

Analysis of Eight Polymorphic Alu Elements in the Teleuts Population

M. G. Swarovskaya^{a, b, c}, A. V. Marusin^b, T. I. Tacheeva^b,
I. Yu. Khitrinskaya^b, N. I. Gafarov^d, and V. A. Stepanov^{a, b}

^aNational Research Tomsk State University, Tomsk, 634050 Russia

^bInstitute of Medical Genetics, Tomsk Research Center, Siberian Branch of the Russian Academy of Medical Sciences, Tomsk, 634050 Russia

e-mail: maria.swarovskaja@medgenetics.ru

^cSiberian State Medical University, Tomsk, 634050 Russia

^dNovokuznetsk State Institute of Postgraduate Medicine, Novokuznetsk, 654005 Russia

Received September 30, 2014; in final form, November 14, 2014

Abstract—Allele frequencies and genetic diversity in the population of Teleuts were assessed by the Alu repeat polymorphism at eight autosomal loci (*ACE*, *APOA1*, *PLAT*, *F13*, *PV92*, *A25*, *CD4*, *D1*). For comparison, the study included previously obtained data on the Alu polymorphism in 19 indigenous populations of Siberia. On the dendrogram of genetic distances, the Teleut population is located in the cluster of Siberian ethnic groups, which are similar in origin, geography, and cultural traditions.

DOI: 10.1134/S1022795415080104

The Teleuts, a people numbering about 3000 individuals, live in the south of western Siberia, mainly in rural areas and in the towns of Kemerovo oblast, Altai Krai, and the Altai Republic. Teleuts speak one of the southern dialects of the Altai language. The written language is based on the Russian alphabet. The ancestors of Teleuts are the tribes of the epoch of ancient Turks. In the Russian chronicles, the Teleuts (white Kalmyks, as opposed to black Kalmyks, i.e., Mongolians) were first mentioned at the beginning of the 17th century. Teleuts actively participated in the ethnogenesis of neighboring peoples (Khakassians, Altaians, various groups of the Ob and Tomsk Tatars, Russians), with which they assimilated over time [1, 2].

In this study, the DNA samples from 155 representatives of the Teleut population living in the Belovo region of Kemerovo oblast were examined with the use of a panel of autosomal polymorphic insertions of Alu repeats (*ACE*, *APOA1*, *PLAT*, *F13*, *PV92*, *A25*, *CD4*, *D1*). Alu repeats are a well-known family of short repetitive elements. The human genome contains about one million copies of Alu repeats. The age of these sequences constitutes more than 100 million years; they can transpose to new loci of the genome and are found in the genomes of many species [3]. Alu repeats are very convenient for population genetic studies and can accurately identify the ancestral (no insertion) and derivative (the presence of an Alu repeat) allelic state.

The study includes previously obtained data on the Alu polymorphism in 19 indigenous populations of Siberia and adjacent territories (Tuvinians, Buryats, Evenks, Altaians, Kets, Yakuts, Kyrgyz, Uzbeks, Dunggans, Kazakhs, Tajiks, Russians, Chukchis, Eskimo,

Nivkhs, Ukrainians, Khanty, Mansi, and Koryaks) [4]. DNA samples were isolated from peripheral blood leukocytes by the standard method [5]. Genotyping was performed by PCR and subsequent electrophoresis in 2% agarose gel. The genetic relationships among the ethnic groups were analyzed by factor analysis. The genetic distances between the populations and the coefficient of genetic differentiation, G_{ST} , was calculated by the Nei method [6]. A phylogenetic tree of the populations was built in the PHYLIP software package (version 3.5) [7] with 1000 bootstrap replicates of the original dataset.

The table shows the allele frequencies and expected heterozygosity at eight loci for the Teleut population and the level of genetic differentiation for 20 populations. The genotype distribution at seven loci was consistent with the Hardy–Weinberg distribution (except for the *F13* locus; $\chi^2 = 11.2618$; $P < 0.01$).

