

Allele distribution of five X-chromosome STR loci in an Italian population sample

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Abstract. Population genetic data for five X-chromosomal STR loci (DXS7423, DXS6789, DXS6795, DXS9898 and DXS8377) were generated by analysing a population sample from Northwest Italy. Intensive stutter bands were observed for the DXS8377 locus. The analysis of the 40 family trios segregation showed no new mutation. © 2003 Elsevier B.V. All rights reserved.

Keywords: X-Chromosome; STRs; Population data

1. Introduction

The aim of this study was to investigate the distribution of five STR loci (DXS7423, DXS6789, DXS6795, DXS9898 and DXS8377) in a population sample from 120 unrelated individuals (60 females and 60 males) and 40 family trios (20 male and 20 female offspring) living in the Pavia area (Northwest Italy).

2. PCR amplification and allele typing

DXS7423, DXS6789 and DXS6795 loci were amplified in one triplex and DXS9898 and DXS8377 loci in one duplex PCR reaction, using primers sequences already reported [1–5]. Fragment lengths (bp) processed by ABI 310 capillary sequencer were compared to the published data [1–5] and allelic ladders were calibrated by typing K562 and 9947A. Seven alleles of the DXS6795 locus were sequenced in order to identify the repeat motif. The consensus repeat structure of alleles 12 to 15 was the following: (TAA)_{10–13}TG(TAA)TTG(-TAA), while allele 10 showed the slightly modified repeat motif (TAA)₆(TGA)(TAA)TG(-TAA)TTG(TAA). The alleles were named as recommended by the ISFH [6].

3. Results and discussion

The allele frequencies calculated for females and males independently are shown in Fig. 1. These typing results were consistent with other previously published data

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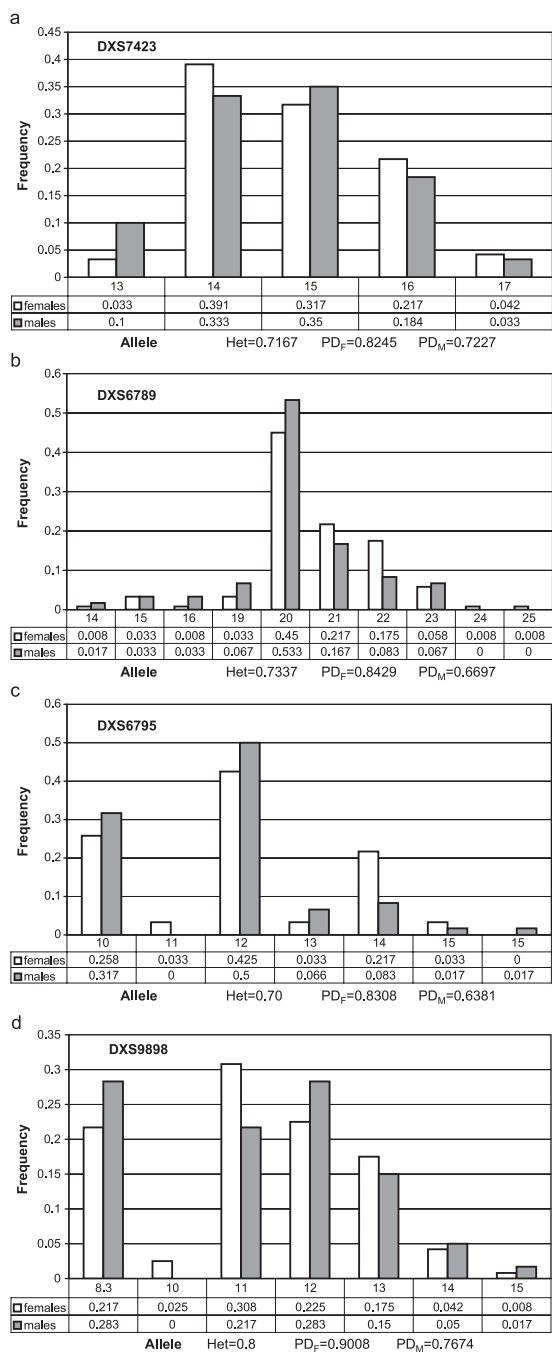


Fig. 1. Allele frequency distribution in females and males.

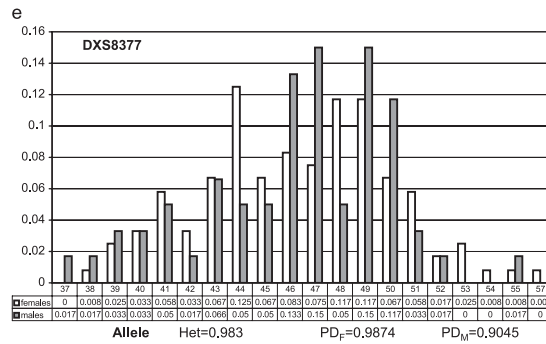


Fig. 1 (continued).

[2,4,5]. Intensive stutter bands were observed by typing the DXS8377 locus, as already shown [5].

Data compared by Fisher test were similar in males and females for all markers. No deviations from HWE were found in females. Statistical parameters were calculated according to Desmarais et al. [7]. No mutation was found by analysing 40 family trios.

The pooled data for the DXS7423, DXS6798, DXS9898 and DXS8377 loci compared with a population sample from Germany showed very similar distributions.

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