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Application of Y-chromosomal STR Haplotypes to Forensic Genetics

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This paper delivers population genetic data on Y-chromosomal short tandem repeat (STR) polymorphisms along with reports of unusual observations and casework. Population studies were carried out on the Y-specific STR polymorphisms DYS19, DYS385 I+II, DYS389 I+II, DYS390, DYS391, DYS392, and DYS393 in population samples from North India, Turkey, and Germany. In all three populations the vast majority of haplotypes was observed only once, especially in the Turkish group. Highly unusual cases are reported. In a German individual, we observed the variant allele DYS392*11.1, whereas a Turkish haplotype revealed a duplication at locus DYS19. Application of Y-chromosomal STR markers to forensic genetics was demonstrated in two cases: 1) a deficient paternity case, and 2) a father/son pair, where the Amelogenin primers failed to amplify the Y-homolog. In forensic genetics, Y-chromosomal STR polymorphisms are highly welcomed as an additional tool.

Key words: *forensic medicine; Germany; haplotypes; India; short tandem repeats; Turkey; Y chromosome.*

DNA-polymorphisms on the human Y chromosome are valuable tools for evolution and migration studies (1-5). In forensics, Y chromosome markers are used particularly in rape cases and in deficient paternity cases with male offspring (6-10).

To date, most forensic Y-chromosome DNA work around the world has focused on a set of 7-9 short tandem repeat (STR) polymorphisms (10,11). This study aimed at collecting data regarding haplotype frequencies in populations from India, Turkey, and Germany. Moreover, unusual haplotypes are reported, which were caused by a mutation and a duplicated locus. Finally, the application of Y-chromosomal markers to a deficient paternity case and to a case with an allelic drop-out at the amelogenin locus is reported.

Material and Methods

Population Samples

Jat Sikhs are endogamous but practice g*otra* (clan like organization) exogamy. Samples from 108 unrelated individuals were analyzed. The samples were collected from the Punjabi University students and villagers belonging to the districts of Patiala, Fatehgarh Sahib, and Sangrur of the Punjab, India.

Unrelated Turkish males living in Germany (n = 281) were tested as they were involved in legal proceedings concerning paternity. In addition, 166 unrelated German Caucasians were analyzed.

DNA Extraction

DNA was extracted either from Iso Sticks (Schleicher and Schuell Inc, Dassel, Germany) following the company's recommendation or from EDTA blood samples with the salting out method (12).

Analysis

Primers and PCR conditions. Primers were employed as described by Kayser et al (8) or Schneider et al (13). Monoplex amplification: DYS385 (HEX) (13); triplex amplification: DYS391 (NED), DYS392 (FAM), DYS393 (FAM) (8); quadruplex amplification: DYS19 (NED), DYS389I/II (NED), DYS390 (FAM) (8).

Monoplex-amplification (DYS385). Polymerase chain reaction (PCR) reaction mixture: 1.5 ng template DNA was amplified in a total reaction volume of 10 μL including 1 μL GeneAmp 10×PCR buffer (Applied Biosystems, Foster City, CA, USA), 0.5 μmol/L each primer of DYS385, 1 mol/L betain (Sigma, St. Louis, MO, USA), 0.2 mmol/L each nucleotide (Pharmacia, Uppsala, Sweden) and 0.8 U AmpliTaq Gold DNA Polymerase (Perkin Elmer Deutschland Gmbh, Weiterstadt, Germany). The cycling conditions were as follows: initial incubation at 95°C for 11 min, followed by a touchdown PCR of 3 cycles: 94°C for 30 s, 59°C for 30 s, 72°C for 60 s, 3 cycles: 94°C for 30 s, 58°C for 30 s, 72°C for 60 s, 3 cycles: 94°C for 30 s, 57°C for 30 s, 72°C for 60 s, 27 cycles: 94°C for 30 s, 56°C for 30 s, 72°C for 60 s, extension at 72°C for 7 min, and a final extension step at 60°C for 45 min in a GeneAmp PCR System 9700 Thermocycler (Perkin Elmer).

