ISSN 1022-7954, Russian Journal of Genetics, 2014, Vol. 50, No. 2, pp. 180–190. © Pleiades Publishing, Inc., 2014.
Original Russian Text © V.N. Kharkov, K.V. Khamina, O.F. Medvedeva, K.V. Simonova, E.R. Eremina, V.A. Stepanov, 2014, published in Genetika, 2014, Vol. 50, No. 2, pp. 203–213.

HUMAN GENETICS

Gene Pool of Buryats: Clinal Variability and Territorial Subdivision Based on Data of Y-Chromosome Markers

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Received April 23, 2013

Abstract—The structure of the Buryat gene pool has been studied based on the composition and frequency of Y-chromosome haplogroups in eight geographically distant populations. Eleven haplogroups have been found in the Buryat gene pool, two of which are the most frequent (N1c1 and C3d). The greatest difference in haplogroup frequencies was fixed between western and eastern Buryat samples. The evaluation of genetic diversity based on haplogroup frequencies revealed that it has low values in most of the samples. The evaluation of the genetic differentiation of the examined samples using an analysis of molecular variance (AMOVA) shows that the Buryat gene pool is highly differentiated by haplotype frequencies. Phylogenetic analysis within haplogroups N1c1 and C3d revealed a strong founder effect, i.e., reduced diversity and starlike phylogeny of the median network of haplotypes that form specific subclusters. The results of a phylogenetic analysis of the haplogroups identified common genetic components for Buryats and Mongols.

DOI: 10.1134/S1022795413110082

INTRODUCTION

Buryats are the most numerous indigenous ethnic group of Siberia. According to the population census of 2010, there are 461389 Buryats on the territory of Russia [1]. They are mostly concentrated in regions to the east and west of Lake Baikal, the Sayan–Baikal Highlands, and Eastern Baikal, as well as the southeastern part and border of the Central Siberian Plateau. In addition, Buryats live in North Mongolia and in northeast China [2]. The Buryat language belongs to the north subgroup of the Mongol group of the Altai language family and has multiple dialects.

The differences between the dialects largely coincide with the ethnic division of their carrier [2]. Buryat is usually divided into three main dialects by the territory, i.e., western, eastern, and southern [3].

Over the whole territory where they are distributed, Buryats demonstrate clearly expressed Mongol anthropological peculiarities. In general, with small variability of a group of features they may be considered as the representatives of one central-Asian anthropological type. The other types inhibiting adjacent territories did not participate in the formation of Buryat nation [4]. In addition, local variants are separate from the anthropological type of Buryats. There are expressed physical differences between the eastern and western Buryats in general, i.e., eastern Buryats belong to Transbaikalian variants, and western Buryats are included in the Angara–Lena variants of the Central Asian anthropological type. In this respect, eastern Buryats are similar to Mongols and western Buryats are close to Yakuts.

It is assumed that the Mongol ethnic nucleus only began to occupy the leading position among the rest the population of Baikal starting in the 11th–13th centuries, during the formation of the Khitan and Mongol Empires. The available data lead us suspect that there was quite intensive contact between the population of the Baikal region and Mongolian tribes during this period up to the 17th century [5]. Since the 3rd century, the populations of Transbaikalia and Pribaikalia have been included in the Central Asian states, i.e., the Huns, Xianbei, Rouran, and other Turks [6]. The new stage in the history of Buryats starts from the formation of the Khitan (Liao) Empire in the 10th century. The distribution of Mongol tribes in Transbaikalia and the mongolization of Buryats began at this time. The main nucleus of the Buryat nation, which assimilated and united different ethnic groups, were Mongol tribes. The question of the origin of these tribes is one of the main problems in Buryat ethnogenesis [7].

The work continues the series of the studies of indigenous Siberian ethnic groups genofond structure [8–17]. The aim of this study is to characterize the Buryat genofond structure, as well as its regional and intraethnic subdivision based on an analysis of composition and structure of Y-chromosome haplogroups determined by genotyping the vast set of informative SNP and STR markers of it nonrecombining part.

