-13-13-15-16-12	0.00833
14-14-30-24-11-13-13-12-14-12	0.00833	15-14-29-23-10-11-13-12-12-13	0.0167
14-14-30-24-11-13-13-12-13-12	0.00833	15-14-30-23-11-11-14-13-15-12	0.00833
14-14-31-25-10-14-13-11-14-12	0.00833	15-15-31-23-9-11-13-14-16-16	0.00833
14-14-32-22-11-13-13-14-16-12	0.00833	16-12-28-22-11-11-12-14-14-13	0.00833
14-14-32-24-10-13-13-17-22-12	0.00833	16-13-28-23-10-11-13-12-12-13	0.00833
15-12-28-22-10-11-14-12-13-17	0.00833	16-13-29-22-11-11-12-12-16-15	0.00833
15-12-29-21-10-11-14-13-15-12	0.00833	16-14-29-23-10-13-13-12-12-12	0.00833
		17-13-28-23-9-11-13-11-12-13	0.00833

panel and will improve the exclusion probabilities in forensic and kinship cases when individuals from Asturias are involved.

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