Triplex-amplification (DYS391, DYS392, DYS393). PCR reaction mixture consisted of 2 ng template DNA in a 10 μL reaction volume, 1 μL GeneAmp 10×PCR buffer (Perkin Elmer), 0.1 μmol/L each primer of DYS391, 0.6 μmol/L each primer of DYS392, 0.15 μmol/L each primer of DYS393, 0.85 mol/L betain (Sigma), 0.2 mmol/L each nucleotide (Pharmacia) and 0.8 U AmpliTaq Gold DNA Polymerase (Perkin Elmer). The cycling

Table 1. Y-chromosomal short tandem repeat frequencies in populations from Germany (G), India (I), and Turkey (T)

DYS19, DYS385, DYS389_I, YS389_II, DYS390, DYS391, DYS392, DYS393	G	I	T
13,11-12,12,28,22,11,16,11			1
13,11-14,13,29,23,10,13,13	1		
13,11-14,13,30,25,10,13,12			1
13,11-18,12,30,24,10,11,12			2
13,12-12,14,31,22,10,15,12			1
13,12-13,13,29,23,10,14,13			1
13,12-16,12,28,22,10,15,13			1
13,12-18,12,28,23,11,11,12			1
13,12-18,12,29,24,10,11,12			1
13,12-18,13,31,24,10,11,12			1
13,12-19,12,29,25,10,11,12			1
13,12-19,13,30,23,10,11,12			1
13,13-15,15,31,22,10,15,13			1
13,13-18,13,30,23,10,11,13			1
13,14-16,13,29,22,10,15,13			1
13,14-17,12,30,24,10,11,13			1
13,14-17,14,30,22,10,15,13			1
13,14-18,13,30,25,10,11,13			1
13,15-17,12,30,22,10,11,13			1
13,15-19,13,30,24,10,11,13			1
13,15-21,14,31,23,10,12,13		2	
13,16-16,13,30,23,10,11,13	1		
13,16-16,13,32,24,10,11,13			1
13,16-17,12,28,23,11,12,12		1	
13,16-17,12,29,24,10,11,12			1
13,16-17,12,30,22,10,11,13			5
13,16-17,13,30,24,10,11,13			1
13,16-18,13,30,23,10,11,13			1
13,16-18,13,30,23,11,11,13			1
13,16-18,13,30,24,10,11,13			2
13,16-18,13,32,23,10,11,13	1		
13,16-18,13,32,24,10,11,13			1
13,16-18,15,32,24,10,11,13			1
113,16-19,13,30,24,10,11,13			1
13,16-19,13,30,24,9,11,14			1
13,16-19,14,32,25,10,11,14	1		
13,17-17,13,30,24,10,11,13			1
13,17-18,13,30,24,10,11,13			1
13,17-18,13,30,24,9,11,14	1		
13,17-18,13,31,22,10,11,13			1
13,17-18,13,32,24,10,11,13			1
13,17-18,13,32,25,10,11,13			1
13,17-19,13,30,24,10,11,14			1
13,17-20,14,31,24,10,11,13			1
14,10-15,13,31,24,11,14,13	1		
14,10-15,14,31,24,10,15,13	1		
14,11-11,13,29,24,11,13,13			1
14,11-13,12,28,23,10,13,13	1		
14,11-13,13,29,23,11,13,13	2		
14,11-13,13,29,24,10,13,12			1
14,11-13,13,29,24,11,13,13	1		
14,11-13,13,29,24,11,14,13	1		
14,11-13,13,30,25,11,13,12			1
14,11-13,14,30,23,11,15,14			1
14,11-14,12,29,23,11,13,13	1		
14,11-14,13,28,23,10,13,13	1		
14,11-14,13,28,24,11,13,13	1		
14,11-14,13,28,24,11,14,12		1	2
14,11-14,13,28,24,11,14,13	1		
14,11-14,13,29,22,11,13,11	1		
