
HUMAN
GENETICS

Molecular Genetic Differentiation of the Ethnic Populations of South and East Siberia Based on Mitochondrial DNA Polymorphism

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Received January 9, 2002; in final form, March 25, 2002

Abstract—Using the data on mitochondrial DNA (mtDNA) polymorphism, genetic structures of the ethnic groups inhabiting South and East Siberia, including Altaians, Buryats, Tuvinians, Tadjins, Tofalars, Yakuts, and Evenks were described. Mitochondrial gene pools of the populations examined were characterized by different ratios between Mongoloid (M*, C, D, E/G, G, A, B, and F) and Caucasoid (H, HV, I, J, K, T, U, and X) mtDNA lineages. All the populations studied carried a marked Mongoloid component, maximum frequency of which was observed in Evenks (92.4%) and Buryats (90.1%). Maximum frequencies of Caucasoid mtDNA lineages were detected in Tofalars (20.7%) and Yakuts (14.5%). Statistically significant interpopulation differences regarding the frequencies of mtDNA haplogroups were observed between all populations examined, excluding the pairs of Evenks–Yakuts, Evenks–Tuvinians, and Tuvinians–Tadjins. Differentiation of the ethnic groups inhabiting South and East Siberia, as well as Central and Middle Asia, is discussed based on genetic, linguistic, and anthropological data.

INTRODUCTION

Based on anthropological, historico-ethnographic and anthropological data, most of the ethnic populations of South and East Siberia are combined into Central Asian cultural zone [1]. The main area of this zone lies in Mongolia, and its northern periphery span the territories of Siberia inhabited by Southern Altaians, Tuvinians, Buryats, Yakuts, Khakassians, and Tofalars. All these populations are characterized by the Central Asian anthropological type, although in some cases (for example, in Tofalars) anthropological features typical of Baikal type are observed. It is suggested that formation of Baikal anthropological type was associated with the ancient population inhabiting taiga regions of the southern East Siberia and northern Cisbaikalia. This type characterizes Siberian branch of Mongoloids, specifically, Tungus-Manchurian populations, i.e., Evenks, Evens, Negidals, Ulchis, and some others. On the other hand, based on craniological data, it was established that Central Asian type was indistinguishable from Baikal type, and thus anthropological closeness of Turkic, Mongolian, and Tungus-Manchurian populations can be explained by their common ancestry [1, 2]. However, the use of anthropological data for more detailed analysis aimed at investigating the origin and

ethnic history of certain populations is hampered by their incompleteness, methodical incompatibility, and, in some cases, also by the absence of paleoanthropological data.

On the contrary, modern molecular genetic methods offer the opportunity to reconstruct the process of the gene pools development based on their present-day state. Complex analysis of variation in highly polymorphic genetic systems, mitochondrial DNA (mtDNA) and nonrecombining portion of the Y chromosome, characterized by the lack of recombinations and the inheritance in one of the parental lineages, makes it possible to estimate the ratios between certain components in the gene pools of the populations with special reference to the contribution of basal and introduced lineages [3]. Phylogeographic analysis of the data on the distribution of mtDNA and Y-chromosome monophyletic clades offers an opportunity to localize the sources of their origin and to estimate their evolutionary age [4, 5].

Our previous investigations on mtDNA polymorphism in ethnic populations of Altai–Sayan and Baikal regions showed that the gene pools of these populations are represented by different ratios between Mongoloid (M*, C, D, E, G, A, B, and F) and Caucasoid (H, U, T, J, and K) mtDNA lineages. The proportion of Cauca-

soid lineages increased westward, reaching its maximum in Shorians (35.7%) [6]. Despite the similarity of the gene pool structures, the populations examined were characterized by the high level of differentiation. Because of this, it seems reasonable to include into the analysis more populations inhabiting large territory of South and East Siberian.