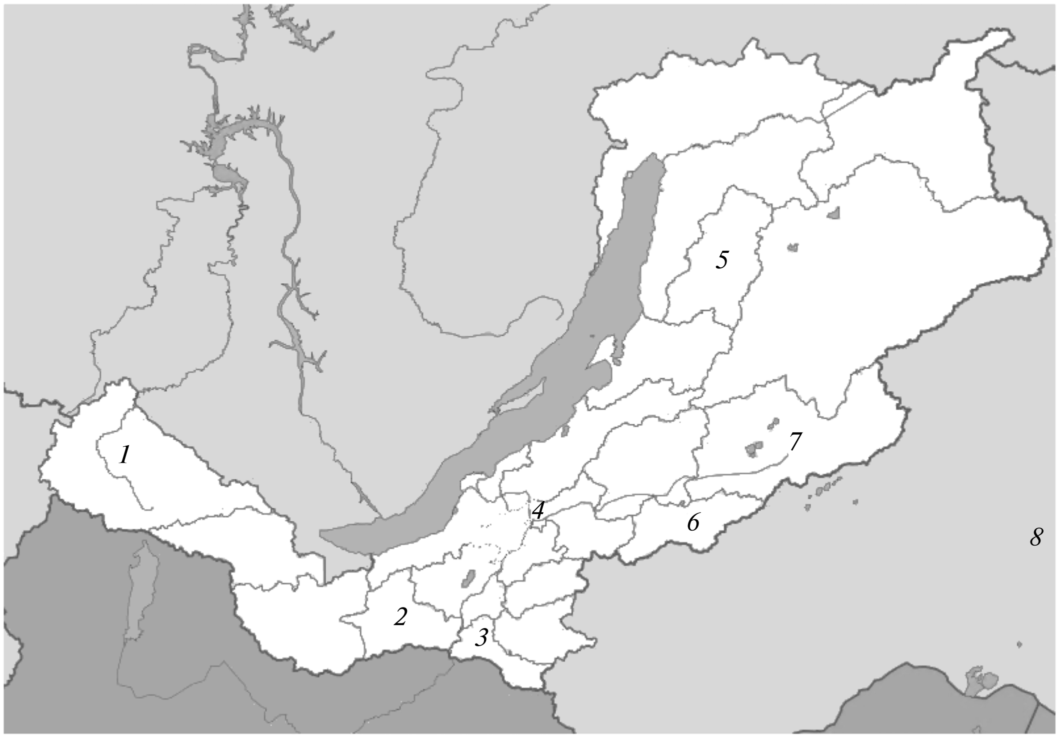

Fig. 1. Map of Buryatia. Numbers designate location of populations (regions). (*1*, Oka; *2*, Dzhida; *3*, Kyakhta; *4*, Ulan-Ude; *5*, kurumkan; *6*, Kizhynga; *7*, Eravninsky; *8*, Aginsky).

Until now, the Buryat genofond has been poorly studied using Y-chromosome markers. There are no separate works devoted to Buryats and, although the most detailed article on South Siberian ethnic groups [14] contains a quite representative sample of Buryats ($N = 238$), no comparative analysis of subethnic groups was conducted. In addition, the results of genotyping Buryat samples were also presented in articles on the phylogeography and origin of different haplogroups [15–17].

MATERIALS AND METHODS

Material for the study was the total DNA extracted from the leukocytes of peripheral blood using standard methods. Population samples of ethnic Buryats with 297 samples in total were studied. All samples from different population samples included in the study were separated into eight geographical groups according to the localization of the place of material gathering (Fig. 1): Okinsky district ($N = 53$) (west of the Republic of Buryatia, ethnoterritorial group of Oka Buryats), Dzhida ($N = 31$) and Kyakhta ($N = 27$) (south, ethnoterritorial group of Selenga Buryats), the Kizhinga ($N = 64$) and Eravninsky ($N = 30$) regions (east, ethnoterritorial group of Khorin Buryats), Kurumkan village ($N = 23$) (northwest, ethnoterritorial group of Barguzin Buryats), Ulan-Ude, Khuramsha (30 km west of Ulan-Ude) ($N = 26$) (ethnoterrotorial group of Kudarinsk Buryats), and Aginskoe village ($N = 44$) (Agin–Buryat Autonomous Region of Chita, Agin Buryats). For the statistical treatment, samples from Ulan-Ude and Khuramsha village were united into one group designated as "Ulan-Ude."

Diallelic loci mainly represented by SNP and polyallelic highly variable microsatellites (YSTR) were used as markers. The classification of the haplogroups was determined based on diallelic markers. The classification of haplogroups is given in accordance with the one suggested by the Consortium with regard to studying the Y chromosome [18] with further modifications [19, 20]. Then, genotyping with a set of microsatellites markers with the determination of individual STR haplotypes was conducted for every sample. Based on the data on the composition of haplotypes inside the groups, their internal diversity and detailed phylogenetic interactions were revealed.

The composition of the haplogroups was studied using 60 markers of the nonrecombinant part of the Ychromosome, including M1 (YAP), M3 (DYS199), M7, M8, M9, M12, M15, M17, M20, M25, M46 (Tat), M47, M56, M64, M67, M70, M73, M77, M86, M89, M92, M102, M117, M119, M120, M122, M124, M128, M130, M134, M157, M170, M172, M173, M174, M175, M178, M198, M201, M204, M207, M217, M223, M231, M242, M253, M267, M269, M324, M346, M407, M458, SRY1532, 97R7, DYF155S2, 12f2, P25, P31, P37, and P43. Genotyping was conducted by PCR and the further study of

DNA fragments was carried by various methods as described earlier [21–22].

Microsatellite markers. The haplotypes were analyzed using 17 microsatellite markers of the nonrecombinant part of the Y chromosome (DYS: 19, 385a, 385b, 388, 389I, 389II, 390, 391, 392, 393, 426, 434, 435, 436, 437, 438, 439). Fluorescent-labeled primers with the HEX, FAM, TET, and NED stains synthesized by Applied Biosystems were used. Genotyping was conducted on ABI Prism 310 and ABI Prism 3130xl genetic analyzers. The following primer sequences were previously described for DYS: in works [8, 23], 19, 389I, 389II, 390, 391, 392, and 393; in the article [24], 385a, 385b, 388, 426, and 438; and, in article [25], 434, 435, 436, 437, and 439. The size of the fragments was analyzed in GeneMapper software. Nomenclature of alleles is given in accordance with the commonly accepted (for DYS389I without taking into account the three-copy TCTG repeat and, for DYS437, without taking into account terminal tandems [TCTG]2–[TCTA]4).

