
HUMAN GENETICS

Gene Pool Differences between Northern and Southern Altaians Inferred from the Data on Y-Chromosomal Haplogroups

V. N. Kharkov^a, V. A. Stepanov^a, O. F. Medvedeva^a, M. G. Spiridonova^a,
M. I. Voevoda^{b, c}, V. N. Tadinova^d, and V. P. Puzyrev^a

^a Institute of Medical Genetics, Tomsk Scientific Center, Russian Academy of Medical Sciences, Tomsk, 634050 Russia;
fax: (3822)51-37-44; e-mail: vadim.stepanov@medgenetics.ru

^b Institute of Internal Medicine, Russian Academy of Medical Sciences, Novosibirsk, 630089 Russia; fax: (3832)64-25-16

^c Institute of Cytology and Genetics, Russian Academy of Sciences, Novosibirsk, 630090 Russia; fax: (383)333-12-78

^d Republican Children Hospital, Gorno-Altai, 649002 Altai Republic, Russia; fax: (38822)2-61-86

Received March 6, 2006

Abstract—Y-chromosomal haplogroups composition and frequencies were analyzed in Northern and Southern Altaians. In the gene pool of Altaians a total of 18 Y-chromosomal haplogroups were identified, including C3xM77, C3c, DxM15, E, F*, J2, I1a, I1b, K*, N*, N2, N3a, O3, P*, Q*, R1*, R1a1, and R1b3. The structuring nature of the Altaic gene pool is determined by the presence of the Caucasoid and Mongoloid components, along with the ancient genetic substratum, marked by the corresponding Western and Eastern Eurasian haplogroups. Haplogroup R1a1 prevailed in both ethnic groups, accounting for about 53 and 38% of paternal lineages in Southern and Northern Altaians, respectively. This haplogroup is thought to be associated with the eastward expansion of early Indo-Europeans, and marks Caucasoid element in the gene pools of South Siberian populations. Similarly to haplogroup K*, the second frequent haplogroup Q* represents paleo-Asiatic marker, probably associated with the Ket and Samoyedic contributions to the Altaic gene pool. The presence of lineages N2 and N3a can be explained as the contribution of Finno-Ugric tribes, assimilated by ancient Turks. The presence of haplogroups C3xM77, C3c, N*, and O3 reflects the contribution of Central Asian Mongoloid groups. These haplogroups, probably, mark the latest movements of Mongolian migrants from the territory of contemporary Tuva and Mongolia. The data of factor analysis, variance analysis, cluster analysis, and phylogenetic analysis point to substantial genetic differentiation of Northern and Southern Altaians. The differences between Northern and Southern Altaians in the haplogroup composition, as well as in the internal haplotype structure were demonstrated.

DOI: 10.1134/S1022795407050110

INTRODUCTION

Ethnic groups of the Altai–Sayan region are especially interesting for ethnogenetic studies. The gene pool of the indigenous populations of Southern Siberia was formed as the result of long and multistage admixture of local gene pools of different Caucasoid and Mongoloid tribes. Mixing of different Turkic, Mongolian, Ket, and Uralian groups at the basis of the genetic substratum of ancient Indo-European tribes resulted in the formation of the varied genetic diversity pattern of the Altaic populations [1]. Anthropologic heterogeneity of South Siberian populations can be traced at the regional, as well as at the intraethnic levels [2, 3].

Altaians represent the indigenous population of the Altai Republic. Linguistically, they belong to the Turkic group of the Altaic linguistic family, and are subdivided by the anthropologists and ethnographers into two isolated groups of Southern and Northern Altaians. Altaians were relatively recently consolidated into one ethnic group. Earlier, they belonged to different tribal and territorial subdivisions. The division of Altaians into Northern and Southern groups was asso-

ciated with remarkable differences in their ontogeny, as well as with their development at the bases of compositionally different ancestral components. The differences between the groups can be traced at the historical, anthropologic, and linguistic levels [3, 4].

According to the classification of Turkic languages, Southern Altaians were attributed to the Kypchak group. A part of Northern Altaic dialects belongs to the Uyghuric (northeastern) language group [4, 5]. Northern Altaians (including three ethnic groups of Kuman-dins, Chelkans, and Tubalars) were formed as a result of the interactions between ancient Samoyedic, Ket, Ugric, and Turkic tribes. Southern Altaians (Altai-Kizhi, Telengits, and Teleses) were formed as a result of admixture between Turkic and Mongolian tribes. In respect of anthropology, Southern Altaians belong to South Siberian and Central Siberian Mongoloid types. At the same time, in Northern Altaians, Uralian anthropologic type prevails [2, 6, 7].

