

ALLELE AND HAPLOTYPE FREQUENCIES OF (6)Y-STR LOCI IN TURKEY

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Introduction

DNA polymorphism of the human Y-chromosome is becoming frequently used in forensic application. Although Y-STR data concerning mutational rates have been limited so far, recent studies indicate similar average mutational rates for autosomal STR loci commonly used in forensic sciences. Y-chromosomal STRs are characterized by male inheritance pattern and remain stable in a given paternal lineage over many generations. Thus, this system is very useful in forensic casework such as rape cases where a mixture of male and female DNA is found, and paternity deficiency cases when the putative father is not available. It is also used in kinship analysis and human identification.

In this study, we analyzed the frequency distributions 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 males, from different regions of Turkey. DNA extraction was performed by salting out procedure or chelex extraction.

PCR Amplification

DNA was amplified and typed using of the Y-Plex 6 kit, which contains 6 Y-STR markers (DYS393, DYS19, DYS389 II, DYS 390, DYS 391, DYS 385; Reliagene Technologies, Inc., New Orleans, LA, USA) according to the manufacturer's protocol. Amplification was carried out in a MJ Research PTC-200 Thermal Cycler (MJ Research, Inc., Watertown, Massachusetts, USA).

Electrophoresis

Capillary electrophoresis was carried out on an ABI 310 genetic analyzer (Applied Biosystems, Foster City, USA). Allele designations were determined according to the fragment size in base pairs, using the allele designation guide provided (Reliagene Technologies, Inc., New Orleans, USA).

Results

Total 200 unrelated Turkish individuals were analyzed. Allele frequencies and gene diversity values are shown in Tables 1 and 2. Haplotype frequencies observed in our study, Y-chromosomal short tandem repeat haplotype reference database (YHRD) and Henke's results for German Turks are listed in Table 3.

We observed total 184 haplotypes. Fourteen haplotypes occurred more than once, 12 of them were snared 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, had 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 in our population (58 %) when they were compared with YHRD database (Table 3). Haplotype, "13/14/29/23/11/11-14", observed once in our data was found 162 times in the YHRD database.

Discussion

Except DYS393 locus, the gene diversity values were similar to data in the previous publications studied with Swiss, German and Chinesees. Decreasing number of haplotype combinations from 184 to 122, when we exclude the DYS385 locus, confirms that this locus has been accepted as one of the most informative Y-STR loci having been published so far.

The genotype "11-14" at the locus DYS385 was the most frequent allele combination (0.125) observed in our population. This was also identified as the most common genotype at a collaborative study including the data of Portugal, Spain, Italy, Greece, Germany, Netherlands, England, Norway and it varied from 0.099 in the Greek to 0.409 in the Dutch.

Only 19 haplotypes were 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. For Y-chromosome specific markers, the haplotype diversity for the whole array of loci is more important than 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 males.

Henke et al. showed a very high degree of haplotype diversity with 2.5 % highest frequency in German Turks, whereas at the same study the most frequent haplotypes were found 12% in Jat Sikhs and 4.8% in Germans. We also found the two most frequent haplotypes occurred three times with the frequency 1.5%.

This result strongly confirm Henke's observation above. This very high degree of haplotype diversity in Turkish population makes Y-STRs more useful tools in Forensic cases.

We have begun to use this system routinely for deficiency paternity testing cases (e.g. where the father is not available for analysis or supporting the autosomal STR data), incest cases, all rapes, homosexual assaults, human identification, homicide investigations, and the Y-STR system has also been planned for mass disasters.

Table 1. Allele Frequencies of 6 Y-STRs (DYS393 / DYS19 / DYS389 I / DYS390 / DYS391) and gene diversity* in Turkish population

Allele	Frequency				
	DYS 393	DYS 19	DYS 389 I	DYS 390	DYS 391
9					0.050
10					0.060
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		0.090			
33		0.020			
Gene Diversity	0.63	0.71	0.76	0.70	0.48

Table 2. Allele frequencies and gene diversity of DYS385 locus in Turkish population

