

Allele and Haplotype Frequencies of Y-Short Tandem Repeat Loci in Turkey

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Aim. To assess the frequency distribution of six Y-chromosomal specific short tandem repeat (STR) polymorphisms and frequencies of haplotypes in Turkish population.

Methods. Blood samples were taken from 200 unrelated Turkish men from different regions of Turkey, who were involved in legal proceedings concerning paternity. The DNA was amplified and typed by use of Y-Plex 6 kit containing 6 Y-STR markers (DYS393, DYS19, DYS389 II, DYS390, DYS391, and DYS385). Capillary electrophoresis was carried out on an ABI 310 Genetic Analyzer.

Results. Allele frequency profiles of the Turkish men showed no significant differences compared with previously published results for Swiss and German populations. The range of gene diversity was determined between 48.3% for DYS391 and 96.0% for DYS385. Of 184 determined haplotypes, 170 occurred once, and the rest more than once. The most common haplotypes had a frequency of 1.5%.

Conclusion. High degree of haplotype diversity in Turkish population makes analysis of Y-STRs a useful tool in forensic cases.

Key words: alleles; gene frequency; haplotypes; forensic medicine; polymorphism (genetics); tandem repeat sequences; Turkey; Y chromosome

DNA polymorphism of the human Y-chromosome is frequently used in forensic casework (1,2). For the past few years, new Y-chromosome short tandem repeat (Y-STR) markers have been described and population data published (3,4). Some international groups have carried out collaborative studies to characterize Y-chromosome STRs (5-7).

Although Y-STR data concerning mutational rates have been limited so far, recent studies have indicated similar average mutational rates for autosomal STR loci commonly used in forensic sciences (8-11). Such mutations are rare: their frequency in the commonly used loci has been measured at approximately 2×10^{-3} per locus per generation (9,12).

Y-chromosomal STRs are characterized by male inheritance pattern and remain stable in a given paternal lineage over many generations (5,13). This makes the system very useful in forensic casework, such as rape cases where a mixture of male and female DNA is found, and paternity deficiency cases where the putative father is not available. It is also used in kinship analysis and human identification (5,8,13-15). In the investigation of sexual assault cases, Y-STRs are especially suitable, because they provide a male specific DNA profile by which the problems of mixed stain interpretation are avoided (6,8,16-20).

In our study, we analyzed the frequency distribution of six Y-chromosomal specific STR polymorphisms and frequencies of haplotypes in Turkish population.

Material and Methods

Sample Preparation

Blood samples were taken from 200 unrelated Turkish men from different regions of Turkey, who were involved in legal proceedings concerning paternity. DNA extraction was performed by salting out procedure or Chelex extraction (21,22).

Polymerase Chain Reaction (PCR) Amplification

DNA was amplified and typed by use of the Y-Plex 6 kit, which contains 6 Y-STR markers (DYS393, DYS19, DYS389 II, DYS 390, DYS 391, and DYS 385; Reliagene Technologies, Inc., New Orleans, LA, USA), according to the manufacturer's protocol. The polymerase chain reaction (PCR) mixture contained 2-10 ng DNA amplified in a total reaction volume of 25 μ L, including 5 μ L 5X Y-PLEX™ 6 Primer Mix, 0.5 μ L AmpliTaq Gold (5 u/ μ L) DNA Polymerase (Applied Biosystems by Roche Molecular Systems Inc., Branchburg, NJ, USA), and double distilled H₂O. In the PCR protocol, DNA was initially denatured at 95 °C for 10 min. This was followed with 94 °C for 30 s, 59 °C for 1 min, and 70 °C for 1 min. The amplification reaction was completed after extension at 60 °C for 60 min. A total of 30 cycles was carried out in a MJ Research PTC-200 Thermal Cycler (MJ Research Inc., Watertown, MA, USA).

Electrophoresis

Capillary electrophoresis was carried out on an ABI 310 Genetic Analyzer (Applied Biosystems, Foster City, CA, USA). AI-

lele designations were determined according to the fragment size in base pairs, using the allele designation guide provided by the manufacturer (Reliagene Technologies, Inc., New Orleans, LA, USA).