In the present study new data on the structure and diversity of mitochondrial gene pools of Altai-speaking populations, including Tungus-Manchurian ethnic groups are presented. These data provided performance of the first estimates of phylogenetic relationships between the populations examined with special attention to their ethnolinguistic and anthropological diversity.

MATERIALS AND METHODS

Tissue samples (blood and hair follicles) were collected from 538 individuals representing seven ethnic populations of Northern Asia during expeditions in 1998–2001. Judging by questionnaire data, all individuals examined were unrelated at least in three generations. The sample of Tofalars ($N = 58$) was represented by the inhabitants of the settlement of Alygdzher, Nizhneudinskii raion, Irkutsk oblast. The sample of Altaians ($N = 110$) consisted of Maimalars ($N = 14$) from Shebalinskii, Maiminskii, and Chemal'ckii raions; Lebedins ($N = 4$) and Kumandins ($N = 2$) from Turochanskii raion; Telenghits ($N = 17$) from Kosh-Agachskii and Ulaghanskii raions; and Altai-Kizhi ($N = 73$) from Ust'-Kanskii, Ongudaiskii and Ust'-Koksinskii raions of the Altai Republic. The sample of Tuvinians ($N = 90$) consisted of residents of the settlements of Dzun-Khemchenskii, Mongun-Taiga, Bai-Taiga, Sut-Khol'skii, Ovyurskii, Tes-Khemskii, Erzya, and Tandinskii raions of the Tuva Republic. Taking into consideration ethnic and territorial differences between Tuvinians and the specific combination of anthropological features in Eastern Tuvinians (Todjins) [7], the samples were collected from Todjins ($N = 48$) inhabiting the settlements of Todja raion of the Tuva Republic (Toora-Khem, Ii-Khem, Ulug-oo, and Adyr-Kezhig). The sample of Buryats ($N = 91$) consisted of residents of the settlements of Kizhinginskii, Khorinskii, Zakamenskii, Eravninskii, Selenginskii, Barguzinskii, and Kabanskii raions of the Buryat Republic. The sample of Western Evenks ($N = 79$) was represented by the inhabitants of the settlements of Chirinda, Ekonda, Kislokan, Poligus, Strelka-Chunya, Solomai, and Tura of Evenk Autonomous okrug. The sample of Yakuts ($N = 62$) consisted of Esseisk Yakuts ($N = 22$), inhabiting the settlement of Essei, Evenk Autonomous okrug, and the settlements of Churapchinskii, Suntarskii, Kobyaiskii, Anabarskii, and Gornyi raions of the Sakha-Yakutia Republic.

In our previous investigation of the Altai–Sayan populations [6] a sample of Atai-Kizhi formed by the residents of the settlements of Mendur-Sokkon and Kaisyn, Ust'-Kansk raion of the Altai Republic was examined. In the present study we examined an Altai-

Kizhi sample comprised of the individuals from other settlements along with the representatives other ethnic territorial groups of Altaians, namely, Maimalars, Lebedins, Telenghits, and Kumandins. Taking into consideration the relatively high population number of Altaians and the existing ethnic territorial, anthropological, and other differences between Northern and Southern Altaians, we think it expedient to examine the degree of differentiation between different groups of Altaians. Since comprehensive information has been currently obtained only for Altai-Kizhi, in the present study this group is examined together with other groups of Altaians, represented so far by rather small samples. The above is also true for Tuvinians and Buryats. The sample of Tuvinians examined previously was rather small (36 subjects) and mainly consisted of individuals from the western regions of the Tuva Republic [6]. The sample examined in the present study was substantially enlarged in size and included Tuvinians from all ethnic territorial groups. Moreover, Eastern Tuvinians (Todjins) were analyzed separately as an independent group. Similarly to our previous study [6], the re-examined sample of Buryats was comprised of individuals from different regions of the Buryat Republic, but the number of specimens was substantially augmented. Thus, at this stage of investigation the data on mtDNA variation were for the first time obtained for representative samples (more than 100 subjects for Tuvinians and Buryats and more than 200 subjects for Altaians) of South Siberian ethnic groups.