Statistical methods. Genetic interactions between the populations were revealed with multidimensional scaling of genetic distances (Slatkin F_{st}). The calculations and charting were carried out using the STATISTICA 7.0 software package. The genetic diversity was assessed by the Nei formula [26]. Genetic differentiation of populations was assessed using the analysis of modular variance (AMOVA) [27]. The coefficient F_{st} was applied with 10000 permutations of the original data set. The significance of the interpopulation differences with regard to the frequencies of haplotypes and YSTR haplotypes was evaluated by an exact test of population differentiation (number of Markov chain pitch = 10 000, number of negligible steps = 1000, significance level = 0.05). Slatkin matrices of pairwise distances (F_{st}) were calculated using 100 permutations of original data set. The calculations were conducted in the ARLEQUIN 3.11 software package (http://cmpg.univbe.ch/software/arlequin3) [28]. The median networks of Y-chromosome haplotypes were constructed in Network v. 4.6.1.1 software (www.fluxus-engineering.com) via the Bandelt median network method with the subsequent application of the reduced-median (RM) and median-joining (MJ) algorithms (parameter ε was taken to be 0) [29, 30]. When constructing networks in order to take into account the difference in mutation speed, the weight in proportion to its variability in the studied set of haplotypes was assigned to each STR locus. The time during which the observed diversity of haplotypes in haplogroups was generated (Age) was assessed based on the rms differences in the number of repeats between all haplotypes and founder haplotype [31]. An evolutionary rate of mutation of 0.00069 per locus for 25 years [31] and a genealogical rate of mutation of 0.0025 per locus for 25 years [32] were used.

RESULTS AND DISCUSSION

Frequencies of Y-Chromosome Haplogroups in Samples of Buryat Population

Eleven haplogroups (C3*, C3c, C3d, E, N1b, N1c1, O3a*, O3a3c*, O3a3c1, R1a1a and R2a) were revealed in the Buryat gene pool (Table 1). The highest frequency in all seven territorial groups of Buryats was observed for N1c1 and C3d haplogroups. Only these haplogroups were present in all eight geographical groups of populations. Their total share varies from 55% in the Kyakhta region to 90% in Eravninsk and Kizhinga. The only exclusion is the population of Selenga Buryats from the Dzhida region where the haplogroups C3* and R2a were the most numerous.

The populations tested in this work are separated into two groups by the frequency of representation in their gene pool of the most frequent haplogroup N1c1, which may be conditionally designated as "western" and "eastern." In the western group (Oka, Barguzin, Selenga and Kudarinsk Buryats), the frequency of the N1c1 haplogroup ranges from 8.7% (Kurumkan village) to 37% (Kyakhta region). The east group (Khorin and Agin Buryats) differ by the much higher frequency of N1c1, i.e., from 60% in the Eravninsk region to 78% in Kizhinga. The difference in frequencies between relatively close samples of Ulan-Ude city and Kizhinga regions is 40%. Thus, the Buryat gene pool is characterized by clinal changes in the N1c1 frequencies in the populations with minimal values in the west and maximal in the east.

The opposite situation is observed for haplogroup C3d, which is the second most common in the total genofond (28.8%). The maximal frequency of this lineage is observed in the western group (maximum in Barguzin Buryats of Kurumkan region was 74%) and minimal frequency in Agin Buryats (4.5%) and quite low in Khorin Buryats of Kizhinga region (12.6%). Therefore, there is a reverse cline on the C3d haplotype group in the gene pool of the Buryat ethnic group.

A remarkable feature of the southern group of populations (Selenga Buryats from Kyakhta and Dzhida regions) is the rather high frequency of R1a1a (18.5%) and R2a (22.6%) haplogroups. Along with these regions, the R1a1a haplogroup was also revealed in the Oka region (3.7%). In the samples from Ulan-Ude and the Dzhida region, the C3* lineage is present with a frequency of more than 20%.

The other haplogroups found in Buryats, i.e., E, N1b, O3a*, O3a3c*, O3a3c1, i.e., are present with small frequencies and only in some populations. The total frequency of the haplotypes from group O3 is significant in Oka and Selenga Buryats from the Kyakhta region (9 and 11%, respectively). The obtained results correspond well to anthropological and linguistic data on the subdivision of the Buryat ethnic group.

The analysis of haplotypes frequencies by the method of multidimensional scaling demonstrates

Table 1. Distribution of Y-chromosome haplotypes in population samples of Buryats