Results

A total of 200 unrelated Turkish men were analyzed for allele frequencies and gene diversity (Tables 1 and 2). At the DYS391 locus, the allele 10 was the most frequent allele among all the loci. Our data for DYS385 locus showed a flat distribution in which most of the genotypes had almost equal frequencies.

The dominant allele combination at the DYS385 locus "11-14" had a frequency of 0.125, followed by

"13-14" with 0.07, and "13-16" and "14-15" with 0.05, respectively. In our study, the total number of genotypes observed at DYS385 locus was 47.

We noted haplotype frequencies and compared them with Y-chromosomal STR Haplotype Reference Database (YHRD) and Henke's (15) results for German Turks (Table 3). We observed a total of 184 haplotypes. Fourteen haplotypes occurred more than once, 12 of them were shared by two individuals, and 2 by three, whereas the remaining 170 combinations were observed only once. The most common haplotypes, "13/16/31/24/11/14-15" and "14/15/29/22/10/13-14," repeated three times, with a frequency of 1.5%. When we excluded the DYS385 locus, we found only 122 haplotypes. Out of these, 34 occurred more than once and 88 only once.

A total of 107 haplotypes were unique for our population (58%) when compared with YHRD data (Table 3). Haplotype "13/14/29/23/11/11-14," which was observed once in our study, was found 162 times in the YHRD database.

Discussion

Except the DYS393 locus, the gene diversity values were similar to those published previously for Swiss (2), German, and Chinese populations (7,23).

The most common alleles in Turkish population were 13, 14, 29, 23, and 10 for the loci DYS393, DYS19, DYS389II, DYS390, and DYS391, respectively. Likewise, allele 13 for DYS393, and allele 10 for DYS391 were the most frequent modal alleles found in the previous survey of African-American, European-American, and Hispanic-American populations. Whereas allele 21 was the most common allele (46%) in African-American population, it was the least common (3%) in the Turkish population for DYS390 locus (24).

The genotype "11-14" at the locus DYS385 was the most frequent allele combination (0.125) observed in our population. It was also identified as the most common genotype in a collaborative study including the data from Portugal, Spain, Italy, Greece, Germany, Netherlands, England, Norway, and Germany, and it varied from 0.099 in the Greek to 0.409 in the Dutch population (5). The informative value of the locus decreases as its frequency increases in a certain genotype, such as that for allele 10 at DYS391. This effect was relatively smaller at the locus DYS385 in our study.

The decreasing number of haplotype combinations from 184 to 122, after exclusion of DYS385 locus, confirms this locus as one of the most informative Y-STR loci so far (5,23).

For Y-chromosome specific markers, the haplotype diversity for the whole array of loci is more important than the allele distribution for each marker, because the haplotype diversity value of the Y-chromosome corresponds to the value of the power of discrimination and the chance of exclusion for unrelated men (25).

Table 1. Allele frequencies of 5 Y-STRs (DYS393 / DYS19 / DYS389II / DYS390/ DYS391) and gene diversity in Turkish population

Allele	Frequency				
	DYS 393	DYS 19	DYS 389 II	DYS 390	DYS 391
9					0.050
10					0.660
11	0.030				0.280
12	0.415				0.010
13	0.430	0.100			
14	0.115	0.365			
15	0.010	0.345			
16		0.170			
17		0.020			
21				0.030	
22				0.135	
23				0.445	
24				0.275	
25				0.105	
26				0.010	
27			0.015		
28			0.095		
29			0.315		
30			0.285		
31			0.210		
32			0.060		
33			0.020		
Gene diversity*	0.63	0.71	0.76	0.70	0.48

* $1-\sum p_i^2 - p_i$ allele frequencies.

Table 2. Allele frequencies and gene diversity of the DYS 385 locus in Turkish population

Allele	Frequency	Allele	Frequency
8-8	0.020	13-19	0.020
9-11	0.005	14-14	0.025
9-13	0.005	14-15	0.050
10-10	0.005	14-16	0.020
11-11	0.025	14-17	0.015
11-13	0.030	14-18	0.005
11-14	0.125	14-19	0.005
11-15	0.045	14-20	0.005
11-16	0.005	15-15	0.020
12-12	0.020	15-16	0.010
12-13	0.040	15-18	0.005
12-14	0.035	15-19	0.005
12-15	0.040	15-20	0.005
12-16	0.020	16-16	0.010
12-17	0.010	16-17	0.025
12-18	0.010	16-18	0.025
12-19	0.010	16-19	0.005
12-20	0.010	17-17	0.010
13-13	0.015	17-18	0.005
13-14	0.040	17-20	0.010
13-15	0.050	18-19	0.005
13-16	0.030	18-20	0.005
13-17	0.045	19-20	0.005
13-18	0.020	Gene diversity*:	0.96

* $1-\sum p_i^2 - p_i$ allele frequencies.