Total DNA was extracted from the tissue samples using standard techniques [8]. Screening for polymorphic sites determining the main haplogroups of mtDNA types distributed in the populations of Eurasia (Table 1) was conducted through the analysis of mtDNA fragments amplified in polymerase chain reaction with the primers described in [9–12]. Restriction fragments were separated by electrophoresis in 8% polyacrylamide gel. Gels were stained with ethidium bromide and DNA fragments were visualized in the UV light. Polymorphism was scored by the presence (+) or absence (–) of restriction endonuclease recognition sites.

Diversity of mtDNA types (h) was calculated according to Nei and Tajima [14]:

$$h = (1 - \sum x^2)N/(N - 1),$$

where x is the population frequency of each mtDNA type and N is the sample size.

Statistical significance of interpopulation differences with respect to the frequencies of mtDNA type haplogroups was evaluated using the identity index I [15].

Phylogenetic analysis was carried out using the PHYLIP 3.5c software package [16]. Genetic distances between the populations were calculated from the frequencies of mtDNA haplogroups in each population using the GENDIST program. Phylogenetic trees were constructed using the NJ (neighbor-joining) method [17].

Table 1. Polymorphic restriction variants determining haplogroups of mtDNA types in the population of Eurasia

mtDNA haplogroup	Key restriction variants
M:	+10394 <i>DdeI</i> , +10397 <i>AluI</i>
C	+10394 <i>DdeI</i> , +10397 <i>AluI</i> , -13259 <i>HincII</i> +13262 <i>AluI</i>
D	+10394 <i>DdeI</i> , +10397 <i>AluI</i> , -5176 <i>AluI</i>
E	+10394 <i>DdeI</i> , +10397 <i>AluI</i> , -7598 <i>HhaI</i>
G	+10394 <i>DdeI</i> , +10397 <i>AluI</i> , +4830 <i>HaeII</i> +4831 <i>HhaI</i>
A	+663 <i>HaeIII</i>
B1	9-bp deletion
B2	9-bp deletion, +10394 <i>DdeI</i>
F	-12406 <i>HpaI/HincII</i>
HV	-14766 <i>MseI</i>
H	-14766 <i>MseI</i> , -7025 <i>AluI</i>
V	-14766 <i>MseI</i> , -4577 <i>NlaIII</i>
U	+12308 <i>HinfI</i>
K	+12308 <i>HinfI</i> , +10394 <i>DdeI</i> , -9052 <i>HaeII</i>
J	-13704 <i>BstNI</i> , +10394 <i>DdeI</i>
T	+13366 <i>BamHI</i> , +15606 <i>AluI</i>
I	-4529 <i>HaeII</i> , +8249 <i>AvaII</i> , +16389 <i>BamHI</i> , +10032 <i>AluI</i>
W	+8249 <i>AvaII</i> , -8994 <i>HaeIII</i>
X	+14465 <i>AccI</i>

Note: Haplogroups of mtDNA types were designated according to classification proposed in [14]. Positions of polymorphic sites are indicated with respect to the Cambridge Reference Sequence of human mtDNA [13].

For comparative analysis the data on the frequencies of mtDNA haplogroups in Western Tuvinians, Buryats, Southern Altaians (Altai-Kizhi), Shorians, Khakassians, and Sojots, reported by us earlier [6], as well as the data for Mongolians [18], Kyrgyzes, Kazakhs and Uighurs [19], were used. In the figure presenting the results of phylogenetic analysis, the sample of Buryats investigated in the present study is designated Buryats-2, while the sample analyzed in our previous work [6] is designated Buryats-1.