Haplogroup	Frequency, % (*N*)								
	West		South			East			Total ($N = 298$)
	Oka region ($N = 53$)	Kurumkan region ($N = 23$)	Ulan-Ude ($N = 26$)	Kyakhta region ($N = 27$)	Dzhidinsky region ($N = 31$)	Kizhynginsky egion ($N = 64$)	Eravninsky region ($N = 30$)	Agin AR ($N = 44$)	
C3*	1.9 (1)	8.7 (2)	23.1 (6)	11.1 (3)	22.6 (7)	1.5 (1)	–	4.5 (2)	7.3 (22)
C3c	3.8 (2)	–	7.7 (2)	3.8 (1)	3.2 (1)	1.5 (1)	6.7 (2)	4.5 (2)	3.7 (11)
C3d	58.5 (31)	73.9 (17)	30.8 (8)	18.5 (5)	19.3 (6)	12.6 (8)	30.0 (9)	4.5 (2)	28.8 (86)
E	–	–	–	–	–	4.7 (3)	3.3 (1)	–	1.3 (4)
N1b	–	4.3 (1)	–	–	–	–	–	–	0.3 (1)
N1c1	22.6 (12)	8.7 (2)	38.4 (10)	37.0 (10)	19.3 (6)	78.2 (50)	60.0 (18)	77.4 (34)	48.0 (142)
O3a*	–	–	–	11.1 (3)	3.2 (1)	–	–	2.3 (1)	1.7 (5)
O3a3c*	7.5 (4)	–	–	–	–	–	–	–	1.3 (4)
O3a3c1	1.9 (1)	4.3 (1)	–	–	–	1.5 (1)	–	–	1.0 (3)
R1a1a	3.7 (2)	–	–	18.5 (5)	9.8 (3)	–	–	4.5 (2)	4.0 (12)
R2a	–	–	–	–	22.6 (7)	–	–	2.3 (1)	2.6 (8)
H	0.6089 ± 0.0623	0.4545 ± 0.1234	0.7262 ± 0.0423	0.7977 ± 0.0479	0.8387 ± 0.0266	0.3770 ± 0.0724	0.5632 ± 0.0720	0.4027 ± 0.0928	0.6799 ± 0.0203

N—sample volume.

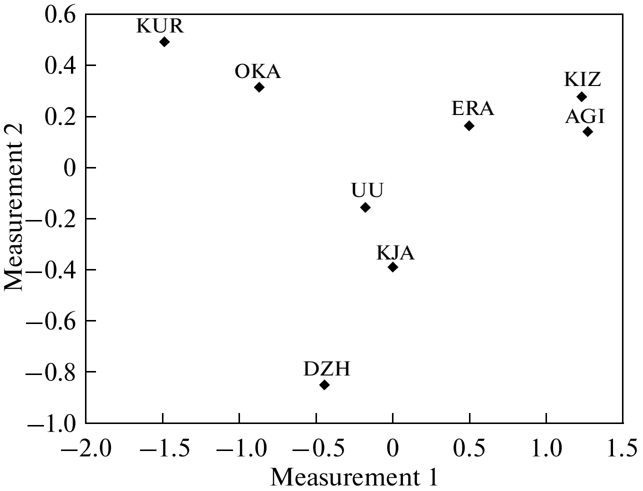

Fig. 2. Position of Buryats population in principle component space by Y-chromosome haplogroup frequencies. Designation of samples by region: OKA is Oka, DZH is Dzhida, KJA is Kyakhta, UU is Ulan-Ude, KUR is Kurumkan, KIZ is Kizhinga, ERA is Eravninsk, AGI is Agin).

vivid differences between Buryat populations (Fig. 2). All populations were separated into three groups locating on the graph in accordance with their geographical localization (the only exclusion is Barguzin Buryats of the Kurumkan region). Cluster analysis indicated almost the same picture of the localization of populations (picture is not shown). The majority of the pairs of compared populations differ statistically significantly from one another by the results of the exact test of the population differentiation by haplotype differences. The samples of eastern regions (Khorin from the Kizhinga, Eravninsk, and Agin regions) were genetically closest; no significant differences were observed between them. The southern population group (Ulan-Ude and Kyakhta regions) also shows no differences with the exception of the Dzhida region, which differs from Ulan-Ude. Oka and Barguzin (Kurumkan village) Buryats do not differ significantly either. This is quite natural because the Buryat population of the northern regions of Transbaikalia was formed as a result of intensive migrations from the west in 17th–18th centuries [2].

Genetic Diversity of Populations

Genetic diversity (*H*) was assessed based on the frequencies of the haplogroups in eight geographical distanced groups and in the total sample (Table 1). In haplotype frequencies, the maximal diversity was obtained for Selenga Buryats from the Kyakhta (0.79) and Dzhida (0.84) regions. According to the data of tribal structure analysis, this region of Buryatia has the most mixed ethnic picture as a result of the mixture of local tribes with natives from the western part of Buryatia and migrants from Mongolia [2]. The minimal diversity is recorded for the Khorin and Agin Buryats, which is connected with very high frequency of the N1c1 haplogroup. The low genetic diversity in the sample of Barguzin Buryats from the Kurumkan region (0.45) is conditioned by the dominance of the C3d haplogroup.

In general, Buryats differ by the very specific composition of haplogroups and decreased genetic diversity compared to the other South Siberian ethnic groups (Altai, Teleuts, Khakhas, Tuvins) [9, 11–13]. Previously, based on the material from these populations, it was indicated that based on Alu-repeats Buryat samples are also characterized by the relatively low genetic difference due to the lack of heterozygotes, which may be a consequence of the high level of inbreeding [33]. The diversity of Buryats according to the data on the variability of nucleotide sequences HVI and HVII mtDNA is quite high [34].

