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X-STR diversity patterns in the Finnish and the Somali population

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ABSTRACT

Autosomal and Y-chromosomal STR markers have been routinely used in kinship analyses already for over a decade, augmented by mitochondrial DNA in more complex cases questioning the maternal relationships of the samples. Recently, a commercial X-chromosome typing kit Mentype Argus X-8 was introduced to supplement the existing forensic toolkit. In this study, X-STR allele frequencies and population diversity indices in two ethnic groups, the Finnish and the Somali, are reported. Several previously unreported alleles and features in the allelic distribution were observed, some of which were further investigated with a small set of family data. Most notably, several alleles showed significant frequency differences between sexes, yet no obvious explanation for this discrepancy was found. As a demonstration of X-chromosome analysis in practice, we describe two family reunion cases, where the X-STR data was successfully utilized.

1. Introduction

Even though the first X-chromosomal STR marker was found early in the DNA-based identification era [1] the X-chromosomal markers have only recently entered the field of forensic genetics. The commercial X-chromosome typing kit for routine use, Mentype Argus X-8 (Biotype AG, Dresden), has been available since 2006. The kit contains eight polymorphic STR loci, four of which have been used in forensic community more widely (HPRTB, DXS7132, DXS7423, and DXS8378) (e.g. [2,3]). As of yet, studies including the four remaining loci (DXS10074, DXS10101, DXS10134, and DXS10135) have been less commonly reported (but see [4,5]).

The advantages of X-chromosome manifest in deficient kinship analyzes, for example determining the relationship between two presumed half-sisters, when the putative father of both examinees is not available. The need for validated, commercially available X-chromosome STR kit has increased for instance due to growing number of complicated kinship questions encountered in family reunion cases. These typically involve refugees separated from their kin, seeking residence permit and reunion with the rest of their family in another country. Partial families and/or information often associated with these cases make them challenging for forensic laboratories. Currently, the family relationships for immigrants are routinely confirmed by DNA-testing in several European countries, e.g. Germany, United Kingdom, Holland, Norway and Sweden. In Finland, as well, residence permits are granted on the grounds of proven family relationship. The state will provide DNA-testing if adequate clarification of family relationships is not available by other means, which has led to a growing number of DNA-tests aimed at providing the evidence.

The X-STR markers are already proven as an invaluable marker system, complementing the autosomal STRs, mitochondrial DNA and Y-chromosomal STRs, in the family reunion and paternity cases [6–8].

This study reports the X-STR diversity patterns in the two ethnic groups most commonly encountered in casework in Finland, the Finns and the Somali. Several noteworthy features in the X-STR variation were observed in the data. In order to clarify these, inheritance patterns of a subset of alleles were examined in a small set of family data. In addition, we describe two family reunion cases, where X-STR data was successfully applied.

2. Materials and methods

Altogether 700 anonymized Finnish and Somali samples were included in this study. These samples were collected either with informed consents for research purposes, or chosen randomly from the casework samples. The main data comprises of altogether 800 X-chromosomes from 600 unrelated individuals, 100 females and 200 males in both populations. In addition, 51 unrelated Somali mothers from family reunification cases were examined in order to clarify X-chromosome inheritance patterns. For 18 mothers showing unexpected allele patterns (see below), the X-STR profiles of their 49 offspring were also obtained.

The samples were typed using Mentype Argus X-8 kit (Biotype AG, Dresden) according to the manufacturer's instructions, apart from the PCR reaction volume and cycle number. These were

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Table 1
Allele frequencies and various discrimination indices in the studied populations. Alleles showing significantly lower frequencies in males are highlighted.

