Mitochondrial DNA Polymorphism in Populations of Aboriginal Residents of the Far East

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Abstract—An analysis of mtDNA polymorphism in eight populations of aboriginal residents (\(N = 519\)) of the Far East has been performed. The majority of haplogroups revealed in the examined groups were of East Eurasian origin. Haplogroup D was revealed in seven populations and its frequency varied from 2.8% in Koryaks to 28.3% and 28.9% in Nanaians and Evenks, respectively. Chukchi and Koryak populations, which belong to the same language family, exhibited haplogroup G, which has the same motive and indicates the genetic kinship of both populations. The presence of East Eurasian haplogroups A and D with a strong predominance of haplogroup A in Chukchi indicates the closer relationship of this population both with Asian and Canadian Eskimos and northern Atapasks on the other side of Bering Strait. The high level of genetic variability was revealed in populations belonging to the Tungus-Manjur group. The high frequency of east Eurasian haplogroups in Nanaians could result from close historical associations with Siberian Evenks.

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INTRODUCTION

Small aboriginal populations of northern Eurasia that have lived under conditions of relative isolation significantly longer than other continental ethnic groups are a unique model for elucidating the evolutionary mechanisms of formation of the gene pool in local populations and for assessing the factors responsible for their mutual differentiation. Modern types of aboriginal populations have formed as a result of their complex history, which mirrored both various stages of ethnic history of separate nations, their interrelationships, merging, moving and the effect of the environment. In this respect, populations of the Far East are of significant interest as this region thus far remained unexplored in respect to anthropologic types of ancient populations, although undoubtedly northern Mongoloids have resided in this area for a long time. However, different populations from southeastern Asia might enter this region long time ago. The modern population of the Amur River anthropologically demonstrates high variability, which corresponds to the complex ethnical relationships and intertribal links observed in the nations in this area.

At present, we have a fairly large number of publications that explore mtDNA in populations of the Far East, including Itel’men, Koryaks, Chukchi, Evenks, Eskimos, Yukaghirs, Chuvance, Negidal’ts, Nivkhs, Oroks, Tubalars, Udeges, and Ulchi [1—9]. Although the populations examined here have been considered previously, our study still is very important. Considering the diversity of north Asia populations and the wide area of their residence, as well as their small numbers, the increase in quantity of the examined samples including samples from other locations increases the statistical significance of the obtained results.

In the present study, we describe the polymorphism of mitochondrial DNA in eight populations of aboriginal residents of the Far East dwelling in the places with high population density (Chukchi, Koryaks, Asian and Canadian Eskimos, Nanaians, Nivkhs, Evenks, and Yukaghirs).

MATERIALS AND METHODS

Sample Collection and Examined Populations

Blood samples were collected during expeditions in 1982—1994. Blood was collected from volunteers (adults and children over the age of 10) from the elbow vein under sterile conditions. The informed consent was obtained from each subject.

Chukchi and Koryaks. Chukchi and Koryaks belong to the group of northeastern paleoasians, which also include Itel’men. A close relationship exists between
Chukchi and Koryaks with regard to both language and various fields of physical and spiritual culture [10]. The sample of unrelated (according to the maternal line) Chukchi consisted of 159 subjects (Kanchalan, Ust’-Belaya, Anadyrsk District, Chukotsk Region). The sample of Koryaks had 69 subjects from Ossora village, Karaginsk District, Koryak Region. The ethnicity of individuals was identified by a questionnaire and an additional cross questionnaire with identification of parental ethnicity at least up to the third ancestor. To identify mother lines, a thorough genealogical analysis of the collected material were used for up to three to six ancestors.

Nanaians. Nanaians represent the most numerous population among Tungus-Manjur populations of Amur region. The majority of Nanaians are located along both shores of the Amur River down from the mouth of Ussuri and to the Kargi village, as well as along the tributaries and lakes of Amur system. Nanai language belongs to Manjur subgroup of Tungus-Manjur language group [10]. The Nanaian sample consisted of 86 subjects, including residents of the Khabarovsk Region.