Genetic Differentiation of Populations

Genetic differentiation of the studied populations was assessed in the analysis of molecular dispersion (AMOVA). In the division of Buryats into eight groups of samples in accordance with the geographical place of birth, the values of F_{st} indicated their significant degree of interpopulation differentiation. The total level of genetic differentiation of the studied system of populations was 19.81%. This value is quite high for the intraethnic level. With the division of population into three groups (western, eastern, and southern), the share of total diversity of intergroup differences was 21.82%. This value is much higher than for the other South Siberian ethnic groups, which indicates significant subdivision of the Buryat gene pool. The sample from the Kurumkan region is included into the eastern group, along with the sample of Oka region because, in the results of the exact test of population differentiation, they do not show statistically significant differences. The proportion of differences between the population inside three geographical groups was lower in degree, i.e., 2.07% for the western group (Oka and Kurumkan regions), 2.74% for the southern group (Kyakhta and Dzhida regions and Ulan-Ude), 3.34% for the eastern group (Kizhinga, Eravninsk, and Agin regions), which well correlates with the results of the exact test of population differentiation. The western group of populations differs a lot from the eastern group (F_{st} = 35.34%). The differences between the southern group and the western and eastern groups are less (F_{st} = 11.70 and 15.40% respectively). Thus, Buryats are remarkable for the significant level of intraethnic subdivision of their gene pool. The significant genetic homogeneity and absence of statistically significant differences between populations is typical in the range of subethnic groups, which indicates the unity of gene pool in ethnoterritorial groups of Buryats. The obtained results confirm the formation of

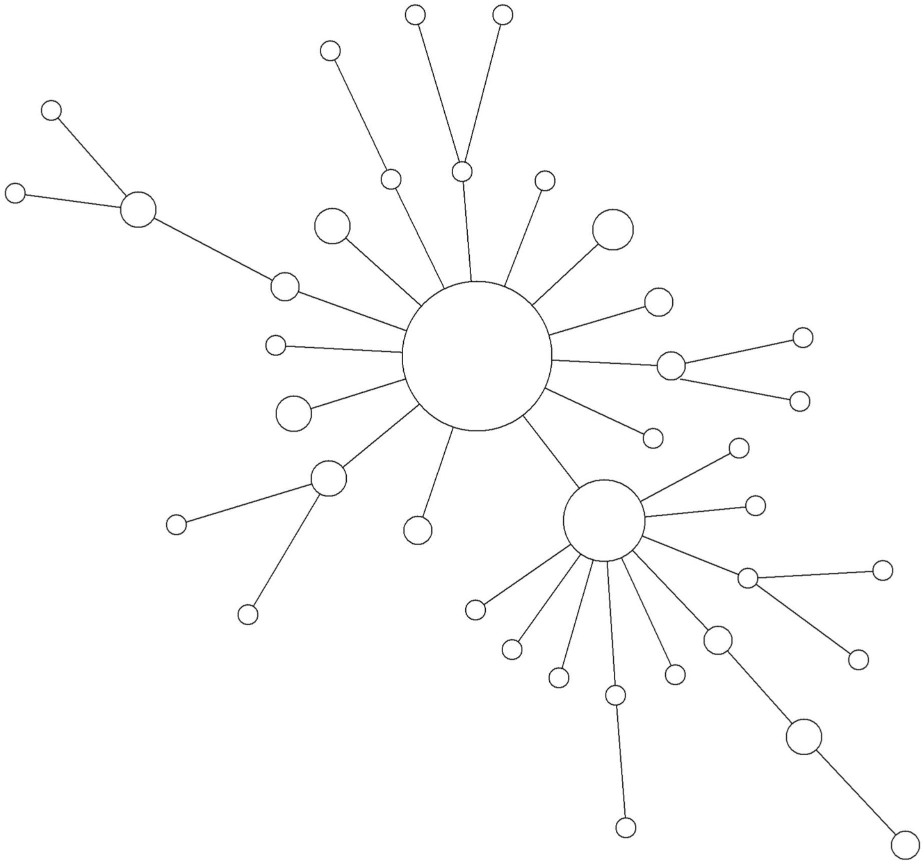

Fig. 3. Median network of microsatellite haplotypes from N1c1 haplogroup in Buryats.

Buryats as a multicomponent ethnic group with different origins of the populations. Very similar results were obtained earlier in the characterization of the Y-chromosome gene pool of the Khakhas, for which the significant genetic homogeneity between different samples of Kachintsy and Sagaitsy with significant differentiation between subethnic groups is typical [12].

An earlier study of the mitochondrial gene pool of Buryats in the material of the same population samples has also revealed significant genetic heterogeneity in them. Haplogroups C and D, which are present with almost equal frequencies, are the main haplogroups in the mitochondrial gene pool of Buryats, as well as in other ethnic groups of South Siberia. The Agin population is also characterized by a decreased frequency of these main lineages (by 21%) and high frequency of haplogroup A (7% compared to 1% in Kurumkan and Ulan-Ude) [35].

Phylogenetic Analysis of N1c1 Haplogroup in Buryats

N1c1 haplogroup is the most common in the total sample of Buryats (48% in total). It prevails in the ethnoterritorial groups of Agin and Khorin Buryats, which belong to Khorin dialects. The median network of this haplogroup demonstrate that the majority of Buryat haplotypes form detached from the other Siberian ethnic groups highly specific spectrum with star-like phylogeny (Fig. 3) [10–12, 15, 42]. N1c1 haplotypes are separated into two uneven clusters. For the majority of the samples, the allele 14 by the DYS393 locus is typical and forms the main cluster of haplotypes (N1c1-A). For the smaller cluster, allele 13 (N1c1-B) is present in the DYS393 locus. It may be concluded that the genetic component marked by this haplogroup inside the Buryat ethnic group indicates the high founder effect and speedy growth of the population of Buryat tribes, as in the situation observed previously in Yakuts [10].

