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Review Article

Genetic Polymorphisms at 17 Y-STR loci in Uzbek Population

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Abstract

Haplotypes and allele frequencies for the 17 Y-chromosomal short tandem repeat (Y-STR) loci, DYS456, DYS389I, DYS390, DYS389II, DYS458, DYS19, DYS385a/b, DYS393, DYS391, DYS439, DYS635, DYS392, Y GATA H4, DYS437, DYS438 and DYS448 were determined in a sample of 1000 unrelated Uzbekistan males living in the regions of Tashkent (100), Fergana (75), Andijan (85), Namangan (55), Sirdarya (55), Djizax (80),Samarqand (80), Kashqadarya (75), Surxandarya (50), Buxara (100), Navoiy (50), Xorezm (97) and from Republic Karakalpakistan (98) using the Y-filer PCR Amplification Kit (Thermo Fisher Scientific).This population was demonstrated 1000 haplotypes, of which 899 were unique. The gene diversity was 0.9988 (standard error:0.005).The haplotype diversity calculated from the 17 Y-STR loci was 0.9967 and the discrimination capacity was 0.8990. The DYS385 locus showed the highest gene diversity value (0.8936), while the DYS391 locus showed the lowest gene diversity value (0.4934).

Keywords: DNA analysis; Mutation; Y chromosome; Population data

Introduction

There have been few reports regarding genetic polymorphisms at the Y-STR loci in Uzbekistan population [3]. This study aimed to investigate the haplotypes and allele frequencies for the 17 Y-STR loci in Uzbekistan population and establish forensic DNA database.

Materials and Methods

Objects of the Research: The subjects of the study were blood samples and dried saliva on sterile gauze tampons, selected from 1000 individuals.

DNA Extraction: Genomic DNA was extracted from peripheral blood and dried saliva samples using the phenol-chloroform-isoamyl alcohol method.

DNA Quantification: After isolation, the quantity of genomic DNA of each sample was determined by quantitative real-time polymerase chain reaction (PCR) using the Quantifiler™ Human Male DNA Quantification kit (Thermo Fisher Scientific), which includes internal positive control to test for the presence of PCR inhibitors in the DNA extracts. Quantitative real-time PCR was performed on 7500 Real-Time PCR System (Applied Biosystems).

PCR Amplification and Detection: To ensure successful amplification, 0.5ng to 1ng of DNA was used for each multiplex amplification reaction. All thermal cycling was conducted on Applied Biosystems® Gene Amp® PCR System 9700 thermal cyclers. PCR amplification using Y-filer PCR Amplification Kit (Thermo Fisher Scientific) was performed as recommended by the manufacturer, although half of the recommended reaction volume (12.5µl) was used. Separation and detection of the 17 Y-STR loci were performed using the 3130xl Genetic Analyzer (Applied Biosystems) 16-capillary array system and filter set G5. Each sample was prepared by adding 1mL PCR product to 14mL of Hi-Di™ formamide and 0.4mL GeneScanTM-500 LIZ™ internal size standard (Thermo Fisher Scientific). The sample run data were analyzed, together with an allelic ladder and positive and negative controls, using GeneMapper ID-X v3.2 (Applied Biosystems) software.

Statistical Analysis

Comparison information of the sample data was generated using an in-house software program involving DNA-expert macros designed to check for allele sharing across all loci. For all analyses the DYS385 locus was treated as a single haplotype and not two separate alleles. The gene diversity (D) was calculated as

$$D = \frac{n}{n-1}\left(1 - \sum p_i^2\right),$$

where p_i is the frequency of the ith haplotype [2].

The discriminatory capacity was determined by dividing the number of different haplotypes by the number of samples in that

population. The discrimination capacity (DC) was determined by the formula n/N where n=the number of observed haplotypes divided by the number of samples [1].

Results and Discussion

This population was demonstrated 1000 haplotypes, of which 899 (Tashkent-100, Fergana-73, Andijan-84, Namangan-36, Sirdarya-54, Djizax-77, Samarqand-77, Kashqadarya-71, Surxandarya-46, Buxara-97, Navoiy-48, Xorezm-90 and from Republic Karakalpakistan-93) were unique. The gene diversity was 0.9988 (standard error:0.005). The haplotype diversity calculated from the 17 Y-STR loci was 0.9967 and the discrimination capacity was 0.8990. The DYS385 locus showed the highest gene diversity value (0.8936), while the DYS391 locus showed the lowest gene diversity value (0.4934). This database of 17 Y-STR loci for the Uzbekistan population would be useful in forensic examinations and human genetic studies.

References

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