Nivkhs. Nivkhs live in the low current of the Amur River and adjacent districts, as well as at Sakhalin. They strongly differ in language from their neighbors, who speak the languages of the Tungus-Manjur group and are related to Paleoasiats. However, their language differs from that of the Tungus-Manjur group. Nivkhs form local race complex of Amur-Sakhalin anthropological type. It could be suggested that this anthropological type was more common and that the ancient Paleoasian population of the low Amur basin belonged this type before the arrival of Tungus-speaking groups. The most direct descendants of this ancient population were Nivkhs, whose language represents a fragment of ancient languages of the Amur River area [10]. The Nivkh sample included 17 subjects from the Nogliki village, Nogliinsk District, Sakhalin Region.

Eskimos. Eskimos reside at the shore of Chukotsk Peninsula from Bering Strait at the North and up to Cross Bay at the west, as well as at Wrangel Island. The majority of Eskimos are located along the Arctic shore of America from Alaska at the west up to Labrador at the east as well as in Greenland. Eskimo language belongs to the Yupik subgroup of the Eskimo group of Eskimo-Aleut languages. The Eskimo are direct descendants of ancient culture that resided on the shores of the Bering Sea starting from the end of the first millennium before Christ [10]. The Asian Eskimo samples (N = 40) were collected in the Lorino village, Chukotsk District, Chukotsk Region; village New Chaplino and Sireniki, Providensk District, Chukotsk Region; and Canadian Eskimo (N = 44) from the Iglulik village, Canada.

Evenks. Evenks are located on the large territory from the left shore of Yenisei River at the west and up to Okhotsk Sea at the east. The southern border goes along the left shore of the Amur and Angara Rivers. They speak the Evenk language, which belongs to the Tungus-Manjur group of the Altai language family. Evenks derived from the aboriginal population of eastern Siberia with Tungus tribes, which arrived from the Lake Baikal area [10]. The Evenk sample (N = 38) included residents of the Tyndinsky District of the Amur Region.

Yukaghirs. This is one of the most ancient population of northeastern Asia, which also was involved in formation of other paleoasian populations. They previously resided on a large territory from Taymyr to Anadyr’ and from the Lake Baikal area to Arctic Ocean. Pre-Yukaghir culture was dated by archeologists since second millennium before Christ. Yukaghir language belongs to the Yukaghir group of Ural-Yukaghir language family [10]. The sample of Yukaghirs consisted of 66 subjects from the Nelemnoe village, Upper Kolyma District, Sakha Republic.

Molecular Genetic and Statistical Methods
Total DNA was isolated from lymphocytes of peripheral blood by standard phenol-chloroform method [11].

We conducted an analysis of the nucleotide sequence at hypervariable segment I (HVSI) in 519 members of the above-mentioned ethnic groups. HVSI fragment mtDNA, which, according to Cambridge mtDNA nucleotide sequence [12], corresponds to 15898–16402 bp, and amplified it using PCR protocol with specific oligonucleotide primers [13]. The sequencing of the nucleotide sequence of hypervariable segment I (HVSI) of control region mtDNA was conducted using an ABI 377 DNA sequencing machine (Perkin Elmer) with a DYEnamic™ ET Terminator cycle sequencing premix kit (Amersham Pharmacia Biotech). Mutations were revealed by comparison with the Cambridge sequence [12]. Haplogroups were also identified by analyzing each group of specific diagnostic restriction sites (RFLP) [2, 14–19].

Multidimensional scaling based on the genotype frequencies was conducted using XLStat software (Addinsoft, Inc., 2011).

Genetic variability h was assessed as follows:

$$h = \frac{n}{(n-1)} \left(1 - \sum_{i=1}^{k} p_i^2\right),$$

where n is total number of sequences, k, is the number of various haplotypes, and $p_i$ is the haplotype frequency.

Genetic segregation between different populations and its statistical significance were assessed using $F_{ST}$ (HG and HVSI) value. The statistical significance of inter-population differences according to haplogroup frequency was estimated using an exact test of population differentiation (the pace numbers of Markov