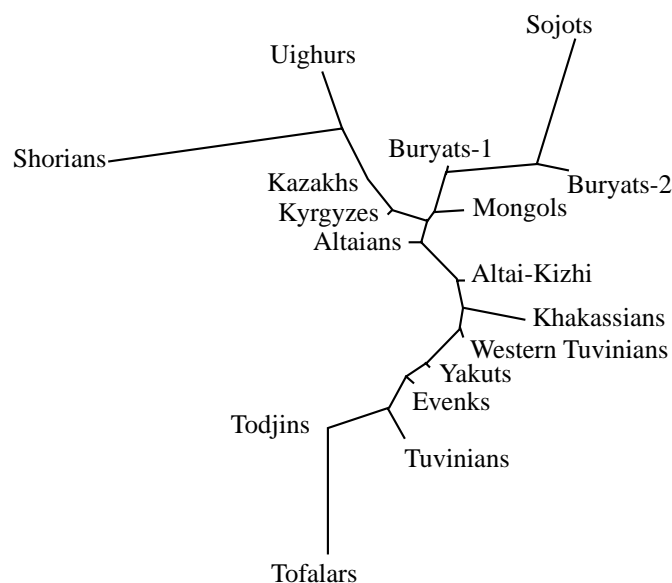
RESULTS AND DISCUSSION

The present study continues description of the mitochondrial gene pool structures in ethnic populations of Northern Asia. Earlier, using high-resolution restriction analysis of mtDNA we characterized the samples of Western Tuvinians, Buryats, Sojots, Southern Altaians (Altai-Kizhi), Khakassians and Shorians [6]. In the

present study the data on mtDNA variation in 538 individuals from seven indigenous populations inhabiting South and East Siberia and belonging to different linguistic groups of the Altaic linguistic family are presented. Altaians, Tuvinians, Tofalars, and Yakuts belong to Turkic-speaking group, while Buryats are Mongolic-speaking, and Evenks represent Tungus-Manchurian linguistic group.

The data on the prevalence of mtDNA haplogroups in Siberian populations examined are demonstrated in Table 2. As shown in the table, mitochondrial gene pools of the populations examined were characterized by different ratios between Mongoloid (M*, C, D, E/G, G, A, B, and F) and Caucasoid (H, HV, I, J, K, T, U, and X) mtDNA lineages. All the populations studied carried the marked Mongoloid component, maximum frequency of which was observed in Evenks (92.4%) and Buryats (90.1%). More than the half of Mongoloid mtDNA lineages in each population belongs to different haplogroups of macrohaplogroup M, and haplogroups C and D of this macrohaplogroup are characterized by maximum prevalence. Maximum frequency of C (62.1%) mtDNA lineages was observed in Tofalars. Tuvinians, Todjins, Evenks, and Yakuts are also characterized by high frequencies of this haplogroup (more than 40%). MtDNA types belonging to haplogroup D were found in all populations except Tofalars. Moreover, the frequencies of D mtDNA in Buryats, Evenks, and Yakuts were about 30%. A specific feature of mitochondrial gene pool in Buryats is the high frequency (14.3%) of mtDNA lineages characterized by the *HhaI* site loss at nucleotide position 7598 (haplogroup E-determining polymorphism) and the *HhaI* site gain at nucleotide position 4830 (haplogroup G-determining polymorphism). Such mtDNA types were assigned to haplogroup E/G. Note that E/G mtDNA variants were observed in all the populations tested (except Todjins) with the average frequency of 2%. In turn, in Todjins the frequency of G mtDNA types was the highest among all populations examined (18.8%). Similarly high frequencies of these mtDNA types were previously observed only among Koryaks and Itel'men [20]. Note also that the gene pools of all populations analyzed contained distinct mtDNA lineages that did not belong to the canonical haplogroups of macrohaplogroup M. These lineages may belong to haplogroup Z, not detectable by means of restriction analysis, and also to other subhaplogroups of macrohaplogroup M distributed in more southern regions.