Table 3. Y-chromosomal short tandem repeat (STR) haplotype frequencies of the loci DYS393/DYS19/DYS389II/DYS390/DYS391/DYS385 in Turkish population and comparison with Y-STR Haplotype Reference Database (YHRD)* and German Turks[†] data[‡]

Haplotypes	Observed in [‡]			Haplotypes	Observed in [‡]		
	Turks No. (%)	YHRD No.	German Turks No.		Turks No. (%)	YHRD No.	German Turks No.
11/13/28/22/11/12-15	1 (0.5)			12/15/30/22/10/16-18	1 (0.5)	1	
11/15/29/23/10/12-16	1 (0.5)			12/15/30/23/10/13-16	2 (1.0)	4	
11/15/30/23/10/12-13	1 (0.5)			12/15/30/24/10/13-15	1 (0.5)		
11/15/30/23/10/13-18	1 (0.5)		1	12/15/31/24/11/12-14	1 (0.5)		
11/15/31/23/10/13-17	1 (0.5)			12/15/31/24/11/11-15	1 (0.5)		
11/16/31/23/10/12-16	1 (0.5)		1	12/15/31/24/11/17-20	1 (0.5)		
12/13/28/23/11/13-18	1 (0.5)			12/16/29/23/9/13-15	1 (0.5)	1	
12/13/29/25/10/12-18	1 (0.5)			12/16/30/23/10/15-15	1 (0.5)		
12/13/30/23/10/13-15	1 (0.5)			12/16/31/24/11/11-14	2 (1.0)		
12/14/28/24/10/11-15	1 (0.5)		1	12/16/32/22/10/13-15	1 (0.5)	2	
12/14/28/24/10/11-18	1 (0.5)			12/16/32/23/11/15-20	1 (0.5)		
12/14/29/22/10/13-19	1 (0.5)			13/13/28/25/10/14-17	1 (0.5)		
12/14/29/23/10/11-13	1 (0.5)			13/13/29/24/10/16-17	1 (0.5)	3	
12/14/29/23/10/11-14	2 (1.0)	1		13/13/29/24/10/16-19	1 (0.5)	2	
12/14/29/23/10/9-13	1 (0.5)			13/13/30/22/10/13-16	1 (0.5)		
12/14/29/23/10/12-15	1 (0.5)	1		13/13/30/22/10/14-16	1 (0.5)	1	
12/14/29/23/10/13-17	1 (0.5)	7		13/13/30/22/10/16-17	2 (1.0)		5
12/14/29/23/10/12-20	1 (0.5)			13/13/30/24/10/14-17	1 (0.5)		1
12/14/29/23/10/16-18	1 (0.5)			13/13/30/24/10/16-18	1 (0.5)	43	2
12/14/29/23/10/12-14	1 (0.5)			13/13/30/25/10/15-19	1 (0.5)		
12/14/29/23/11/8-8	1 (0.5)			13/13/31/23/10/15-16	1 (0.5)		
12/14/29/23/11/11-15	1 (0.5)			13/13/31/24/10/16-17	1 (0.5)	5	
12/14/29/23/11/12-15	1 (0.5)			13/13/31/24/10/17-20	1 (0.5)		1
12/14/27/23/10/13-17	1 (0.5)			13/13/32/24/10/11-14	1 (0.5)		
12/14/29/24/10/12-15	1 (0.5)			13/13/32/24/10/17-17	1 (0.5)	1	
12/14/29/24/10/13-14	2 (1.0)			13/14/28/22/10/13-14	1 (0.5)	97	
12/14/29/24/10/13-16	1 (0.5)	1	1	13/14/28/23/10/11-13	1 (0.5)	4	
12/14/29/24/11/12-13	1 (0.5)			13/14/28/23/10/11-14	1 (0.5)	22	
12/14/29/25/10/12-15	1 (0.5)			13/14/28/23/10/12-13	1 (0.5)	2	
12/14/30/22/9/13-19	1 (0.5)			13/14/28/24/11/13-13	1 (0.5)		
12/14/30/23/9/13-18	1 (0.5)			13/14/29/23/10/10-10	1 (0.5)		
12/14/30/23/10/11-14	1 (0.5)	1		13/14/29/23/10/11-11	1 (0.5)	5	
