

## Analysis of Polymorphism in Alu-Insertional Loci in the Population of Tatar in Russia

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**Abstract:** The study of variability and diversity of the human genome in the population had a rapid development. The purpose of this paper is to compare the data on Alu- insertion polymorphism in the population of the Kazan and baptized Tatars, Russian Federation, as well as in the neighboring populations. The material for the genetic analysis was DNA collected from 192 unrelated individuals, representatives of Tatar nationality (96- Kazan Tatars and 96 - baptized Tatars), Russian Federation. Data on ethnicity was found out by the survey, blood sampling was carried out on a voluntary basis with signed informed consent. Alu- insertion polymorphism in the population of the Kazan and baptized Tatars, Russian Federation, as well as in the neighboring populations was analysed.

**Key words:** Alu- insertion • Polymorphism • Tatars

### INTRODUCTION

Over the past years, the study of variability and diversity of the human genome in the population had a rapid development. However, the question about the demographic history of certain ethnic groups, such as the Tatars, remains open.

To address these issues the proven system of genetic markers, such as autosomal Alu- insertion, mitochondrial DNA, Y- chromosome, is widely used. In particular, some properties of the Alu- repeats, such as high stability, low insertion *de novo*, no mechanism of removal from a specific locus, methodical simplicity of genotyping are making them very convenient to use. If by studying the polymorphism of mitochondrial DNA and Y chromosomes the characteristic of male and female gene pool in the population could be found, then the study of polymorphism of autosomal Alu- insertion allows you to define some features of the human genome diversity in the population as a whole [1]. Unlike other biallelic systems, Alu- insertions advantage is that initial and final state, before and after insertion into the locus Alu- member, are always known [2]. In human population genetics the Alu- insertions, which appeared relatively recently, are used. They

coincide with the resettlement of people across continents. Some of the loci containing Alu- elements have been very informative markers of populations differentiation in Europe and Asia - Ya5NBC148, PV92, TPA25 and Ya5NBC27 [3].

The purpose of this paper is to compare the data on Alu- insertion polymorphism in the population of the Kazan and baptized Tatars, Russian Federation, as well as in the neighboring populations.

### MATERIALS AND METHODS

The material for the genetic analysis was DNA collected from 192 unrelated individuals, representatives of Tatar nationality (96- Kazan Tatars and 96 - baptized Tatars), Russian Federation. Data on ethnicity was found out by the survey, blood sampling was carried out on a voluntary basis with signed informed consent.

DNA was isolated from peripheral blood by phenol-chloroform extraction. Genotyping of 10 Alu- insertion loci (PV92, B65, ACE, APOA1, Ya5NBC123, Ya5NBC182, Ya5NBC148, Ya5NBC102, Ya5NBC480, Ya5NBC485) was performed by PCR amplification of DNA. Statistical analysis was carried out using Genepop and popSTR software.

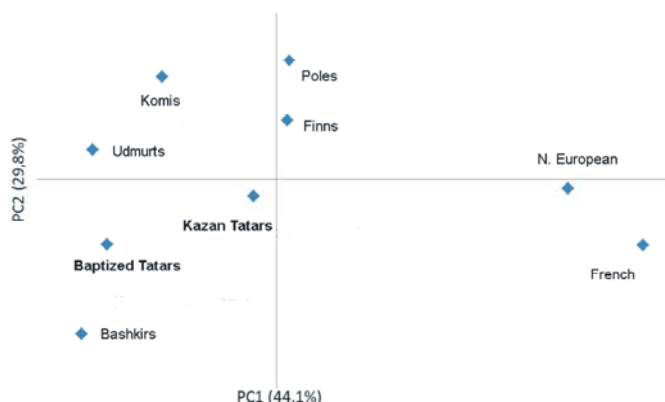


Fig. 1: Tatar population status compared to some other populations of VUR in the space of the first two principal components according to the frequencies of 10 Alu-insertions (PV92, B65, ACE, APOA1, Ya5NBC123, Ya5NBC182, Ya5NBC148, Ya5NBC102, Ya5NBC480, Ya5NBC485)

## RESULTS AND DISCUSSION

Different Alu- insertions in the population of the Kazan Tatars are found with a frequency of 0.301 to 0.938, while among the baptized Tatars from 0.335 to 0.926. For these populations the occurrence of certain insertions characteristic more for the Volga-Ural region (VUR) has shown. The highest frequency of insertions detected by APOA1 locus (0.938), is consistent with the literature on VUR. Among the populations of the Volga-Ural region the highest frequency of APOA1 - insertion of the Tatars (0.914) and the Komi - Perm (0.914) has been previously shown, while the lowest was recorded in Bashkir (0.804). Among the Central Asian ethnic groups, the highest rate is among the Uzbeks (0.903) and the Kazakhs (0.801). The lowest rate was observed in the insertion of the Uighurs (0.548) [4]. In general, the frequency of this insertion, described in the literature, in Asian populations is lower (0.856) than in European populations (0.965) [6, 8]. This indirectly indicates the presence of the European component in the population of Tatars.

The low frequency in the population is observed for the locus ACE (0.493), corresponding to the previously obtained data for the Volga-Ural region [4] and Central Asia [3, 4]. In the study of the Kazan Tatars population all 10 loci were polymorphic. The average value of expected heterozygosity of 10 Alu- repeats is 0.3377.

It was previously shown that Tatar demonstrated some genetic structure which is a similar to that in other populations Turkic VUR. This could indicate to long-term contacts and / or similar demographic processes that took place in these populations [3]. Figure 1 shows that the first two principal components explain 73.9% of the variability of allelic frequencies. In this case, the first principal component separates the population of the

Volga-Ural region, including the baptized Tatars and the populations of Western Europe. At the same time, the second principal component shows the gradient between the Bashkirs and some of the Finno-Ugric populations and the Poles. Given the lack of discriminatory effect on 10 Alu- insertion loci, the interpretation of these data is difficult.

In the future, more comprehensive genetic study of sub-populations of Tatars with additional markers systems with greater phylogenetic resolution, will reveal not only the genetic relationship with other populations of Tatars, but also, perhaps, will find specific features within the population itself.

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