	HPRTB						DXS7423						DXS10074					
	Somali all	Somali male	Somali female	Finn all	Finn male	Finn female	Somali all	Somali male	Somali female	Finn all	Finn male	Finn female	Somali all	Somali male	Somali female	Finn all	Finn male	Finn female
6	0.0125	0.0150	0.0100															
7													0.1100	0.1200	0.1000	0.0704	0.0950	0.0455
8				0.0025		0.0050							0.1700	0.1800	0.1600	0.1030	0.1000	0.1061
9	0.0850	0.1000	0.0700	0.0050	0.0050	0.0050							0.0125	0.0050	0.0200	0.0075		**0.0152**
10	0.0025	0.0050		0.0025	0.0050								0.0025		0.0050			
11	0.1100	0.0950	0.1250	0.0825	0.0900	0.0750												
11.2				0.0025	0.0050													
12	0.2675	0.2600	0.2750	0.3825	0.3850	0.3800							0.0225	0.0250	0.0200			
13	0.2450	0.2300	0.2600	0.2950	0.3000	0.2900	0.0675	0.0500	0.0850	0.1075	0.0900	0.1250	0.1125	0.1200	0.1050	0.0075	0.0100	0.0051
14	0.1800	0.1800	0.1800	0.1425	0.1300	0.1550	0.4675	0.4850	0.4500	0.3025	0.2950	0.3100	0.1025	0.1250	0.0800	0.0075	0.0150	
15	0.0800	0.0850	0.0750	0.0600	0.0600	0.0600	0.3425	0.3550	0.3300	0.4350	0.4550	0.4150	0.0425	0.0500	0.0350	0.0578	0.0550	0.0606
16	0.0175	0.0300	0.0050	0.0225	0.0200	0.0250	0.0875	0.0750	0.1000	0.1425	0.1500	0.1350	0.0975	0.1000	0.0950	0.1859	0.1700	0.2020
17				0.0025		0.0050	0.0275	0.0200	0.0350	0.0125	0.0100	0.0150	0.1225	0.1350	0.1100	0.2940	0.3000	0.2879
18							0.0050	0.0100					0.1225	**0.0800**	**0.1650**	0.1809	0.1950	0.1667
19							0.0025	0.0050					0.0700	**0.0500**	**0.0900**	0.0729	0.0550	0.0909
20													0.0125	0.0100	0.0150	0.0101	0.0050	0.0152
21																0.0025		0.0051
MEC_{KRU}	0.6266	0.6442	0.6043	0.5145	0.5077	0.5137	0.3943	0.3679	0.4177	0.4360	0.4209	0.4471	0.7736	0.7625	0.7736	0.6551	0.6456	0.6538
MEC_{KIS}	0.7840	0.7958	0.7690	0.6987	0.6917	0.6972	0.5910	0.5645	0.6144	0.6357	0.6208	0.6456	0.8780	0.8713	0.8777	0.8021	0.7950	0.8007
MEC_{DES}	0.7840	0.7958	0.7690	0.6940	0.6917	0.6972	0.5910	0.5645	0.6144	0.6333	0.6208	0.6456	0.8780	0.8713	0.8777	0.7998	0.7950	0.8007
MEC_{DESDUO}	0.6622	0.6775	0.6431	0.5551	0.5526	0.5587	0.4455	0.4196	0.4690	0.4877	0.4745	0.5009	0.7909	0.7811	0.7907	0.6832	0.6768	0.6843
PIC	0.7840	0.7958	0.7690	0.6940	0.6917	0.6972	0.5910	0.5645	0.6144	0.6333	0.6208	0.6456	0.8780	0.8713	0.8777	0.7998	0.7950	0.8007
H_E	0.8121	0.8238	0.8021	0.7366	0.7364	0.7413	0.6526	0.6333	0.6735	0.6884	0.6786	0.7011	0.8908	0.8872	0.8927	0.8238	0.8218	0.8268
	±0.0195	±0.0269	±0.0282	±0.022	±0.0312	±0.031	±0.0238	±0.0341	±0.0332	±0.0232	±0.033	±0.0324	±0.0156	±0.0224	±0.0219	±0.0191	±0.0271	±0.0269
H_O			0.7900			0.7600			0.6700			0.6900			0.8600			0.7900
PD_{FEMALE}	0.9378	0.9436	0.9301	0.8889	0.8875	0.8908	0.8182	0.7976	0.8354	0.8485	0.8401	0.8566	0.9770	0.9747	0.9770	0.9463	0.9441	0.9466
PD_{MALE}	0.8101	0.8197	0.7981	0.7348	0.7327	0.7375	0.6510	0.6301	0.6702	0.6867	0.6752	0.6976	0.8886	0.8828	0.8883	0.8217	0.8177	0.8226

	DXS7132						DXS8378					
	Somali all	Somali male	Somali female	Finn all	Finn male	Finn female	Somali all	Somali male	Somali female	Finn all	Finn male	Finn female
7	0.0025	0.0050										
8												
9										0.0100	0.0150	0.0050
10							0.3400	0.3700	0.3100	0.4050	**0.3600**	**0.4500**
11				0.0025	0.0050		0.3325	0.3300	0.3350	0.2950	0.3100	0.2800
12	0.0525	0.0400	0.0650	0.0950	0.1050	0.0850	0.3075	0.2950	0.3200	0.2775	0.3050	0.2500
13	0.1775	0.1650	0.1900	0.2750	0.2800	0.2700	0.0200	**0.0050**	**0.0350**	0.0100	0.0050	0.0150
14	0.3725	0.3950	0.3500	0.3300	0.3050	0.3550				0.0025	0.0050	
15	0.2925	0.2950	0.2900	0.2250	0.2250	0.2250						
16	0.0875	0.0850	0.0900	0.0575	0.0600	0.0550						
17	0.0150	0.0150	0.0150	0.0150	0.0200	0.0100						
MEC_{KRU}	0.5045	0.4847	0.5161	0.5257	0.5415	0.5061	0.3930	0.3742	0.4088	0.3902	0.3983	0.3766
MEC_{KIS}	0.6935	0.6753	0.7031	0.7122	0.7242	0.6964	0.6103	0.5945	0.6231	0.6048	0.6139	0.5893
MEC_{DES}	0.6900	0.6753	0.7031	0.7111	0.7242	0.6964	0.6103	0.5945	0.6231	0.6048	0.6139	0.5893
MEC_{DESDUO}	0.5500	0.5338	0.5646	0.5736	0.5889	0.5568	0.4629	0.4466	0.4763	0.4574	0.4668	0.4414
PIC	0.6900	0.6753	0.7031	0.7111	0.7242	0.6964	0.6103	0.5945	0.6231	0.6048	0.6139	0.5893
H_E	0.7353	0.7243	0.7485	0.7541	0.7667	0.7438	0.6806	0.6705	0.6915	0.6737	0.6844	0.6596
	±0.0221	±0.0316	±0.0307	±0.0215	±0.0299	±0.0309	±0.0233	±0.0332	±0.0327	±0.0234	±0.0329	±0.0335
H_O			0.6500			0.6900			0.6200			0.6800
PD_{FEMALE}	0.8855	0.8766	0.8932	0.8975	0.9051	0.8887	0.8283	0.8166	0.8377	0.8252	0.8312	0.8149
PD_{MALE}	0.7335	0.7207	0.7448	0.7522	0.7629	0.7401	0.6789	0.6671	0.6881	0.6720	0.6810	0.6564