12/14/30/23/10/12-15	1 (0.5)			13/14/29/23/10/11-13	1 (0.5)	9	
12/14/30/23/10/13-16	2 (1.0)	6		13/14/29/23/10/12-13	2 (1.0)	4	7
12/14/30/23/10/14-16	1 (0.5)			13/14/29/23/10/13-18	1 (0.5)	2	
12/14/30/23/10/12-19	1 (0.5)			13/14/29/23/10/14-16	1 (0.5)	2	1
12/14/30/23/10/12-17	1 (0.5)	3	1	13/14/29/23/11/11-11	1 (0.5)	162	
12/14/30/23/10/13-17	1 (0.5)	2	1	13/14/29/23/11/11-15	1 (0.5)	30	
12/14/30/23/10/13-18	1 (0.5)	3		13/14/30/23/11/11-15	1 (0.5)	12	
12/14/30/23/10/14-18	1 (0.5)			13/14/30/23/11/13-18	1 (0.5)		
12/14/30/23/11/12-16	1 (0.5)			13/14/31/21/10/16-16	1 (0.5)		
12/14/30/23/11/12-18	1 (0.5)			13/14/31/23/10/14-17	1 (0.5)	1	
12/14/30/23/11/13-16	1 (0.5)			13/14/30/24/10/16-16	1 (0.5)		
12/14/30/23/11/14-19	1 (0.5)			13/14/31/24/10/18-19	1 (0.5)	2	
12/14/30/24/10/11-14	1 (0.5)	6		13/14/33/24/9/12-14	1 (0.5)		
12/14/30/24/11/12-16	1 (0.5)			13/15/28/22/11/13-14	1 (0.5)	1	
12/14/30/25/11/11-14	1 (0.5)	6		13/15/29/22/10/12-14	1 (0.5)	4	
12/14/31/22/10/14-15	1 (0.5)	2		13/15/29/22/10/12-19	1 (0.5)		
12/14/31/23/10/13-17	2 (1.0)	6	1	13/15/29/23/10/11-14	2 (1.0)	12	
12/14/31/23/10/13-18	1 (0.5)	3	5	13/15/29/23/11/11-14	1 (0.5)	20	
12/14/31/23/10/14-20	1 (0.5)			13/15/29/24/11/13-18	1 (0.5)		
12/14/31/24/10/12-17	1 (0.5)			13/15/29/24/12/11-14	1 (0.5)		
12/14/31/24/10/13-14	1 (0.5)	1		13/15/30/23/10/12-13	1 (0.5)		
12/14/32/23/10/12-15	1 (0.5)			13/15/30/23/10/12-14	1 (0.5)	1	
12/15/27/24/9/13-14	1 (0.5)			13/15/30/23/10/14-16	1 (0.5)	5	
12/15/28/22/10/13-18	1 (0.5)			13/15/30/23/10/15-16	1 (0.5)	4	
12/15/28/24/10/14-14	1 (0.5)			13/15/30/23/10/16-18	1 (0.5)		
12/15/28/24/11/11-14	1 (0.5)			13/15/30/23/11/12-12	1 (0.5)	1	
12/15/29/23/9/13-16	2 (1.0)	10	1	13/15/30/24/10/11-14	1 (0.5)	15	
12/15/29/23/9/13-19	1 (0.5)			13/15/30/25/10/11-11	1 (0.5)		
12/15/29/23/10/8-8	1 (0.5)			13/15/30/25/11/11-15	1 (0.5)	17	
12/15/29/23/10/12-12	1 (0.5)			13/15/30/25/11/12-14	1 (0.5)	14	
12/15/29/23/10/17-17	1 (0.5)			13/15/30/26/10/14-14	1 (0.5)		
12/15/29/23/10/13-15	1 (0.5)	2		13/15/31/22/11/14-15	1 (0.5)		
12/15/29/23/10/13-19	1 (0.5)			13/15/31/24/10/17-18	1 (0.5)	1	
12/15/29/23/11/11-14	1 (0.5)	2		13/15/31/24/10/19-20	1 (0.5)		
12/15/29/24/9/8-8	1 (0.5)			13/15/31/25/11/11-14	2 (1.0)	20	1
12/15/29/24/10/12-14	1 (0.5)			13/15/31/25/11/12-13	2 (1.0)		
12/15/29/25/12/11-14	1 (0.5)			13/15/31/26/11/11-14	1 (0.5)	1	
12/15/30/22/10/15-15							