Allele	Frequency	Allele	Frequency
10-8	0.002	14-15	0.008
10-11	0.008	14-16	0.002
10-13	0.005	14-17	0.001
10-10	0.008	14-18	0.003
11-11	0.008	14-19	0.003
11-13	0.002	14-20	0.003
11-14	0.008	14-21	0.002
11-15	0.008	14-22	0.002
11-16	0.008	14-23	0.002
11-17	0.008	14-24	0.002
11-18	0.008	14-25	0.002
11-19	0.008	14-26	0.002
11-20	0.008	14-27	0.002
11-21	0.008	14-28	0.002
11-22	0.008	14-29	0.002
11-23	0.008	14-30	0.002
11-24	0.008	14-31	0.002
11-25	0.008	14-32	0.002
11-26	0.008	14-33	0.002
11-27	0.008	14-34	0.002
11-28	0.008	14-35	0.002
11-29	0.008	14-36	0.002
11-30	0.008	14-37	0.002
11-31	0.008	14-38	0.002
11-32	0.008	14-39	0.002
11-33	0.008	14-40	0.002
11-34	0.008	14-41	0.002
11-35	0.008	14-42	0.002
11-36	0.008	14-43	0.002
11-37	0.008	14-44	0.002
11-38	0.008	14-45	0.002
11-39	0.008	14-46	0.002
11-40	0.008	14-47	0.002
11-41	0.008	14-48	0.002
11-42	0.008	14-49	0.002
11-43	0.008	14-50	0.002
11-44	0.008	14-51	0.002
11-45	0.008	14-52	0.002
11-46	0.008	14-53	0.002
11-47	0.008	14-54	0.002
11-48	0.008	14-55	0.002
11-49	0.008	14-56	0.002
11-50	0.008	14-57	0.002
11-51	0.008	14-58	0.002
11-52	0.008	14-59	0.002
11-53	0.008	14-60	0.002
11-54	0.008	14-61	0.002
11-55	0.008	14-62	0.002
11-56	0.008	14-63	0.002
11-57	0.008	14-64	0.002
11-58	0.008	14-65	0.002
11-59	0.008	14-66	0.002
11-60	0.008	14-67	0.002
11-61	0.008	14-68	0.002
11-62	0.008	14-69	0.002
11-63	0.008	14-70	0.002
11-64	0.008	14-71	0.002
11-65	0.008	14-72	0.002
11-66	0.008	14-73	0.002
11-67	0.008	14-74	0.002
11-68	0.008	14-75	0.002
11-69	0.008	14-76	0.002
11-70	0.008	14-77	0.002
11-71	0.008	14-78	0.002
11-72	0.008	14-79	0.002
11-73	0.008	14-80	0.002
11-74	0.008	14-81	0.002
11-75	0.008	14-82	0.002
11-76	0.008	14-83	0.002
11-77	0.008	14-84	0.002
11-78	0.008	14-85	0.002
11-79	0.008	14-86	0.002
11-80	0.008	14-87	0.002
11-81	0.008	14-88	0.002
11-82	0.008	14-89	0.002
11-83	0.008	14-90	0.002
11-84	0.008	14-91	0.002
11-85	0.008	14-92	0.002
11-86	0.008	14-93	0.002
11-87	0.008	14-94	0.002
11-88	0.008	14-95	0.002
11-89	0.008	14-96	0.002
11-90	0.008	14-97	0.002
11-91	0.008	14-98	0.002
11-92	0.008	14-99	0.002
11-93	0.008	14-100	0.002

* Gene Diversity = 1/n

Table 3. Y-chromosomal short tandem repeat haplotype frequencies of the first 100 Y-STRs (DYS19, DYS390, DYS391, DYS385) in Turkish population and comparison with YHRD and Henke's results

Haplotype	Frequency	YHRD	Henke	DYS19		DYS390		DYS391		DYS385	
				Allele	Frequency	Allele	Frequency	Allele	Frequency	Allele	Frequency
11-14-15-16-17-18	0.015	1	1	11	0.015	14	0.015	15	16	17	18
11-14-15-16-17-19	0.015	1	1	11	0.015	14	0.015	15	19	17	18
11-14-15-16-17-20	0.015	1	1	11	0.015	14	0.015	15	20	17	18
11-14-15-16-17-21	0.015	1	1	11	0.015	14	0.015	15	21	17	18
11-14-15-16-17-22	0.015	1	1	11	0.015	14	0.015	15	22	17	18
11-14-15-16-17-23	0.015	1	1	11	0.015	14	0.015	15	23	17	18
11-14-15-16-17-24	0.015	1	1	11	0.015	14	0.015	15	24	17	18
11-14-15-16-17-25	0.015	1	1	11	0.015	14	0.015	15	25	17	18
11-14-15-16-17-26	0.015	1	1	11	0.015	14	0.015	15	26	17	18
11-14-15-16-17-27	0.015	1	1	11	0.015	14	0.015	15	27	17	18
11-14-15-16-17-28	0.015	1	1	11	0.015	14	0.015	15	28	17	18
11-14-15-16-17-29	0.015	1	1	11	0.015	14	0.015	15	29	17	18
11-14-15-16-17-30	0.015	1	1	11	0.015	14	0.015	15	30	17	18
11-14-15-16-17-31	0.015	1	1	11	0.015	14	0.015	15	31	17	18
11-14-15-16-17-32	0.015	1	1	11	0.015	14	0.015	15	32	17	18
11-14-15-16-17-33	0.015	1	1	11	0.015	14	0.015	15	33	17	18
11-14-15-16-17-34	0.015	1	1	11	0.015	14	0.015	15	34	17	18
11-14-15-16-17-35	0.015	1	1	11	0.015	14	0.015	15	35	17	18
11-14-15-16-17-36	0.015	1	1	11	0.015	14	0.015	15	36	17	18
11-14-15-16-17-37	0.015	1	1	11	0.015	14	0.015	15	37	17	18
11-14-15-16-17-38	0.015	1	1	11	0.015	14	0.015	15	38	17	18
11-14-15-16-17-39	0.015	1	1	11	0.015	14	0.015	15	39	17	18
11-14-15-16-17-40	0.015	1	1	11	0.015	14	0.015	15	40	17	18
11-14-15-16-17-41	0.015	1	1	11	0.015	14	0.015	15	41	17	18
11-14-15-16-17-42	0.015	1	1	11	0.015	14	0.015	15	42	17	18
11-14-15-16-17-43	0.015	1	1	11	0.015	14	0.015	15	43	17	18
11-14-15-16-17-44	0.015	1	1	11	0.015	14	0.015	15	44	17	18
11-14-15-16-17-45	0.015	1	1	11	0.015	14	0.015	15	45	17	18
11-14-15-16-17-46	0.015	1	1	11	0.015	14	0.015	15	46	17	18
11-14-15-16-17-47	0.015	1	1	11	0.015	14	0.015	15	47	17	18
11-14-15-16-17-48	0.015	1	1	11	0.015	14	0.015	15	48	17	18
11-14-15-16-17-49	0.015	1	1	11	0.015	14	0.015				