DNA-TP-06

Allele Frequencies and Haplotypes of 6 Y-STR Loci in Turkish Population

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DNA polymorphism on the human Y-chromosome are becoming of increasing interest in forensic application. During the last few-years, new Y-chromosome STR markers have been described and a huge amount of population and sequencing data published. Some international groups have carried out collaborative studies to characterize Y-chromosome STRs. Large number of databases (YHRD - database of European Y STRs haplotype I have been collected by participating laboratories and announced via internet site to facilitate the forensic exploitation of human Y-chromosomal STR markers and to support the presentation of Y STR evidence in court.

Y-chromosomal STR's are characterized by a male inheritance pattern and remain stable in a given paternal lineage over many generations. Thus, these systems are *very* useful tools for evolutionary studies and human identification, especially in rape cases where a mixture of male and female DNA is present and in paternity deficiency cases when the putative father is not available. For investigation of sexual assault cases Y STRs are especially suitable, since they provide a male specific DNA profile which avoids problems of mixed stain interpretation. Therefore, this technique has been accepted by the courts in various countries. Recently commercial kits have been available, extending the routine forensic casework.

Because of importance of detailed databases of population, in this study we present frequency distributions of six Y-chromosomal specific STR polymorphisms and frequencies of hapiotypes in Turkish population.

Blood samples were taken from 200 unrelated Turkish males from different regions of Turkev DNA extraction was performed by salting out procedure or chelex extraction.

PCR amplification were typed by use of the Y-Plex 6 kit which contains 6 Y-STR markers (DYS393, DYS19, DYS389II, DYS 390, DYS 391, DYS 385)(Reliagene Technologies, Inc., New Orleans. USA) according to the manufacturer's protocol. The PCR was carried out in a MJ Research PTC-200 Thermal Cycler.

Capillary electrophoresis was carried out on an ABI 310genetic analyser (Applied Biosystems, Foster City, USA) as described in the "ABI Prism Genetic Analyser. User's Manual" (1998) provided by Applied Biosystems. Allele designations were determined according to fragment size in base pairs, using the allele designation guide provided (Reliagene Technologies, Inc., New Orleans, USA).

A total of 200 unrelated Turkish individuals were analyzed. Except DYS393 locus, the gene diversity values were similar to data in the previous publications studied with Swiss, German and Chinees.

Bimodal distribution at the loci DYS393, and DYS 19 were present. At the DYS391 locus, allele 10 was the most frequent allele among all the loci. Our data for DYS 385 locus showed flat distribution such as described at some population like Chinese, Thai and German.

We observed total 184 hapiotypes with 6 Y-STR. including DYS385, Fourteen hapiotypes occurred more than once, whereas the remaining 170 combinations were observed once. The most common hapiotypes "13/16/31/24/11/14-15" and "14/15/29/22/10/13-14" repeated 3 times in this study had a frequency of 1.5%. Whereas when we exclude DYS385 locus, we found only 122 hapiotypes. Out of these l22 hapiotypes, 34 occurred more than once, and only 88 occurred once. This results confirm that this locus has been accepted one of the most informative single Y-linked STR published so far.

Haplotype frequencies observed in our study, Y-chromosomal short tandem repeat haplotype reference database (YHRD) and Henke's results *for* German Turks are were also compared in this paper. A total of 107 hapiotypes were unique in our population (58 %) when they were compared with YHRD database. Haplotype, "13/14/29/23/11/11-14". observed once in our data was found 162 times in the YHRD database. Only 19 hapiotypes were same for both the Turks in the homeland and in Germany. It can therefore be expected that a larger number of hapiotypes will be revealed in studies based on larger sample sizes.

Henke et al. showed a very high degree of haplotype diversity with 2.5 *9c* highest frequency in German Turks, whereas at the same study the most frequent hapiotypes were found 12% in Jat Sikhs and 4.8% in Germans [16]. As given before, we also found the two most frequent hapiotypes occurred three times with the frequency 1.5%. This result strongly confirms Henke's observation above. This very high degree of haplotype diversity in Turkish population makes Y-STRs more useful tools in Forensic cases.

The dominant allele combination at the DYS385 locus "11-14" determined in this study had a frequency of 0.125, followed by 13-14 with 0.07. 13-16 and 14-15 with 0.05 respectively. The genotype 11-14 was also identified as the most common genotype at a collaborative study which consists of the data of Portugal. Spain, Italy, Greece. Germany. Netherlands. England. Norway, Germany varied 0.099 in the Greek and 0.409 in the Dutch population [5] Whereas in Chinese with the 13-13(9%), Japanese with the 13-17 (14%) and Thai with the 14-18 genotypes (7%) were the most common genotypes.

In our study, the total number of genotypes observed at DYS385 locus were 47. At the above mentioned collaborative study, the total number of genotypes observed varies 23 in the Dutch and 42 in the Greek population. Our result is appropriate with the knowledge that Mediterranean countries exhibit more genotype heterogeneity compared to the Central and Northern European countries [5]. In another study in the German and three Asian populations has been found 69 different genotypes at DYS385 locus. Of these. 36 were observed in Germans. 36 in Chinese, 33 in Japanese, and 44 in Thai.

We began 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, corps identification, homicide investigations and

plan to use mass disaster. Indeed, inclusion of Y-STR typing in all forensic cases involving males should become routine.

