Available online at www.sciencedirect.com
ScienceDirect
Forensic Science International 175 (2008) 73–78
Forensic Science International
www.elsevier.com/locate/forsciint

Hungarian population data of eight X-linked markers in four linkage groups

Andrea Zalán [a], Antónia Völgyi [a], Werner Brabetz [b], Dorit Schleinitz [b], Horolma Pamjav [a,*]

[a] *Institute of Forensic Medicine, Budapest, Hungary*
[b] *Biotype AG, Moritzburger Weg 67, D-01109 Dresden, Germany*

Received 15 December 2006; received in revised form 10 May 2007; accepted 12 May 2007
Available online 22 June 2007

Abstract

The X chromosomal STR markers DXS10135 and DXS8378 in linkage group 1, DXS7132 and DXS10074 in linkage group 2, HPRTB and DXS10101 in linkage group 3, and DXS10134 and DXS7423 in linkage group 4 were studied in the Hungarian population. After genotyping unrelated men (219) and women (165), forensic efficiency parameters were calculated. Deviations from Hardy-Weinberg equilibrium could not be detected. There were several microvariant and rare alleles were sequenced: four in locus DXS10135 (alleles 17.1, 18.1, 20.1 and 26.1), one in locus DXS10074 (alleles 11), three in locus DXS10101 (alleles 26, 34.2 and 35) and five in locus DXS10134 (alleles 35.3, 37.2, 38.2, 39.2, 41).

Keywords: X-chromosome; STRs; Deficiency paternity testing; Sequencing; *G* statistics

1. Introduction

Fathers transmit their X-chromosome to daughters as haplotypes. Analysis of X-chromosomal loci might be beneficial in deficiency paternity cases, when half-sisters and/or grandmothers are to be examined [1]. The commercially available Mentype® Argus X-8 kit (Biotype AG, Dresden, Germany) makes it possible to examine the DXS10135-DXS8378, DXS7132-DXS10074, HPRTB-DXS10101 and DXS10134-DXS7423 microsatellite markers, which belong to the four linkage groups of the X-chromosome in one multiplex reaction [2]. The four unlinked X-chromosomal loci (DXS8378, DXS7132, HPRTB and DXS7423) have been studied in other populations and compared to each other with *G*-test previously [3].

This paper is aimed to continue the process for establishing a suitable X-STR database of the Hungarian population for the eight loci, mainly for complementary testing in deficiency paternity cases with the autosomal DNA markers [1,4].

2. Materials and methods

2.1. Population

Peripheral blood was collected from 219 unrelated males and 165 females derived from independent regions of Hungary [3].

2.2. DNA extraction, PCR and typing

Genomic DNA was isolated from blood samples with QIAmp Blood Mini Kit (Qiagen GmbH, Hilden, Germany).

The genomic DNA was amplified using the Mentype® Argus X-8 kit (Biotype AG, Dresden, Germany).

The PCR products were analyzed by capillary electrophoresis using an ABI Prism 310 Genetic Analyzer (Applied Biosystems, Foster City, CA). The fragment sizes were determined using the GeneScan 3.1.2 software and the allele designations were performed using Genotyper 2.5.2 software with the Argus X-8_v0.gta macro (Biotype AG).

2.3. Sequencing

PCR products of sequenced alleles were obtained with unlabeled primer pairs. The amplicons were cloned into *Escherichia coli* TOP10F′ (Invitrogen, Carlsbad, CA) using the plasmid pCRBlunt-TOPO from the Zero Blunt™ TOPO® PCR Cloning Kit for Sequencing (Invitrogen). Sequencing of both strands of the DNA inserts of at least two bacterial colonies per transformation was performed using the Big Dye Terminator Cycle Sequencing Kit (Applied

* Corresponding author. Tel.: +36 1 4570183; fax: +36 1 4570182.
E-mail addresses: phorolma@hotmail.com, hori58@excite.com (H. Pamjav).

0379-0738/$ – see front matter © 2007 Elsevier Ireland Ltd. All rights reserved.
doi:10.1016/j.forsciint.2007.05.012

Biosystems). The primer, amplicon sequences and PCR conditions are available at dbSTS (dbSTS_Id 826864–826876) and GenBank (accession numbers BV695821–BV695833).

2.4. Analysis of the data

Polymorphism information content (PIC), expected heterozygosity (HET^{exp}), observed heterozygosity (HET^{obs}), power of discrimination (PD) for males and females were calculated [5–7]. Mean exclusion chance for (MEC) for deficiency cases (mother, daughter, putative grandmother) was determined using the formula of Krüger [8]. MEC values for normal trios (mother, daughter, putative father) were calculated according to Kishida et al. [9]. MEC for duo cases (daughter, putative father or son, putative mother) was estimated according to Desmarais et al. [6]. Arlequin 2.0 software (http://lgb.unige.ch/arlequin.html) was used to perform testing of Hardy-Weinberg equilibrium for female samples. The *G*-tests were performed with the PopTools software (http://www.cse.csiro.au/poptools.html).

3. Results and discussion

3.1. Hungarian population data and statistics

Allele frequencies and forensic efficiency parameters (PIC, PD^{male}, PD^{female}, HET^{exp}, HET^{obs}, $MEC^{Krüger}$, $MEC^{Kishida}$, $MEC^{Desmarais\ duo}$, *p*-values for the HW equilibrium) of the eight X-STR loci were determined in the Hungarian population based on the genotyping of 165 females and 219 males (Table 1). No significant differences were observed between male and female allele frequencies using *G*-test. Table 1 shows the cumulated allele frequencies of the loci DXS10135, DXS10074, DXS10101 and DXS10134. The allele frequencies for the loci DXS8378, DXS7132, HPRTB and DXS7423 included in the Mentype® Argus X-8 kit were published before [3]. Table 2

Table 1
Allele frequencies for four X-STR markers in the Hungarian population

Allele	DXS10074	DXS10135	DXS10101	DXS10134	Combined[a]
7	0.0801				
8	0.1494				
9	0.0128				
11	0.0036				
12	0.0018				
13	0.0055				
14	0.0128				
15	0.0929				
16	0.1949	0.0055			
17	0.2423	0.0200			
17.1		0.0018			
18	0.1384	0.0401			
18.1		0.0073			
19	0.0546	0.0656			
19.1		0.0109			
20	0.0109	0.0638			
20.1		0.0128			
21		0.0801			
21.1		0.0164			
22		0.0710			
22.1		0.0036			
23		0.0929			
23.1		0.0055			
24		0.0856			
25		0.0893			
25.1		0.0018			
25.2			0.0018		
26		0.0638	0.0036		
26.1		0.0018			
26.2			0.0109		
27		0.0692	0.0055		
27.2			0.0565		
28		0.0656	0.0364		
28.2			0.1056		
29		0.0364	0.0146		
29.2			0.1056		
30		0.0455	0.0401		
30.2			0.1876		
31		0.0219	0.0801	0.0055	
31.2			0.1038		
32		0.0109	0.1075	0.0091	
32.2			0.0583		
33		0.0055	0.0401	0.0546	
33.2			0.0219		
34		0.0018	0.0164	0.1184	