14,11-14,13,29,22,11,13,12	1		
14,11-14,13,29,23,10,13,13	1		
14,11-14,13,29,23,11,13,13	2		1
14,11-14,13,29,23,11,13,14	1		
14,11-14,13,29,24,10,13,12	1		1
14,11-14,13,29,24,10,13,13	5		
14,11-14,13,29,24,10,14,13	1		
14,11-14,13,29,24,11,13,12			2
14,11-14,13,29,24,11,13,13	6		1
14,11-14,13,29,25,11,13,13	2		
14,11-14,13,30,23,11,12,13	1		
14,11-14,13,30,23,11,13,13	1		
14,11-14,13,30,24,10,13,13	1		
14,11-14,13,29,23,10,14,13	1		
14,11-14,14,29,24,11,14,12			1
14,11-14,14,29,24,12,14,12			1
14,11-14,14,30,24,11,13,13	1		
14,11-14,14,30,24,11,14,14			1
14,11-14,14,30,24,12,13,12			1
14,11-15,12,27,24,10,14,12			1
14,11-15,13,28,24,10,15,12			1
14,11-15,13,28,24,11,13,12			1
14,11-15,13,29,23,10,13,13			1
14,11-15,13,29,23,11,13,13	1		
14,11-15,13,29,23,12,14,13			1
14,11-15,13,29,24,10,13,13	3		
14,11-15,13,29,24,11,13,12	1		
14,11-15,13,29,24,11,13,13	3		
14,11-15,13,29,24,11,14,13	1		
14,11-15,13,30,24,10,13,12			1
14,11-16,13,28,24,10,14,12			1
14,11-17,13,29,24,12,13,13	1		
14,12-13,13,29,23,10,14,13			7
14,12-14,12,28,23,10,11,13	1		
14,12-14,12,29,23,10,12,13	1		
14,12-14,13,29,23,10,14,13			2
14,12-14,13,29,23,11,13,13	1		
14,12-14,13,29,23,11,14,13	1		
14,12-14,13,29,23,8,13,13	1		
14,12-14,13,29,24,10,13,13	1		
14,12-14,13,29,24,11,13,13	1		
14,12-14,13,29,24,11,14,12			1
14,12-14,13,29,25,10,13,12			1
14,12-14,13,30,23,10,14,13			2
14,12-14,13,30,24,11,13,12	1		
14,12-14,13,30,24,11,13,13	1		
14,12-14,13,30,25,11,11,13	1		
14,12-14,13,31,24,10,13,13	1		
14,12-15,12,28,24,11,13,12			1
14,12-15,13,28,24,11,13,13	1		
14,12-15,13,30,24,10,13,13	1		
14,12-15,14,30,24,11,13,12	1		
14,12-15,14,31,23,10,11,13			1
14,12-16,13,29,24,11,13,12			3
14,12-16,13,29,24,11,14,12			1
14,12-16,13,30,23,10,11,12			2
14,12-16,13,30,24,10,11,12			1
14,12-16,14,32,22,11,11,12			1
14,12-17,13,29,24,11,13,12			2
14,12-17,14,30,23,10,12,12			1
14,12-18,13,31,23,10,11,13			1
14,12-18,13,31,24,10,11,12			1
14,13-13,12,28,22,10,11,14	1		
14,13-13,12,28,22,11,11,13	1		
14,13-13,12,28,22,11,11,14		1	
14,13-13,13,29,23,10,14,13			1
14,13-13,14,31,22,10,11,12			2
14,13-13,14,31,23,10,11,12			1
14,13-14,12,27,22,10,11,13	1		
14,13-14,12,28,22,10,11,13	8		
14,13-14,12,28,22,10,11,14	2		
14,13-14,12,28,23,10,11,13	3		2
14,13-14,13,29,23,10,11,13	2		
14,13-14,13,30,23,10,11,13			1
14,13-14,13,30,24,10,11,12			1
14,13-14,14,30,22,10,11,12			1
14,13-15,12,28,22,10,11,13	1		
14,13-15,12,28,24,10,12,13			1
14,13-15,12,31,22,10,11,13	1		
14,13-15,13,29,24,10,12,12			1