We believe that the database size must be large enough. Future studies are needed to increase the database size that would allow accurate frequency estimation for rare haplorypes.

Keywords: Y-STR. Turkish Population. Haplotypes.

DNA-TP-07

Y -**STR** Typing **in** Forensic Casework

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DXA evidence in forensic analysis and paternity testing is based the interpretation of similarities or differences at genetic marker loci. The human Y chromosome represents only 2 % of the human genome ar.d is approximately 60 Mb in length. The role of the human Y chromosome in male sex determination requires being haploid. and this haploidy ha- the consequence that most of the chromosome escapes from recombination. Due to the lack of recombination, except for mutation events, all male relatives of the paternal lineage will share the same allele combination.

During the last decade. STR loci or microsatellites evolved to be the markers of the choice for forensic cases. The first *Y* -STR declined was Y27H39 known a: present as OYS19. Over 25 Y- STRs have been described and a basic set of nine or an extended set of eleven Y -STR were thoroughly investigated. Its applicability was demonstrated in a wide range of forensic cases such as: deficiency paternity cases of male offspring, victim identification in mass disasters. Application of Y -STRs in forensic casework can especially useful whenever mixtures of small amount of male DXA and large amount of female DNA have to be analyzed.

So far. the usefulness of Y -STRs was mostly presented in sexual assault case-, whereas two case studies presented here demonstrate that Y -STR- could be used effectively in paternity cases and other forensic investigations.

Case examples: DXA of both cases were extracted by salting out or chelex procedure. DXA was amplified using of the Y -Plex 6 and ProfilerPius kits in a MJ Research PTC-2OO Thermal Cycler and typed on an ABI?100 genetic analyzer.

Case 1: In. paternity testing, differences at genetic marker loci between the putative father and the offspring are attributed to non-biologica: paternity, and thus, lead to the exclusion of biological paternity. However, spontaneous mutations in the germ line of the putative fatner at any genetic marker locus used in the analysis can lead to an erroneous exclusion since such mutations result in difference- between the raren: and the offspring. At this case, nine STR loci including. 03S135S. HumvWA. HumFGA. 08S1179. 021S11. 018S51. 05SS1S. 013S31" and 0"SS20. were analyzed. Among these loci, at the two loci 021S11: 018S51. differences has been found between the putative father and the child. These differences were one repeat decrease. The common practice in paternity testing is that a difference at one or two out of 6-15 STR loci commonly analyzed is attributed to mutation rather than non-paternity, whereas differences a: more than two loci are interpreted as non-biological paternity. Therefore, we decided to perform additional loci and Y -STRs were carried out. The results of Y -STR were very informative with 5 exclusion at 6 Y -STR loci.

Case 2: A piece of bloodstained wood was found at the scene of wood robbery. This bloodstained material and 5 samples taken from the suspects were sent for DXA identification. Autosomal testing showed match between the bloodstained wood and the one of the suspects, but this matched biood was not labeled sufficiently in which there was the date of the blood taken and some other details but not the name of the person. After reporting this result, the prosecutor sent a blood taken from this suspect. However the second coming blood did not match with the first coming blood sample with autosomal testing, but match with Y -STRs. Therefore we reported this situation as mentioning that these two bloods were not belonging to the same persons but to the persons who are in the same patrilineage. After all, prosecutor requested the repeat of the. analysis as saying that the second blood sample was not taken under his control and sent the suspected person to our lab. At the end. Y -STR testing and criminal investigation revealed that the second blood received was taken from the brother of the suspect.

Keywords: Y -STR. Paternity. Human Identification

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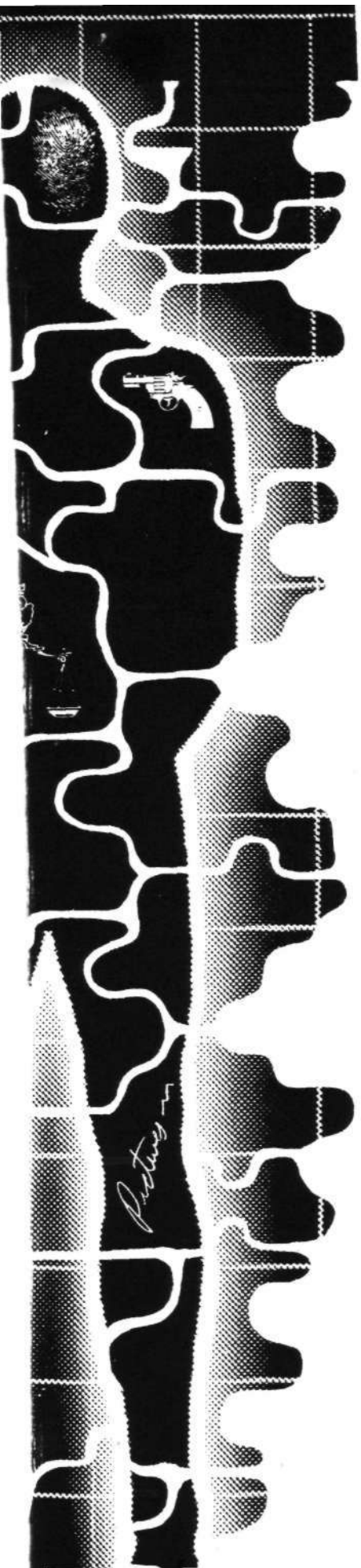

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