Despite of intense migrations, structural subdivision of the Altaic ethnic population underwent through no dramatic changes during the last century. Analysis of

the 1897 census showed the isolation of Southern and Northern Altaians, which was manifested not only in the names of seoks (tribes), but also in the tribe settling patterns [6]. Analysis of the tribal composition of Altaians showed that during the last century, their tribal structure at the level of ethnic groups remained unchanged [8, 9]. Genetic demographic analysis of rural populations of the Altai Republic by the surname and tribal structure suggested that the isolation of the two groups of Altaians have been preserved till present [10]. These results, of course, implied the existence of the genetic differentiation of the two groups of Altaians.

The studies of biochemical markers in the indigenous groups of South Siberia, including Altaians, were rather extensive. Analysis of the frequency distributions of the phenotypes and genes of the blood group system, serum proteins, and erythrocytic enzymes revealed the clear subdivision of Altaians into southern and northern groups, as well as the presence of local subgroups within ethnic groups. Taken together, most of the polymorphic systems in Altaians were characterized by the frequencies typical of the Siberian and Central Asian Mongoloids. At the same time, relative to some of the genes examined, Altaians occupied the intermediate position between Caucasoids and Mongoloids, pointing to the presence of ancient Caucasoid component, mostly expressed in Northern Altaians [11–16].

The uniparentally inherited genome components, mitochondrial DNA (mtDNA) and Y chromosome, occupy a special place among the genetic marker systems. Principal importance of the DNA markers of specific human genome regions lies in the fact that they provide individual analysis of maternal and paternal components.

In recent years, mtDNA diversity was extensively studied in the ethnic groups from South Siberia [17–21]. It was demonstrated that mitochondrial gene pools of the populations studied were characterized by different ratios between the Mongoloid and Caucasoid mtDNA lineages. The frequency of Caucasoid lineages declined in the direction from the south to the north, and from the west to the east, which was generally consistent with the anthropological data [18, 19]. Most of these lineages have south Caucasoid origin, while the others belong to east Caucasoid lineages [22].

Studies of the indigenous populations of South Siberia by use Y-chromosomal DNA markers started only recently. Analysis of Y-linked microsatellite markers showed the presence of several major components in the male gene pool of Tuvinians, which were categorized as the Caucasoid and Mongoloid components [23, 24]. Studies of a number of ethnic groups from Altai–Sayan region (including Altaians) also showed the presence of profound paleo-Caucasoid component in their gene pools [25, 26]. These data in general are consistent with those obtained upon the analysis of mtDNA.

The comprehensive studies performed with the use of biallelic Y-chromosomal markers showed that indigenous populations of South Siberia, compared to other indigenous populations of Siberia, possessed the most variable haplogroup composition. Furthermore, Altaians were characterized by a high frequency of haplogroup R1a1, which marked the Caucasoid gene pool component [15, 26, 27]. However, the pattern obtained still rather schematically reflected the actual gene pool structure of Altaians. Effective haplogroup specification requires investigations involving additional samples and possibly larger number of genetic markers.

In the present study, an analysis of the composition and structure of Y-chromosomal haplogroups, distinguished based on the genotyping of 37 biallelic and seven microsatellite markers of its non-recombining part, was performed. The data obtained provided characterization of the gene pool structure in Northern and Southern Altaians. This study is a part of the ongoing project on the analysis of Y-chromosomal gene pool of the indigenous populations of Siberia.

MATERIALS AND METHODS

Total DNA for the experiments was isolated from peripheral blood lymphocytes using standard techniques [28]. Population samples consisting of 146 individuals representing the indigenous population of the Altai Republic were examined. The samples were comprised of males unrelated in at least three generations.

Northern Altaians ($N = 50$) were represented by the samples, formed from the inhabitants of the city of Gorno-Altaiisk ($N = 20$) and the settlements of Kurmach-Baigol ($N = 11$) and Turochak ($N = 19$), Turochak raion. Southern Altaians ($N = 96$) were represented by the samples formed from the inhabitants of the settlement of Kulada ($N = 46$), Ongudaisk raion; the settlement of Beshpel'tir ($N = 43$), Chemal'sk raion; and the settlement Kosh-Agach ($N = 7$), Kosh-Agachskii raion of the Altai Republic.

The Y-chromosomal haplogroup composition and structure was examined using two genetic marker systems: biallelic loci, mostly represented by SNPs, and polyallelic, highly variable microsatellites (YSTR).

Using biallelic markers, attribution of the samples to certain haplogroup was performed. Nomenclature of haplogroups is as defined by the Y-Chromosome Consortium [29]. Genotyping was performed using a set of microsatellite markers, and for each sample its individual STR haplotype was determined. Based on the data on haplotype composition within the haplogroups, their internal diversity and detailed phylogenetic relationships were established.

Biallelic markers. Variability of 37 biallelic markers from the nonrecombining portion of Y chromosome were investigated, including *SRY1532*, *SRY2627*, *M1* (*YAP*), *92R7*, *DYF155S2*, *12f2*, *M3* (*DYS199*), *M9*, *M15*, *M17*, *M20*, *M25*, *M46* (*Tat*), *M70*, *M77*, *M89*,