The modal haplotype for N1c1-A (14-11-13-12-11-16-23-10-14-14-11-8-11-12-8-10-10) is the most common in Buryats and Mongols. The other haplotypes of this cluster also overlap with Mongol haplotypes. All samples of the most eastern sample of Agin Buryats, where the N1c1 lineage dominates, only belongs to this groups of haplotypes. The smaller cluster of haplotypes N1c1-B is specific to Buryats and is not revealed in Mongols and sample from Agin Autonomous Area. F_{st} value by N1c1 haplogroup for Buryat samples was 20.65%. The exact test of population differentiation indicates the reliable differences between all territorial groups by N1c1 haplogroup. The age of

Table 2. Assessment of age (thousand years) of haplotypic variety generation in haplogroups N1c1 and C3d

Haplogroup	"Evolution" mutation rate [31]	"Genealogic" mutation rate [32]
N1c1, total	3.89 (±0.91)	1.07 (±0.25)
N1c1-A	3.21 (±0.82)	0.88 (±0.23)
N1c1-B	2.50 (±1.01)	0.69 (±0.28)
C3d-A, buryats	3.36 (±1.17)	0.92 (±0.32)
C3d-A, soyots	1.97 (±0.43)	0.55 (±0.12)
C3d-A, hamnigans	1.59 (±1.00)	0.44 (±0.28)
C3d-A, total	3.01 (±1.06)	0.83 (±0.29)
C3d-B	1.05 (±0.44)	0.29 (±0.12)

the generation of haplotype diversity shows that N1c1-B is significantly younger than N1c1-A (Table 2).

The graph of the multidimensional scaling of genetic distance matrix by N1c1 haplotypes between Asian ethnic groups vividly demonstrates the genetic differences between Buryats and other populations of Siberia (Fig. 4), which is conditioned by the specificity of their haplotypes. Mongols turned out to be the group closest to Buryats. According to the second measurement, Buryats are the most remote from the other populations, except for Soyot and Teleuts.

The origin of the N1c1 haplogroup in the Buryat gene pool is directly connected with Mongols. The distribution of Mongol tribes to the North on the territory of contemporary Buryatia reflects in the overlapping of the majority of the spectrum of N1c1-A cluster haplotypes. Specific for Buryats haplotypes cluster N1c1-B is probably a result of the population growth of the ancestral population after the separation of Protoburyats and Mongols. The gradual accumulation of differences in the haplotype content of N1c1 between western and eastern Buryats is apparently determined by the influence of migration factor. It appears that this lineage also marks the expansion of the Khorins.

The noticeable decrease in frequency of N1c1 haplogroup in western direction and the presence of a significant proportion of unique haplotypes in Buryats indicate the absence of the intensive gene drift from Buryats to Mongols.

Phylogenetic Analysis of C3d Haplogroup in Buryats

Haplogroup C3d (29%) is the second most common in Buryats, is dominant in western ethnoterritorial groups (Oka, Barguzin, and Kudarinsk Buryats), and almost completely resembles the structure of N1c1. It is really surprising how much the haplotypes median networks of N1c1 and C3d-A haplogroups are similar to each other in Buryats (Fig. 5). C3d haplogroups also shows a starlike phylogeny of haplotypes and their separation into two clusters. As for the N1c1 haplogroup, with regard to the C3d haplogroup, Buryats demonstrate differences in the specificity of the two main clusters. Haplotypes of the main cluster inside C3d-A were revealed in almost all analyzed samples of Buryats (modal haplotype 15-11-18-12-10-15-23-10-11-14-11-11-11-12-8-10-12). As opposed to modal haplotype C3d-A and haplotypes close to it, which were encountered in almost all Buryat samples, as well as in the Soyot and Altai [16], the second most common haplotype from the studied population is specific for the population of Khorins from Kurumkan and Eravninsk regions. This geographic specificity apparently reflects the differentiation of different Buryat tribes.

SNP marker M407, which defines the C3d subgroup, was only discovered recently and, in the first work [36], it was only revealed in two Yakuts and one Chinese individual. It was originally assumed that it is quite rare,but some years after the area of this haplogroup, it had expanded significantly. Zhong et al. [37] revealed 23 samples that belong to C3d, including thirteen Hans, five Manchus, and two Uighurs, as well as one each of Bai, Tujia, and Khmer.

On the territory of Siberia this line was revealed in addition to Buryats in Teleuts (5.3%) and Tuvinians (1.9%) [38]. According to the data of Malyarchuk et al. [16], C3d was also revealed in Hamnigans (52.9%), Mongols (15.2%), Soyot (53.6%) and Kalmyks (12.1%). It appeared that two samples from a medieval burial ground on the territory of Yakutia also belong to this haplogroup [39].

The C3d haplogroup is common mainly for Mongol-speaking ethnic groups and neighboring Turkic speaking Soyot and Tuvinians. The main area of this group is quite narrow and limited to the territory of Mongolia and South Siberia (except for Kalmyks, who are also of Mongol origin). This line seems to mark the demographic expansion of Mongol populations in Central Asia.