14,13-15,13,30,23,10,11,12			1
14,13-16,12,28,22,10,14,11		2	
14,13-16,13,29,24,10,11,12			1
14,13-16,13,29,24,11,11,12			1
14,13-16,13,30,23,10,11,12			3
14,13-16,13,30,24,10,11,12			1
14,13-16,14,30,24,10,11,12			1
14,13-16,14,31,23,10,11,12	1		1
14,13-16,14,31,23,10,11,13			1
14,13-17,12,28,23,10,11,12			1
14,13-17,12,28,24,10,11,12			1
14,13-17,13,29,23,10,17,13		2	
14,13-17,13,30,22,9,11,12			1
14,13-17,13,30,23,10,11,12			1
14,13-17,13,30,23,11,11,12			1
14,13-17,13,31,23,11,11,12			1
14,13-17,14,31,23,10,11,12			1
14,13-17,14,31,26,10,11,12			1
14,13-18,12,28,23,10,11,12			1
14,13-18,12,29,23,10,11,12			1
14,13-18,13,28,24,10,10,15		1	
14,13-18,13,29,24,10,10,14		2	
14,13-18,13,29,24,10,10,15		3	
14,13-18,13,31,23,10,11,12			4
14,13-18,14,30,23,10,10,14		1	
14,13-18,14,31,23,10,12,12			1
14,13-19,12,29,23,10,11,12			1
14,13-19,13,29,23,10,11,12			1
14,13-19,13,29,24,10,11,13		1	1
14,13-19,14,30,23,10,11,12	1		
14,13-19,14,31,24,10,11,12			1
14,13-20,14,31,22,11,12,13		1	
14,13-21,13,29,23,10,11,12			1
14,13-22,13,30,23,10,12,13		1	
14,14-14,12,28,22,10,11,12	1		
14,14-14,12,28,22,10,11,13	2		
14,14-14,12,28,22,10,12,12	2		
14,14-14,12,28,23,10,11,13	2		
14,14-14,13,28,23,11,13,13			1
14,14-14,13,29,22,10,11,13	1		
14,14-15,12,28,23,10,11,12			1
14,14-15,13,29,25,10,11,12			2
14,14-15,13,30,22,10,11,12			1
14,14-16,13,29,23,10,13,13			1
14,14-16,14,30,25,10,11,12			1
14,14-16,14,31,22,10,11,12	1		
14,14-16,14,31,23,10,11,12			1
14,14-16,14,31,25,10,11,12			1
14,14-17,13,30,23,10,11,12	1		
14,14-17,13,30,25,10,11,12			1
14,14-17,15,32,23,10,13,13			1
14,14-18,12,28,22,10,14,11		1	
14,14-18,13,29,23,9,11,12			1
14,14-18,14,30,23,10,10,14		1	
14,14-19,13,28,23,10,10,14		1	
14,14-21,14,31,23,10,10,14		1	
14,15-16,13,30,24,10,13,13			1
14,15-16,13,31,24,10,13,13			1
14,15-17,12,28,23,10,12,12			1
14,15-17,14,31,23,10,10,14			1
14,15-18,14,30,23,10,11,13			1
14,15-19,14,30,23,10,11,12		5	
14,16-18,14,30,23,10,11,12		1	
14,16-19,12,29,25,10,11,13	1		
14,17-18,12,30,24,10,11,13			1
14,17-18,13,29,23,10,14,11			1
14,17-18,13,30,23,10,11.1,13	1		
14,9-14,13,29,23,11,13,13	1		
15,10-14,13,29,24,11,11,13		1	
15,10-14,13,29,25,11,11,13		1	
15,10-14,13,29,25,12,11,13		2	
15,10-16,12,28,24,10,11,12	1		
15,10-19,13,30,22,10,11,12			1
15,10-19,13,30,23,10,11,12		1	
15,11-11,13,30,25,10,11,13		1	
15,11-11,14,31,23,10,11,13		1	
15,11-13,13,30,25,11,11,13			1
15,11-13,14,29,23,11,14,14	1		
15,11-13,14,30,23,10,14,14	1		