MtDNA types belonging to haplogroups A, B, and F from macrohaplogroup R and distributed predominantly among Mongoloid populations were found in all populations examined with the frequencies not higher than 10%. Haplogroup B in the populations studied was represented by two subhaplogroups, of which subhaplogroup B1 was most common. Note that B1 mtDNA types with almost equal frequencies are distributed both in the south and in the east of Siberia. The gene pools of Altaians and Buryats also contained B2 mtDNA lin-



Unrooted NJ tree illustrating phylogenetic relationships between ethnic populations of South and East Siberia, and Middle and Central Asia. Populations are designated as it is described in Materials and Methods.

eages, characterized by the presence of the 9-bp deletion in region V and the +10394 *DdeI* variant. These mtDNA types were earlier described only in Altai-Kizhi and Sojots from Siberia [6], and also in Central Asian populations (Mongols [18] and Koreans [21]).

Caucasoid component in the gene pools of the ethnic populations of South and East Siberia is represented

by haplogroups HV, H, U, K, J, and T from macrohaplogroup R, and also by phylogenetically related haplogroups I and X from mitochondrial macrohaplogroup N. Maximum frequencies of Caucasoid mtDNAs were typical of Tofalars (20.7%) and Yakuts (14.5%), while Altaians displayed maximum diversity regarding the number of Caucasoid mtDNA haplogroups revealed

Table 2. Prevalence (in %) of mtDNA haplogroups and genetic diversity of South and East Siberian ethnic groups

MtDNA haplogroup	Tofalars (N = 58)	Altaians (N = 110)	Tuvinians (N = 90)	Todjins (N = 48)	Buryats (N = 91)	Evenks (N = 79)	Yakuts (N = 62)
C	62.08	19.09	47.78	47.91	28.56	48.10	41.94
D	0	15.45	17.79	4.17	32.96	26.58	27.42
E/G	1.72	0.91	2.22	0	14.28	2.53	1.61
G	0	0.91	4.44	18.75	0	2.53	0
M*	5.17	11.82	1.11	4.17	4.40	2.53	1.61
A	5.17	0	1.11	4.17	2.20	3.80	0
B1	3.45	2.73	7.78	4.17	4.40	5.06	4.85
B2	0	0.91	0	0	2.20	0	0
F	0	8.18	2.22	2.08	1.10	1.28	1.61
HV	0	0	0	0	0	0	1.61
H	6.90	6.36	1.11	2.08	2.20	2.53	6.45
U	0	16.36	3.33	6.25	1.10	0	0
K	0	0	0	0	0	2.53	0
J	8.62	3.64	5.56	0	2.20	0	6.45
T	5.17	0.91	1.11	0	1.10	0	0
I	0	1.82	0	0	1.10	0	0
X	0	2.73	0	0	0	0	0
“Other”	1.72	8.18	4.44	6.25	2.20	2.53	6.45
<i>h</i>	0.603	0.886	0.733	0.735	0.789	0.699	0.745

Note: Diversity of mtDNA types (*h*) was calculated as described in Materials and Methods; *N*, the sample size.

Table 3. Genetic differences between ethnic populations of South and East Siberia

Populations	1	2	3	4	5	6	7
1. Evenks		26.66	91.42***	66.31***	13.93	35.49***	31.84**
2. Yakuts	0.904		60.93***	54.86***	40.74***	55.30***	28.26*
3. Altaians	0.751	0.808		105.78***	77.29***	57.89***	71.29***
4. Tofalar	0.752	0.771	0.652		72.49***	53.19***	77.76***
5. Tuvinians	0.959	0.861	0.805	0.743		27.34	47.08***
6. Todjins	0.851	0.744	0.784	0.747	0.891		72.24***
7. Buryats	0.906	0.904	0.821	0.726	0.870	0.713	

Note: Above diagonal, identity indices I ; below diagonal, similarity indices r [15]. * $P < 0.05$. ** $P < 0.01$. *** $P < 0.005$.