According to the structure of haplotypes, all samples that belong to C3d are divided into two uneven groups. The combination of alleles by DYS385a,b 11-18 loci and sometimes 11-17 or 11-19 (detected in capillary electrophoresis as two different PCR products) is typical of the majority of samples; they form the main haplotype cluster (C3d-A). For the smaller cluster (C3d-B), only the value 11-11 (detected as one PCR product) is revealed. The development of this branch of haplotypes is unlikely to be connected with the decrease in the size of DYS385b loci in seven tandem

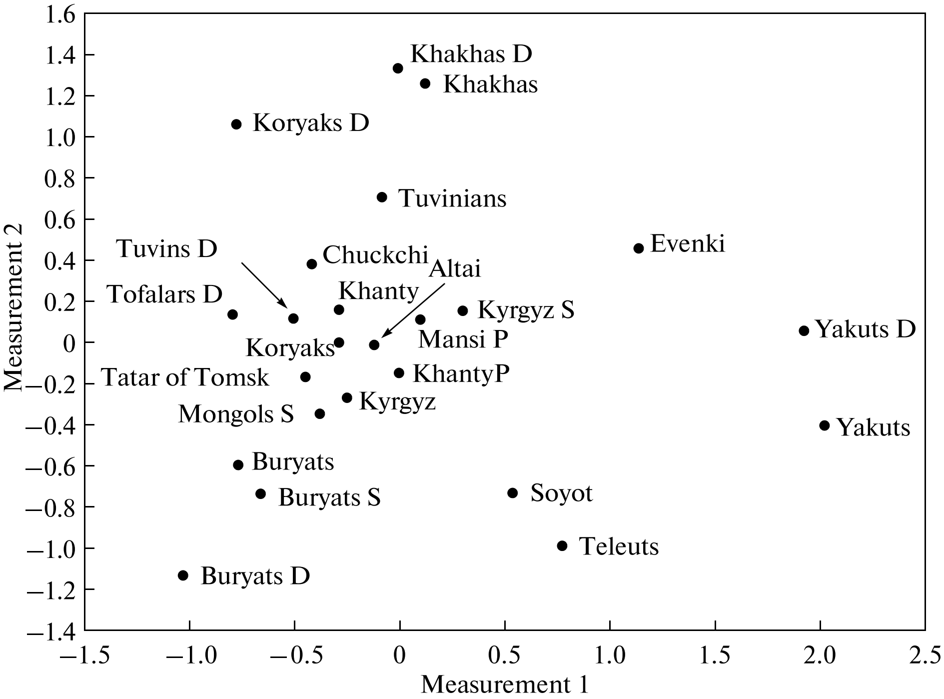

Fig. 4. Multidimensional scale of matrix of genetic distances by YSTR-haplotypes of N1c1 haplogroup (Asian ethnic groups). Populations marked by additional Latin letter were taken from literature. D is Derenko et al., 2007 [15], P is Pimenoff et al., 2008 [42], S is Sorenson Molecular Genealogy Foundation Database [43].

repeats at once. Most likely, a large deletion took place of either the whole locus or part of it, including the primer region. This event may be considered an analogue of diallelic mutation that leads to the appearance of the C3d1 sublineage in the C3d haplogroup. This sublineage is common mainly for Kalmyks, Tuvinians, and Teleuts. Four out of five samples of Buryats of this cluster belong to the western sample from the Oka region. Among eastern Buryats, Hamnigans and Mongols were not encountered at all.

The evaluation of the degree of genetic differentiation of seven ethnic groups (Buryats, Hamnigans, Tuvinians, Soyot, Mongols, and Teleuts) by all haplotypes revealed extremely high interpopulation differences between them (F_{st} = 62.63%). In this case, statistically significant differences (with significance level $p < 0.05$) are not observed in a pairwise comparison of the samples in the Buryats–Hamnigans–Soyot and Tuvinians–Teleuts–Kalmyks–Mongols groups. All pairwise comparisons of the samples from two different groups have statistically significant differences. This is connected with the fact that the clusters C3d-A and C3d-B are distributed unevenly among the studied populations.

If one removes samples from C3d-B cluster and estimates the genetic differentiation of Buryats, Hamnigans, and Soyot (because only C3d-B is present in Teleuts and, in Tuvinians, Mongols, and Kalmyks C3d-A, the cluster is represented by single samples) from the analysis, then interpopulation differences make the proportion of the total variability negligible (F_{st} = 0.31%). In this case, the exact test of population differentiation logically does not reveal any significant differences between different ethnic groups. The same analysis of the samples of Buryats, Tuvinians, Teleuts, Kalmyks and Mongols by C3d-B cluster only, showed greater significant genetic differentiation between them (F_{st} = 25.69%) and statistically significant differences between the pairs Buryats–Kalmyks and Buryats–Tuvinians.

Thus, by C3d haplogroup all ethnic groups are divided into two subgroups: (1) genetically close to each other Buryats, Soyot, and Hamnigans may be conditionally designated as East group; (2) more heterogeneous West group including Mongols, Kalmyks, Tuvinians, and Teleuts. The estimation of time of generation of haplotype diversity indicates that the most ancient is the component C3d in Buryats (Table 2). In all other ethnic groups the diversity is much smaller. However, for Mongols, it may be connected with the rather smaller size of the sample.