Table 1. Continued...

DYS19, DYS385, DYS389_I, YS389_II, DYS390, DYS391, DYS392, DYS393	G	I	T
15,11-13,14,30,23,11,14,14	1		
15,11-14,12,28,24,10,11,13			1
15,11-14,13,29,22,10,13,13	1		
15,11-14,13,29,23,10,13,12			1
15,11-14,13,29,24,10,13,13	1		
15,11-14,13,29,24,11,13,12			1
15,11-14,13,29,24,11,13,13	1		
15,11-14,13,29,25,10,11,13	1		
15,11-14,13,29,25,11,11,13		1	1
15,11-14,13,30,24,10,11,13		1	
15,11-14,13,30,24,10,13,13	1		
15,11-14,13,30,25,10,11,13	2		
15,11-14,13,30,25,11,11,13	1		1
15,11-14,13,30,26,11,13,13	1		
15,11-14,13,31,25,10,11,13		1	
15,11-14,13,31,25,11,11,13			1
15,11-14,13,31,26,9,11,14	1		
15,11-14,13,32,25,10,11,13		1	
15,11-14,13,32,25,11,11,13			1
15,11-14,14,30,24,11,13,13	1		
15,11-14,14,31,24,10,11,13		1	
15,11-14,14,31,24,12,13,12			1
15,11-14,14,31,25,11,11,14		1	
15,11-14,14,32,25,11,11,13		1	
15,11-15,13,29,24,10,13,13	1		
15,11-15,13,30,25,10,11,13	1		
15,11-15,13,30,25,11,11,13		2	
15,11-15,13,30,25,12,11,14		1	
15,11-15,13,30,26,11,11,13		1	
15,11-15,13,31,24,10,11,14			1
15,11-15,13,32,24,10,11,14			1
15,11-15,14,30,24,11,13,13	1		
15,11-16,14,30,24,10,13,12			1
15,11-16,14,32,25,10,11,13		1	
15,12-12,12,29,24,10,11,13	1		
15,12-12,13,29,22,10,11,13			1
15,12-12,13,30,25,10,11,14	1		
15,12-12,14,31,23,10,13,13			1
15,12-13,12,28,22,10,11,14	1		
15,12-13,12,29,21,10,11,14	1		
15,12-13,13,30,23,11,13,13			2
15,12-13,14,30,24,10,13,13			1
15,12-14,12,29,23,10,13,13			1
15,12-14,12,30,22,10,11,14			1
15,12-14,13,29,25,11,13,12			1
15,12-14,13,30,25,11,11,13	1		
15,12-14,13,30,25,11,12,13	1		
15,12-14,13,31,24,10,11,12			1
15,12-14,13,31,25,10,11,13		2	
15,12-14,14,29,25,11,11,13			1
15,12-15,12,28,24,11,13,12			1
15,12-15,13,29,23,10,11,12			1
15,12-15,13,30,24,10,11,13			1
15,12-15,13,30,24,11,13,12			1
15,12-16,13,28,23,10,11,12			1
15,12-16,13,29,23,10,11,12			3
15,12-16,13,29,23,11,11,12			1
15,12-16,13,30,22,10,11,12			1
15,12-17,12,28,24,10,11,12		1	
15,12-17,13,29,23,11,11,12			1
15,12-18,14,30,23,10,11,12			1
15,13-13,12,28,22,10,11,14	1		
15,13-13,13,29,24,11,11,14			1
15,13-13,14,28,22,9,11,12			1
15,13-14,12,28,20,10,11,14	1		
15,13-14,12,28,22,10,11,14			1
15,13-14,13,29,22,10,11,14			1
15,13-14,13,29,23,10,11,13	1		
15,13-14,13,30,22,10,11,12			1
15,13-14,13,30,25,10,11,13			1
15,13-14,13,31,22,10,11,14			1
15,13-15,12,29,21,10,11,14			1
15,13-15,12,30,21,10,11,15			1
15,13-15,13,29,23,9,11,15			1
15,13-15,13,30,23,9,11,14			1
15,13-15,14,30,21,10,11,14			1
15,13-16,11,28,21,10,11,15			1
15,13-16,12,28,21,10,11,14			1
15,13-16,12,28,23,10,12,12			1
15,13-16,12,29,21,11,11,14	1		
15,13-16,12,29,21,8,11,15			2
15,13-16,12,29,23,10,11,12			1
15,13-16,12,31,23,9,11,12			1
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17,16-19,13,27,21,10,11,14	1		
17,16-21,12,29,22,10,12,14		1	
17,18-Var.,13,31,21,10,11,12			1

conditions were as follows: after an initial incubation at 95°C for 11 min followed by 31 cycles with 94°C for 1 min, 55°C for 1 min, 72°C for 2 min and a final extension step at 60°C for 45 min in a GeneAmp PCR System 9700 Thermocycler (Perkin Elmer).

