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Forensic evaluation of three closely linked STR markers in a 13 kb region at Xp11.23

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Abstract. Searching for markers located in the region p11 of the X-chromosome the clone AF196972 was checked for its content of microsatellites. The two tetranucleotide STRs DXS10076 and DXS10078 and the trinucleotide STR DXS10077 were characterised and evaluated for their forensic efficiency in a population sample of 352 unrelated individuals from North-West Germany. Whereas at locus DXS10076 10 alleles with PIC and HET values of 0.767 and 0.747 and at locus DXS10078 13 alleles with PIC and HET values of 0.811 and 0.861 could be observed locus DXS10077 only consists of 5 alleles leading to much lower HET and PIC values (0.492 and 0.507) and therefore to a decreased individualization capacity. © 2006 Published by Elsevier B.V.

Keywords: Forensic DNA-typing; Short tandem repeat (STR); X-chromosome; German population

1. Introduction

Compared to autosomal and Y-STR-markers X-chromosomal STRs are less established in forensic routine case work until now. Nevertheless, they have proven to be a very strong tool in the analysis of complex kinship cases [1–4]. In order to enable haplotyping strategies on the X-chromosome the aim of the present study was to look for new STR-

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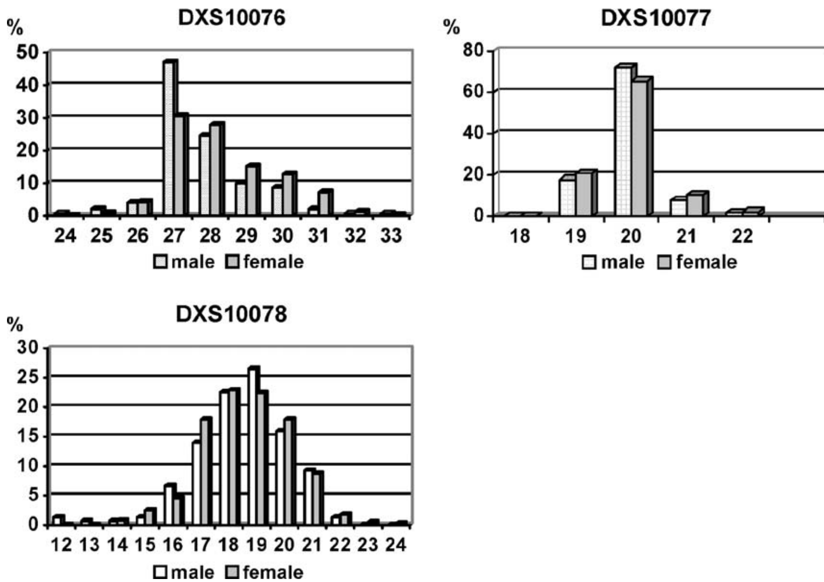

Fig. 1. Allele frequencies of markers DXS10076, DXS10077 and DXS10078 in a population sample from North-West Germany ($n = 151$ females and 201 males).

markers in the Xp11 of the clone AF196972 and to evaluate them for their forensic efficiency.

2. Materials and methods

Blood samples were taken from 151 female and 201 male individuals from North-West Germany in routine paternity cases. DNA was extracted with the QiaAmp Mini Kit (Qiagen). Primers were designed according to GenBank information with the Primer3 software. Amplification was carried out as a triplex reaction in a total volume of 12.5 µl using 0.4 µM of each primer, 1× Gold-Star™ buffer (Promega), 1 U of AmpliTaqGold-DNA-Polymerase (ABI) and 5 ng DNA. PCR conditions were as follows: initial denaturation at 94° for 10 min followed by 30 cycles of 94° for 45 s, 58° for 45 s and 72° for 1 min, then final extension at 68° for 1 h. Primers used for amplification:

DXS10076 A: 5′-JOE-cctgggcaacagagcaagac-3′; B: 5′-gttcttatggagattcagctgg-3′
DXS10077 A: 5′-NED-ctgggctgggcaaaggctc-3′ B: 5′-ggatgccaagcttggatcatg-3′
DXS10078 A: 5′-FAM-aaaaaaaagaaggaagggagga-3′ B: 5′-ttgtgttgcactgattttctcc-3′

Table 1
Parameters of forensical relevance

Locus	HET_{obs}	HET_{exp}	PIC	MEC	PD(♀)	PD(♂)
DXS10076	0.746	0.784	0.767	0.751	0.921	0.782
DXS10077	0.507	0.518	0.492	0.468	0.717	0.516
DXS10078	0.861	0.825	0.811	0.799	0.945	0.823

HET_{obs}: observed heterozygosity; HET_{exp}: expected heterozygosity; PIC: polymorphic information content; MEC: mean exclusion chance; PD(♀): power of discrimination (females); PD(♂): power of discrimination (males).

PCR products were analysed by capillary electrophoresis on a ABI PRISM 310 Genetic Analyser. Sequencing of the alleles was performed using the Big Dye Terminator Cycle Sequencing Kit (ABI).

3. Results and discussion

Three STR-markers not used for forensic purposes so far were detected in the clone 196972 at the Xp11 region of the X-chromosome and evaluated with respect to their forensic potential. The two polymorphic tetranucleotide and one trinucleotide repeats are located between 48.065 and 48.078 Mb from the Xptel, spanning a region of 13 kb.

Frequency data and statistical parameters of the markers are given in Fig. 1 and Table 1. No deviation from Hardy–Weinberg-Equilibrium was observed for the three markers. The tetranucleotide STR DXS10076 exhibits 10 alleles and PIC and HET values of 0.747 and 0.767 (Table 1). A significant difference ($p = 0.0047$) in the allele frequency distribution between male and female individuals was observed. Whereas the frequency of allele 27 was much higher in males, the frequency of allele 31 was much higher in females. This phenomenon requires further investigations. More individuals and other populations will be typed soon. The trinucleotide DXS10077 exhibits 5 alleles and shows only moderate HET and PIC values of 0.507 and 0.492. The second tetranucleotide DXS10078 consists of 13 alleles and has a high discriminative efficiency due to HET and PIC values of 0.861 and 0.811. In 150 families with proven paternity two single-step mutations were observed at the locus DXS10078, no mutations at loci DXS10076 and DXS10077.

Due to the short distance between the three markers that spans only 13 kb the markers are very closely linked. Theoretically a crossing over frequency within this cluster < 0.001 (genetical distance < 0.1 cM) can be assumed. Hence this cluster can be a very useful tool in solving complex kinship cases via haplotyping strategies, as could be shown recently with other X-marker cluster [1,4].

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