The high frequency of the insertion allele (75–82%) was observed at the *APOA1*, *F13*, and *CD4* loci. A very low insertion frequency (9%) was detected at the *A25* locus. Comparing the allele frequencies in the Teleut population with the same loci in other populations, the most interesting is the comparison of three markers, *PV92*, *F13B*, and *APOA1*, because these three loci differentiate Caucasoid and Mongoloid populations well [8, 9]. The frequencies of the insertion allele in the Teleut population at the *PV92* and *F13B* loci (0.54 and 0.75, respectively) unite this population with Mongoloids. With respect to the Alu-insertion occurrence at the *APOA1* locus (0.82), which in Mongoloids has a tendency toward a lower frequency, Teleuts occupy an intermediate position between Mongoloid and Caucasoid populations.

Frequency of the insertion allele (I) at the examined Alu loci, the heterozygosity index, and the level of genetic differentiation in 20 populations of Northern Eurasia

Index	<i>ACE</i>	<i>APOA1</i>	<i>PLAT</i>	<i>F13</i>	<i>PV92</i>	<i>A25</i>	<i>CD4</i>	<i>D1</i>
Frequency of allele I in the population of Teleuts	0.516	0.823	0.542	0.745	0.539	0.097	0.786	0.418
H_e in the population of Teleuts	0.499	0.292	0.497	0.380	0.497	0.175	0.337	0.487
G_{ST} value (for 20 populations), %	3.6	4.2	1.5	11.9	13.2	0.4	10.3	6.3

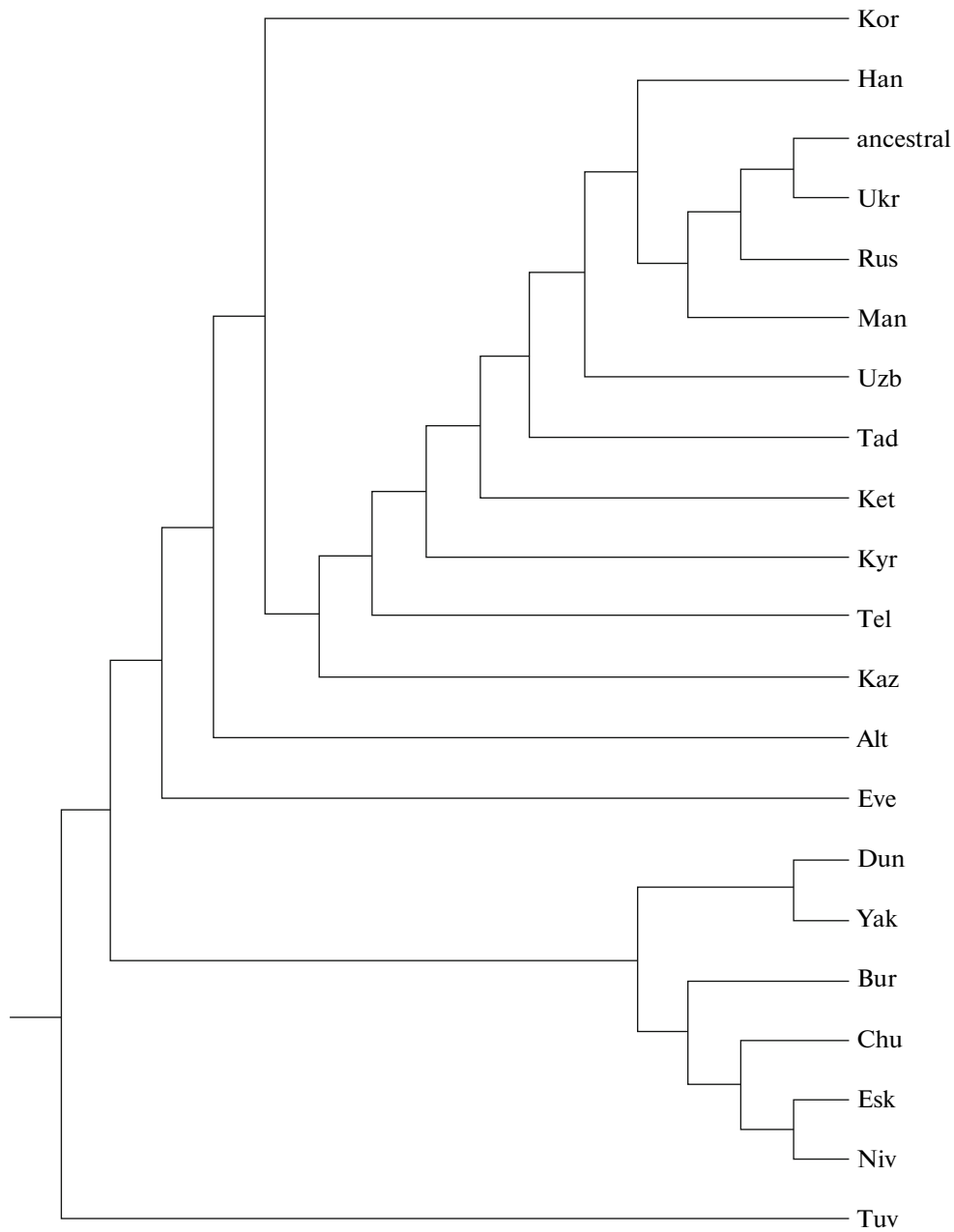
The Teleut population is characterized by a rather high level of genetic diversity. Specifically, the average expected heterozygosity of the population over all eight loci constituted 0.39, which was close to the average value obtained for five Alu insertions in the previously studied populations of Siberia and Central Asia [10] (from 0.343 in the population of Tatars to 0.450 in the population of Northern Altaians). An especially high level of diversity was observed at the *ACE* ($H_e = 0.499$), *PLAT* ($H_e = 0.497$), *PV92* ($H_e = 0.497$), and *D1* ($H_e = 0.487$) loci.