Quadruplex-amplification (DYS19, DYS389I, DYS389II, DYS390). PCR reaction mixture contained 2 ng template DNA in a 10 μL reaction volume mixed with 1 μL GeneAmp 10×PCR buffer (Perkin Elmer), 0.25 μmol/L each primer of DYS19, 0.125 μmol/L each primer of DYS389I/II, 0.104 μmol/L each primer of DYS390, 1 mol/L betain (Sigma), 0.2 mmol/L each nucleotide (Pharmacia), and 0.8 U AmpliTaq Gold DNA Polymerase (Perkin Elmer). PCR cycling conditions were as described for the triplex amplification.

Electrophoresis

Capillary electrophoresis was carried out on an ABI 310 genetic analyzer (Applied Biosystems) with the internal standard CXR 60-400 (Promega, Madison, USA). The allele attribution was made by comparison with allelic ladders constructed from reference samples kindly provided by Lutz Roewer, Institut f. Rechtsmedizin, Berlin. Correct allele calling was additionally assured by successful participation in the quality control tests of the Y chromosome STR haplotype reference database (*http://ystr.charite.de*).

Results

Population Studies

Haplotypes of DYS19, DYS385 I+II, DYS389 I+II, DYS390, DYS391, DYS392 and DYS393 are shown in Table 1. There were only few (thirteen) haplotypes that could be observed in 2 of the 3 populations. Only 1 haplotype was found in all 3 groups.

In 108 Jat Sikhs, 68 different haplotypes were observed, 245 haplotypes were observed in 281 Turkish males, and 133 haplotypes were found in 166 German Caucasians.

The most frequent haplotype in Jat Sikhs was found in 13 individuals (12%), while in Turks and Germans the highest frequencies were 2.5% and 4.8%, respectively. These data show a very high degree of diversity in Turks (14,15).

Interestingly, a German individual carried a DYS392*11.1 allele, which was caused by an insertion of A (adenine) (data not shown). The position of the mutant allele *11.1 in comparison to the allelic ladder of locus DYS392 is illustrated in Figure 1.

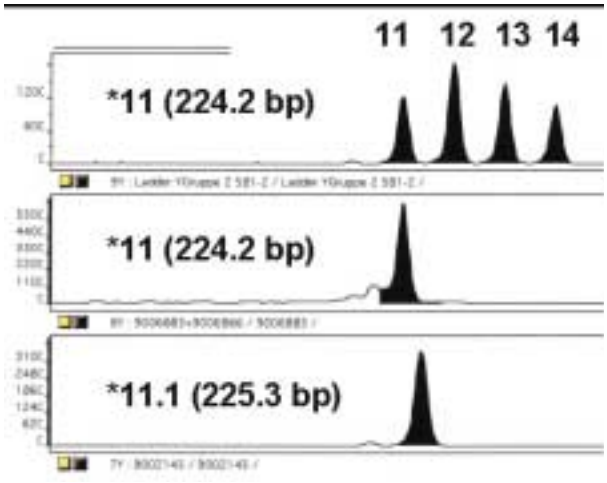

Figure 1. New variant allele DYS392*11.1 caused by insertion of A (adenine).

A Turkish father carried two alleles (*15 and *16) at locus DYS19 (Fig. 2); he inherited his haplotype to his son. Duplicated alleles have been observed at various Y-STR loci (8,16).

Application to Paternity Testing and Impact on Forensic Casework

Application of Y-STRs to a deficient paternity case. Because an alleged father could not be reached for testing, the court ordered his two brothers to be tested. An excerpt from the typing results is shown in Table 2. Strong evidence for kinship was found in the Rhesus blood group system, in DNA minisatellite polymorphisms (despite the fact that there existed 5 RFLP alleles in the pedigree at loci D12S11 and D5S110!), and in Y-chromosomal STRs. The observed Y-chromosomal STR haplotypes revealed that the two men were half-brothers (sharing the same mother but having different fathers). We have calculated that the probability for P. being the uncle of the pursuing child exceeded 99.99% (based on the analysis of 5 conventional markers, 6 DNA minisatellite polymorphisms, and 8 autosomal STR polymorphisms).

