

ESTIMATION OF THE Y-CHROMOSOMAL SHORT TANDEM REPEAT (Y-STR) MUTATION RATES IN UZBEKISTAN

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When determining the degree of solution on the paternal line, if discrepancies between the child's father and other paternal relatives are not taken into account, population-specific mutation rates should be used to determine if this is a mutation or a true exception. Therefore in this study, we aim to determine the mutation rates of 17 Y-STR(Y-filer PCR Amplification Kit) loci in Uzbekistan.

Within the framework of the research, the speed of mutations in 300 pairs of fathers and sons throughout Uzbekistan was studied. Among 5100 translations of alleles, mutations were detected in seven cases, giving an average mutation rate of $7/5100 = 1.373 \cdot 10^{-3}$ at all loci by generation.

Also, the rate of mutations in two large related families from districts Zangiota and Kibray was studied. According to the data obtained the first family consisted of 15 generations, and the second family consisted of 14 generations. As a result of our analysis, one mutation was detected in each family of the locus DYS385a.

As a result of our study of 150 people living in the city of Angren, it was revealed that they had a genetic similarity of the Y chromosome. Among this random sample, 80 people who had close haplotypes, turned out to be relatives. Among them, in the study of loci with a high mutation frequency, 19 individual mutations were found.

The total number of mutations per sample (19 mutations) is divided by the number of haplotypes (11 haplotypes) and the number of markers in the haplotype (17 markers)

$$\frac{19}{11 \cdot 17} = 0.101604 \text{ (Mutations per haplotype)}$$

In this sample study, the average observed number of mutations per marker is 0.101604.

Now we can approximately estimate the age of the common ancestor. The rate of mutations for our 17-marker haplotype: 0.031 mutations per haplotype, or 0.0018 mutations per marker. The duration of one generation is assumed to be 25 years.

The age of a common ancestor is obtained by dividing the average observed number of mutations per marker by the rate of mutations (also on the haplotype): $\frac{0.101604}{0.0018} = 55.71$ generations.

Multiplying 55.71 generations of 25 years, we obtain 1392 years. 1390 years is an approximate, rough estimate of the age of a common ancestor.