In this study, the genetic subdivision of twenty populations (including the Teleuts) residing on the territory of northern Eurasia was evaluated at individual loci and at a combination of eight Alu insertions. The values of the coefficient of genetic differentiation, which estimates the proportion of genetic diversity within the population and is determined by interpopulation differences (G_{ST}), are demonstrated in the table for each locus in the set of populations. The gene pool of the population of Northern Eurasia, which represents various geographic regions and linguistic groups, was found to be highly differentiated. The data for individual loci show that the largest contribution to interpopulation diversity is made by differences in the Alu insertion frequencies at the *PV92*, *F13B*, and *CD4* loci. The lowest degree of interpopulation diversity is observed for the Alu insertions at the *PLAT* and *A25* loci.

The dendrogram of the genetic relationship of the 20 populations over eight examined markers clearly demonstrates the position of Teleuts on the phylogenetic tree among the indigenous ethnic groups of Siberia (figure). On the phylogenetic tree, Teleuts are located in one large cluster in the vicinity of Kazakhs, Altaians, Kirghiz, Evenks, and Kets. Analysis of the Y-chromosomal markers produced conflicting data on the relatedness of Altaians and Teleuts [11]. On the other hand, the anthropological data provided evidence of their close relationship [12]. The low proportion of interpopulation differences identified by the Alu repeats is typical of the indigenous ethnic groups of southern Siberia, which points to the homogeneity

of the gene pool of this region [13]. Our data on the Alu insertions also demonstrate the closeness of these groups. Moreover, analysis of the Y-chromosomal haplogroups in Teleuts revealed the components of their gene pool associated with ancient Turkic and Samoyedic ethnic components. The genetic relatedness of Teleuts to other ethnic groups of the Altai and Sayan regions can be also traced by cultural, ritual, and ceremonial folkloric ties with the Altaian ethnic groups, which are located geographically close to the target group. For example, to record cattle, Teleut herders used special wooden counting sticks, *cherbik*, as did the Kumadins and Khakassians, who live on the adjacent territories [12]. The representatives of one *seok* (genealogical tribe) often longed to settle near each other. In the 19th–early 20th century, typical types of dwellings included permanent surface frame constructions of cone type, which were made of poles and bark or birch bark, *Alan-chyk*. Similar constructions were used by the closest neighbors of Teleuts, Khakassians and Tuvinians, as well as by small Altaian ethnic groups, Altai-Kizhi and Telengits. One more illustration of the organization of Teleut constructions (encasing of the exterior walls with turf, which were additionally covered by sheathed wattle from inside for the purpose of durability; an entrance oriented to the east, an adobe oven located to the right of the entrance) is very similar to the description of the half-dugouts typical of the Altaian ethnic groups, Chelkans and Kumadins, as well as of Shorians and Khakassians [12]. The oldest genre of oral traditions, funeral laments (*sygyt*), goes back to the ancient Turkic period. The traditional religion of the Teleuts is shamanism, which is similar to the shamanism of Altaian ethnic groups [14].