Table 3. Continued ...

13/16/29/25/10/11-14	1 (0.5)	72		14/14/28/22/10/13-13	1 (0.5)	6
13/16/29/25/11/11-11	1 (0.5)			14/14/30/23/10/14-15	1 (0.5)	
13/16/30/23/11/14-14	1 (0.5)			14/15/29/22/10/12-12	1 (0.5)	1
13/16/30/24/10/13-13	1 (0.5)			14/15/29/22/10/13-14	3 (1.5)	7
13/16/31/22/10/14-14	1 (0.5)			14/15/29/23/10/13-14	1 (0.5)	1
13/16/31/23/11/14-15	1 (0.5)			14/15/30/23/11/15-15	1 (0.5)	1
13/16/31/24/11/11-14	1 (0.5)	4		14/15/30/24/10/18-20	1 (0.5)	
13/16/31/24/11/14-15	3 (1.5)	31		14/15/30/25/11/11-15	1 (0.5)	
13/16/31/25/11/11-14	1 (0.5)	28		14/15/31/23/10/13-14	1 (0.5)	
13/16/32/22/10/16-18	1 (0.5)			14/15/31/23/11/13-14	1 (0.5)	
13/16/32/23/10/11-14	1 (0.5)	2		14/15/32/24/10/11-15	1 (0.5)	1
13/16/32/24/10/14-15	1 (0.5)	2		14/16/28/21/10/13-14	1 (0.5)	
13/16/32/24/11/15-15	1 (0.5)	1		14/16/28/21/11/13-15	1 (0.5)	
13/16/33/24/9/12-12	1 (0.5)			14/16/28/22/10/11-16	1 (0.5)	
13/16/33/24/11/14-15	1 (0.5)	3		14/16/29/22/11/8-8	1 (0.5)	
13/16/33/25/11/11-15	1 (0.5)			14/16/29/24/10/11-13	1 (0.5)	
13/17/30/23/10/13-15	1 (0.5)			14/16/30/23/11/11-13	1 (0.5)	1
13/17/31/24/11/14-15	1 (0.5)	5	1	14/16/31/24/10/13-15	1 (0.5)	
13/17/31/25/11/9-11	1 (0.5)			14/16/31/25/11/11-14	1 (0.5)	4
13/17/32/22/10/16-18	1 (0.5)			15/15/29/21/10/13-14	1 (0.5)	1
14/13/30/23/10/16-17	1 (0.5)	1		15/15/29/21/10/13-16	1 (0.5)	
14/14/27/22/11/13-14	1 (0.5)			15/16/28/21/10/14-14	1 (0.5)	

*YHRD reference database was accessed online at <http://www.ystr.charite.de>.

†Data on German Turks data were taken from reference 15.

‡Blanks indicate haplotypes which were observed in our study but not in YHRD and German Turks.

Only 19 haplotypes were the same for both the Turks in the homeland and in Germany. It can, therefore, be expected that a larger number of haplotypes will be revealed in studies based on larger sample sizes.

Henke et al (15) showed a high degree of haplotype diversity in German Turks, with the highest frequency of 2.5%, and the most frequent haplotypes of 12% in Jat Sikhs and 4.8% in Germans. We also found the two most frequent haplotypes occurring three times with the frequency 1.5%, which strongly confirms the observations of Henke et al (15). Such a high degree of haplotype diversity in Turkish population makes Y-STRs a very useful tools in forensic cases.

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