(six out of eight). As it was already mentioned [6], the main impact to the Caucasoid component of Siberian populations was made by mtDNA types of haplogroups U, H, and J, with haplogroup H present in the gene pools of all ethnic haplogroups examined. In the populations of South and East Siberia haplogroups HV, K, I, and X are considered as rare, since they are observed only occasionally in different populations (Table 2).

The presence of X mtDNA types in the gene pool of Altaians deserves special interest. Earlier it was shown that haplogroup X was characterized by mosaic distribution. On the one hand, these mtDNAs were found in the populations of Europe, Caucasus, and Western Asia, and on the other hand, in the populations of North American Indians.

Comprehensive phylogenetic analysis of the X mtDNAs control region sequences has demonstrated the common ancestry of Caucasoid and American X mtDNA lineages. The time of their divergence was estimated as 12 000 to 36 000 years [22]. Thus, according to modern ideas, haplogroup X is one of the major founding mitochondrial haplogroups (A, B, C, and D), which formed the gene pools of Native Americans. It should be noted in this respect, that the absence of haplogroup X in the populations of Eastern Asia has long been a major obstacle to the reconstruction of the process of peopling of America. However, with the revealing of haplogroup X lineages in the populations of Northern and Southern Altaians [23], it can be suggested that the presence of this rare mtDNA haplogroup in South Siberian populations is the evidence of the involvement of these populations in the colonization of America, since, according to the data of phylogenetic analysis, X mtDNA variants found in Altaians occupy an intermediate position between Caucasoid and North American X lineages [23].

It should be noted that Altaians displayed maximum levels of genetic diversity of all seven populations examined ($h = 0.886$). Minimum diversity values were observed in Tofalars ($h = 0.603$) and Western Evenks ($h = 0.699$). In case of Tofalars, low genetic diversity can be explained by their low population number, isolation, and the effect of inbreeding. It has been estab-

lished that population of the settlement of Alygdzher, examined in the present study, was formed from small, rather isolated inbred tribe groups. The founders of the population of this settlement were Tofalars from only two tribes, Chogda and Kara-Chogda (according to [24] 38 and 46.6% of the individuals examined identified themselves as the members of these tribes, respectively). The extremely high (62%) frequency of haplogroup C in this population evidences that the gene pool of Tofalars was founded by few mitochondrial lineages (see Table 2).

Table 3 presents the data on genetic differences between the populations of South and East Siberia. Despite the fact that the gene pools of the populations studied were represented by similar sets of mtDNA haplogroups, statistically significant differences ($P < 0.05$) were found between almost all population pairs compared. The pairs Evenks–Yakuts ($P = 0.063$), Evenks–Tuvinians ($P < 0.1$), and Tuvinians–Todjins ($P = 0.053$) were the exceptions. Thus, taking into consideration the data on interpopulation differences between the ethnic groups of Altai–Sayan region [6], it can be concluded that mitochondrial gene pools of Siberians can be considered as highly differentiated systems. The existence of genetic differentiation of the ethnic groups with respect to mtDNA types is consistent with the conclusion on the prevalence of any type of differentiation over integration in populations of Northern Asia, which is based on the data on biochemical polymorphism of the indigenous population of this region [25].

The results of phylogenetic analysis of mtDNA variation in 17 populations from Middle and Central Asia, and South and East Siberia are presented in the figure. The NJ tree shows that the populations examined differentiate into two groups. The first group comprises the majority of Siberian populations (Altaians, Khakasians, Tuvinians, Yakuts, Evenks, Todjins, and Tofalars), while the second includes only Buryats and Sojots, which cluster together with Mongols. The populations of Central Asia, represented by Uighurs, Kazakhs, and Kyrgyzes, also form a separate cluster. Generally, the distribution of the populations on the phylogenetic tree pointed to the presence of certain geographic

trends, which probably reflected the existing ethnic relationships and the gene migrations associated with them.