The presence of C3d haplogroup in the Buryat gene pool is certainly also connected with Mongols. The differences in the evaluation of the age of the main clusters of haplotypes in N1c1 and C3d haplogroups may reflect two different waves of migration from the territory of Mongolia. East Mongolian tribes that carry C3d-A haplogroup in their genofonds distributed it mainly in the northern areas, where Buryats and Hamnigans were formed. Two haplotypes with

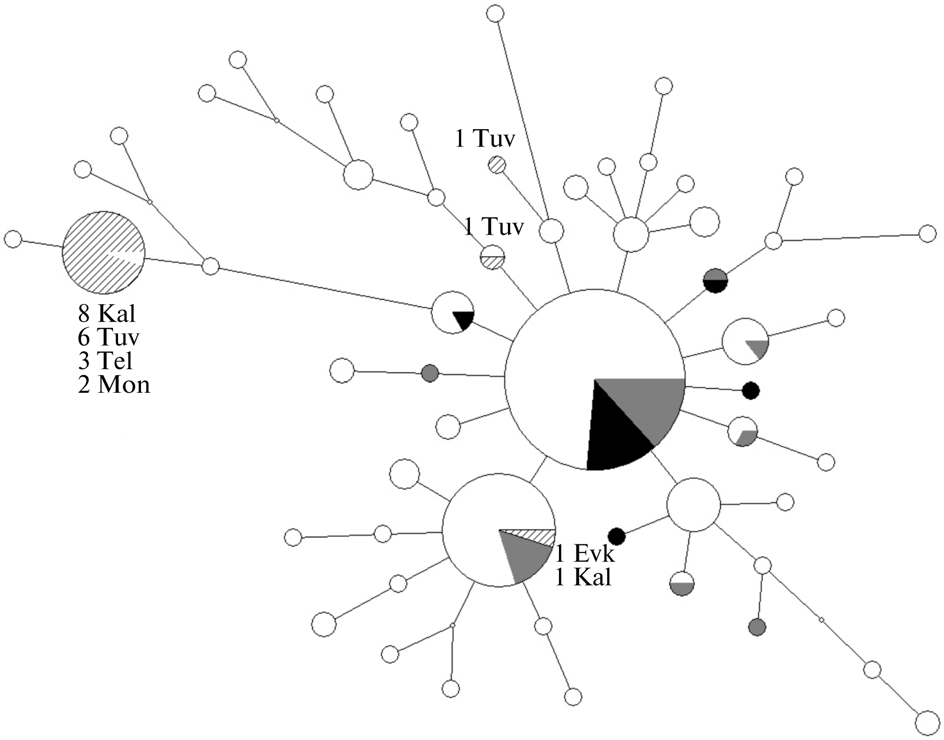

Fig. 5. Median network of microsatellite haplotypes of C3d haplogroup built on the basis of our own data and literature data [15]. White color designates Buryats, gray is Soyot, black is Hamnigans, all other ethnic groups are shaded and designated as follows: Evk is Evenks, Kal is Kalmyks, Mon is Mongols, Tel is Teleuts, Tuv is Tuvinians.

high frequency and the starlike distribution of their derivatives on the median network of C3d-A must reflect the fast demographic growth of Protoburyat tribes. Their genetic connection with Mongols is obvious. The appearance and expansion of C3d-B cluster have apparently occurred near the borders of Tuva and Mongolia, where it came to Teleuts and Tuvinians.

The majority of the samples that belong to the third haplogroup C3* (7.3%) with regard to frequency in Buryats belongs to a starlike cluster of haplotypes that are common for Central Asian and South Siberian populations, which is presumably connected with the expansion of Mongols in the 8th century and, probably, with the descendants of Genghis Khan [40, 41]. Haplotype diversity in this cluster is quite low for Buryats. Thus, this group reflects the much later influence of Mongol tribes than the N1c1 and C3d haplogroups.

Therefore, the study provides evidences, first, on the significant genetic subdivision of the indigenous population of Buryatia into three groups of populations (western, eastern, and southern) and, second, on the high genetic proximity of Buryats and Mongols. The spectrum and frequencies of haplotypes of all main haplogroups between these two ethnic groups are very close. The differences between Buryats and other South Siberian ethnic groups are much higher than between Buryats and Mongols. Haplogroups typical of populations of Altai–Sayan, i.e., R1a1a, N1b, and Q1a3, i.e., are almost absent in Buryat gene pool, and the haplotype composition in the N1c1 line is completely unlike the one observed in the Khakhas, Tuvinians, and Altai. Buryats stand out against other populations of South Siberia by high frequency of C3d haplogroup. A strong founder effect and differences between ethnoterritorial groups by the YSTR spectrum are observed in Buryats by two main haplogroups (N1c1 and C3d).

In summary, it may be concluded that the genetic barrier that separates the populations of Altai, Sayan, and all of Western Siberia from the more eastern neighbors with a predominance of the Central Asian component goes on the border between Sayan and Transbaikalia. This genetic border is confirmed both by the Y chromosome [38] and other marker systems [44].

ACKNOWLEDGMENTS

The works was conducted under the financial support of Federal Target Program “Studies and Developments on Priority Areas of Science and Technology” (11.519.11.2036), Federal Target Program “Scientific and Pedagogical Staff of Russia” (agreement no. 8042) and the Russian Foundation for Basic Research (projects nos. 12-04-00595a and 13-04-02023a).

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Translated by N. Sokueva