Thus, we investigated the molecular genetic structure of Teleuts by eight polymorphic Alu insertions. The allele frequencies and the level of genetic diversity in the Teleut population were assessed, and the place of the examined population among the ethnic groups of northern Eurasia was determined. In addition to ethnogenetic relatedness, an important component of



Dendrogram of genetic relationships between 20 populations of Northern Eurasia at eight examined Alu markers. Tuv, Tuvinians; Niv, Nivkhs; Esk, Eskimo; Chu, Chukchis; Bur, Buryats; Yak, Yakuts; Dun, Dungans; Eve, Evenks; Alt, Altaians; Kaz, Kazakhs; Tel, Teleuts; Kyr, Kyrgyz; Ket, Kets; Tad, Tajiks; Uzb, Uzbeks; Man, Mansi; Rus, Russians; Ukr, Ukrainians; ancestral, ancestral population; Han, Khanty; Kor, Koryaks.

the Teleuts' closeness to the Altaian ethnic groups on the phylogenetic tree is probably the deep interaction of the cultural and linguistic traditions of the studied ethnic groups.

ACKNOWLEDGMENTS

This work was supported by the Program of Tomsk State University: Mendeleev Science Foundation (2014–2015).

REFERENCES

1. *Korennyye malochislennyye narody Severa i Sibiri: rukovodstvo dlya issledovatelei* (The Small Indigenous Nations of Northern Russia and Siberia: A Guide for Researchers), Funk, D.A. and Sillanpää, L., Eds., Vaasa, 1999.
2. *Tyurkskie narody Sibiri* (Turkic Peoples of Siberia), Funk, D.A. and Tomilov, N.A., Eds., Moscow: Nauka, 2006.

3. Khitrinskaya, I.Yu., Stepanov, V.A., and Puzyrev, V.P., Alu repeats in the human genome, *Mol. Biol.* (Moscow), 2003, vol. 37, no. 3, pp. 325–333.
4. Stepanov, V.A., Ethnogenomics of population of Siberia and Central Asia, *Med. Genet.*, 2002, vol. 1, no. 3, pp. 113–123.
5. Johns, M. and Paulus-Thomas, J., Purification of human genomic DNA from whole blood using sodium perchlorate in place of phenol, *Anal. Biochem.*, 1989, vol. 180, no. 2, pp. 276–278.
6. Nei, M., *Molecular Evolutionary Genetics*, New York: Columbia Univ. Press, 1987.
7. Felsenstein, J., *PHYLIP (Phylogeny Inference Package), Version 3.5*, Seattle: Dep. Genet., Univ. Washington, 1993.
8. Batzer, M.A. and Deininger, P.L., Alu repeats and human genomic diversity, *Nat. Rev. Genet.*, 2002, no. 3, pp. 370–379.
9. Stoneking, M., Fontius, J., Clifford, S., et al., Alu-insertion polymorphisms and human evolution: evidence for a larger population size in Africa, *Genome Res.*, 1997, vol. 7, pp. 1061–1071.
10. Stepanov, V.A., *Etnogenomika naseleniya Severnoi Evrazii* (Population Ethnogenomics of Northern Eurasia), Tomsk: Pechatnaya manufaktura, 2002.
11. Kharkov, V.N., Medvedeva, O.F., Luzina, F.A., et al., Comparative characteristics of the gene pool of Teleuts inferred from Y-chromosomal marker data, *Russ. J. Genet.*, 2009, vol. 45, no. 8, pp. 994–1003.
12. Potapov, L.P., *Ocherki po istorii altaitsev* (Essays on History of Altaians), Moscow: Akad. Nauk SSSR, 1953, pp. 133–164.
13. Khitrinskaya, I.Yu., Kharkov, V.N., Voevoda, M.I., and Stepanov, V.A., Genetic diversity and relationships of populations of northern Eurasia by polymorphic Alu insertions, *Mol. Biol.* (Moscow), 2014, vol. 48, no. 1, pp. 58–69.
14. Gekman, L.P., *Mifologiya i fol'klor Altaya* (Mythology and Folklore of Altai), Barnaul: AGIHK, 2000.

Translated by N. Maleeva