The marginal phylogenetic position of eastern Sayan populations of Tofalars, Shorians, and Tuvinians is of special interest. Similar differentiation of eastern Sayan ethnic groups was described earlier based on protein polymorphism data. Rychkov *et al.* [24] suggested that the formation of the contemporary population genetic pattern of Eastern Sayans occurred with the participation of three genetically isolated groups, the traces of which are preserved in Tofalars, Todjins, and Tuvinians. This initial heterogeneity was further enhanced by gene drift under conditions of population isolation and inbreeding caused by the existence of tribe groups. At the same time, the direction of gene migrations from Tuva through Todja to Tofalaria is confirmed by the gradient in the changes of the anthropological type from Central Asian in Tuvinians, through mixed in Todjins, to Katangsk variant of Baikal type in Tofalars, described by anthropologists. Moreover, linguistic studies showed that Samoyedic language in Tofalaria was replaced by Turkic language, which penetrated from the south through the East Sayan Ridge [24].

The results of phylogenetic analysis of mtDNA variation in populations from South and East Siberia also show some other examples of the linguistic replacement, which, probably, was not accompanied by the change of the population and the disruption of genetic succession before and after the period of the language change. These are Turkic-speaking Sojots, which are genetically close to Mongolic-speaking Buryats and Mongols, and also Western Evenks, which belong to Tungus-Manchurian linguistic group, but at the same time are genetically close to their eastern neighbors, Yakuts. Genetic closeness of Western Evenks to Tuvinians and Todjins suggest possible involvement of Central Asian Turkic components into their ethnogeny. This suggestion can be confirmed, for example, by the presence of mitochondrial haplogroup B in the gene pool of Western Evenks and Yakuts. This haplogroup is prevalent in the Central Asian and Southern Siberian populations, but is absent in Northwestern Asian Evenks, belonging to Tungus-Manchurian linguistic group [26, 27].

Some discrepancies in the phylogenetic position of Buryats, Tuvinians, Yakuts, and Western Evenks are also observed upon comparison with anthropological data. According to the classification of anthropological types of the Siberian population presented by Alekseev and Hohman [1], Buryats, Tuvinians, Yakuts, and partly Tofalars are characterized by the Central Asian anthropological type, while Evenks belong to the Baikal type. At the same time, high genetic similarity revealed between Western Evenks and Yakuts by means of mtDNA analysis suggest that their gene pools were formed on the basis of very similar maternal lineages. Moreover, closeness of Yakuts and Evenks naturally follows from their ethnogeny, since considerable part of

Evenks was swamped by Yakuts at their expansion throughout Siberia. Evenks, in turn, have accumulated substantial part of paleo-Siberian populations. Nevertheless, at the present stage of investigation we failed to reveal any substantial genetic difference between Evenks/Yakuts and Altai–Sayan populations. The presence of such differences was expected based on the hypothesis on the presence of paleo-Siberian component in the gene pool of Evenks.

Thus, analysis of mtDNA variation in populations of South and East Siberia and adjacent territories shows that the gene pools of Siberian populations were developed on a rather heterogeneous genetic basis. This heterogeneity can result from the diversity of Mongoloid components, both introduced at different times from Middle and Eastern Asia, and autochthonous, substantially developed in the gene pools of Siberian paleopopulations beginning from the Paleolithic, rather than from the presence of Caucasoid component, which in the populations studied was expressed at different levels. Comprehensive phylogenetic analysis of mitochondrial lineage sequences determined in the populations of Northern Asia will make it possible to elucidate the origins of all genetic components, and also to describe the patterns of genetic processes in the populations of Northern Asia.

ACKNOWLEDGMENTS

This work was supported by the Russian Foundation for Basic Research (grant nos. 99-06-80448, 00-06-80374, and 01-06-06026) and the Russian State Program “Frontiers in Genetics” (grant no. 99-04-03).

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