Allelic drop-out at the Amelogenin locus. In a paternity test, 3 red cell antigen systems (ABO, Rh, MNSs), 3 minisatellite polymorphisms (D7S21, D12S11, D16S309), and 17 autosomal STR polymorphisms were analyzed. By employing 2 PCR profiling kits (SGM Plus from Applied Biosystems and Power Plex 16 from Promega) a father from Morocco and his son were genotyped as female, as demonstrated by the absence of respective 'Y peaks' (Fig. 3). In order to determine whether this observation was possibly caused by an erroneous sample mix-up, Y-chromosomal STRs were typed additionally. Because we were successful in typing Y-chromosomal STR markers, we concluded that the unexpected genotyping was caused by a mutation at a respective primer binding site. In a similar case described recently (17) the potential investigative problems were extensively elaborated.

In our case, we received the alleged father's blood sample first, and after considering the typing results, concerns were raised regarding sample mix-up or manipulations. These concerns could be ruled out after typing the samples from the mother and the child: no exclusion from paternity was found in 23 autosomal polymorphisms or in 9 Y-chromosomal STRs.

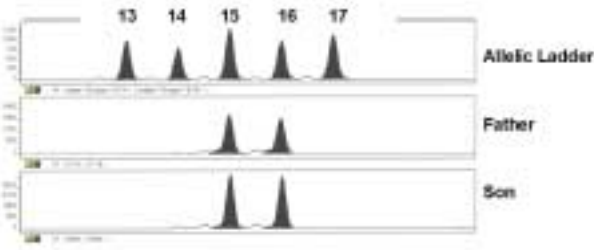

Figure 2. Duplication at locus DYS 19.

Table 2. Excerpt from typing results in a deficient paternity case

System / Locus	Child	Mother	Uncle P.	Uncle C.
Serology				
Rhesus	(C)cD.Ee	CcD.ee	(C)cD.EE	ccD.Ee
DNA-minisatellite-systems (sizes in kilobases)				
D1S7	6.1/4.7	6.1/4.8	9.4/4.3	9.4/5.6
D7S21	9.0/**6.4**	9.0/5.7	8.1/6.0	**6.4**/6.0
D12S11	10.8/7.9	7.9/6.7	10.3/4.9	9.4/9.2
D2S44	**4.3**/3.3	3.3	**4.3**/2.6	3.1/2.7
D16S309	**4.0**/2.1	2.7/2.1	2.8/2.7	**4.0**/2.4
D5S110	7.3/5.8	7.3/2.8	5.3/2.7	6.9/4.1
Autosomal-microsatellite-systems				
D3S1358	16/17	16	17	17
vWA	14/18	16/18	14/15	16/17
FGA	21/24	20/21	24	24
TH01	6	6/9.3	6/9	6/7
TPOX	10/11	10/11	8/9	8/11
CSF1PO	11/12	12/12	11	11/13
D5S818	11/13	10/11	13	9/13
D13S317	9/12	9	12/13	11/12
Y-chromosomal-microsatellite-systems				
DYS19	14		14	13
DYS389.I	10		10	9
DYS389.II	26		26	27
DYS390	23		23	23
DYS391	10		10	8
DYS392	11		11	11
DYS393	12		12	13

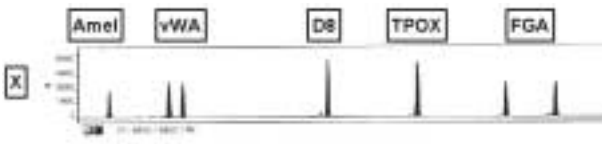

Figure 3. Genotyping results of male DNA. Allelic dropout at the amelogenin locus in a PowerPlex 16 profile; failure to detect the Y homolog.

Discussion

The compilation of haplotypes shows that the vast majority of haplotypes was observed only once, reflecting the enormous genetic heterogeneity, especially in the male Turkish population. This confirms a previous study by Decorte et al (14) and corroborates "early" immunoglobulin allotype data (15). Duplicated alleles at Y-STR loci were described earlier (8,16). It was assumed that they reflected duplication events of larger regions, including the STR locus followed by a mutation in the number of repeats. For DYS19 a frequency of 0.12% was reported (16).

As a failure to detect the Y-homolog at the "amelogenin" locus was described (17), the genetic determination of sex based on the amelogenin sequences from specimens of unknown origin (e.g., crime scene samples) must not be considered infallible.

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