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## Mitochondrial DNA Variation in Two Russian Populations from Novgorod Oblast

A. V. Lunkina, G. A. Denisova, M. V. Derenko, and B. A. Malyarchuk

*Institute of Biological Problems of the North, Russian Academy of Sciences, Magadan, 685000 Russia;*

*fax: (4-13-22) 34-463; e-mail: malyar@ibpn.kolyma.ru*

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**Abstract**—Mitochondrial DNA (mtDNA) polymorphism was examined in two Russian populations of Novgorod oblast, from the city of Velikii Novgorod ( $n = 81$ ), and the settlement of Volot ( $n = 79$ ). This analysis showed that the mitochondrial gene pool of Russians examined was represented by the mtDNA types belonging to 20 haplogroups and subhaplogroups distributed predominantly among the European populations. Haplogroups typical of the indigenous populations of Asia were found in the population sample from Velikii Novgorod with the average frequency of 3.7% (haplogroups A, Z, and D5), and with the frequency of 6.3% (haplogroups Z, D, and M\*) in the Volot population. It was demonstrated that the frequency of the mitochondrial lineages combination, D5, Z, U5b-16144, and U8, typical of the Finnish-speaking populations of North-eastern Europe, was somewhat higher in the urban population (7.4%) compared to rural one (3.8%). The problem of genetic differentiation of Russians from Eastern Europe inferred from mtDNA data, is discussed.

### INTRODUCTION

Northwestern regions of the European part of Russia, including Novgorod oblast, form the original area of the eastern Slavic tribes [1]. According to anthropological and ethnographic data, these regions were colonized in the early Middle Ages predominantly by the tribes of Slovians from Novgorod and Krivichi from Pskov [2, 3]. It is suggested that the formation of the eastern Slavic population of Novgorod oblast, as well as of the other parts of the Northwestern region, was accompanied by the intense interethnic contacts with local Finno-Ugric tribes, including Vod' and Izhora, and northward, with the ancestors of Komi-Zyryans [1]. According to anthropological data, the Russian population from the northwestern and northern regions of the European Russia belongs to the group of European populations characterized by the features of the northern branch of Caucasoid race. For this reason, in respect of their racial and genetic features, these populations in Eastern Europe appear to be close to Western Estonians and Latvians, and to some extent, to Karelians and Northern Komis [4]. Application of integral cartographic analysis to anthropological investigations showed that at the background of the features of different pre-Slavic groups of Eastern Europe, modern Russians exhibit some features typical of proper Slavs [5]. One of the latter elements in the anthropological composition of Russians is represented by the Pskov nucleus, which is similar in origin to the annalistic Krivichi from Pskov. The appearance of this nucleus dates back to the early stages of the development of the ancient Russian population (before 9th century C.E.). Cartographic analysis of anthropological data, however, shows that the modern Russian population from

the Northwestern region carries only the traces of the Novgorod–Sloven nuclear structure. Specific features of this structure may have existed in the past, but then were lost, bringing the physical appearance of the members of the Novgorod population close to the general Russian anthropological type [5].

The data on biochemical polymorphism among Russian population from Pskov oblast and Novgorod oblast show that, in general, the population distribution pattern of the polymorphisms observed coincided with the “average Russian” genetic marker distribution pattern. At the same time, the population of Novgorod oblast (settlement of Volot) had rare polymorphic variants, for example, of esterase D [6, 7]. In wider sense, the results of gene geographic studies point to the specific features of the Russian gene pool and also to the fact that the direction of biochemical variability is somewhat consistent with the main stages of the evolution history of the Russian population, including one of the most interesting steps, the development of the northern Russian territories [8].

Mitochondrial DNA (mtDNA), a highly polymorphic genetic system, which is maternally inherited without recombination, is a powerful tool for studying population genetic history. The present study is a continuation of a series of works on the structure and diversity of the mitochondrial gene pool of the Russian population from Eastern Europe [9–11]. Here, we present the data on mtDNA polymorphism in two populations of Novgorod oblast, from the city of Velikii Novgorod and the settlement of Volot, located close to the border with Pskov oblast. According to anthropological data [1, 2], Russian population of Novgorod oblast was formed under the influence of the annalistic tribes of Novgorod

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

mtDNA Haplogroup	Markers
H	–14766 <i>Mse</i> I, –7025 <i>Alu</i> I
pre*V	–14766 <i>Mse</i> I, +15904 <i>Mse</i> I, –16297 <i>Mse</i> I
V	–4577 <i>Nla</i> III, –14766 <i>Mse</i> I, +15904 <i>Mse</i> I, –16297 <i>Mse</i> I
HV*	–14766 <i>Mse</i> I
U	+12308 <i>Hin</i> fI
K	+12308 <i>Hin</i> fI, –9052 <i>Hae</i> II, +10394 <i>Dde</i> I
J	–13704 <i>Bst</i> NI, +10394 <i>Dde</i> I
T*	+13366 <i>Bam</i> HI, +15606 <i>Alu</i> I
T1	+13366 <i>Bam</i> HI, +15606 <i>Alu</i> I, –12629 <i>Ava</i> II
N1	–12498 <i>Nla</i> III
I	–4529 <i>Hae</i> II, +8249 <i>Ava</i> II, +10032 <i>Alu</i> I, +10394 <i>Dde</i> I, –12498 <i>Nla</i> III, +16389 <i>Bam</i> HI
W	+8249 <i>Ava</i> II, –8994 <i>Hae</i> III
X	+14465 <i>Acc</i> I
C	+10394 <i>Dde</i> I, +10397 <i>Alu</i> I, –13259 <i>Hinc</i> II/+13262 <i>Alu</i> I
D	+10394 <i>Dde</i> I, +10397 <i>Alu</i> I, –5176 <i>Alu</i> I
G	+10394 <i>Dde</i> I, +10397 <i>Alu</i> I, +4830 <i>Hae</i> II/+4831 <i>Hha</i> I
E	+10394 <i>Dde</i> I, +10397 <i>Alu</i> I, –7598 <i>Hha</i> I
M*	+10394 <i>Dde</i> I, +10397 <i>Alu</i> I
A	+663 <i>Hae</i> III
B	9-bp deletion in the region V
F	–12406 <i>Hpa</i> I / <i>Hinc</i> II

Note: Haplogroups of mtDNA types were designated according to classification suggested in [12, 13]. Positions of the polymorphic restriction sites are indicated with respect to Cambridge reference sequence of human mtDNA [14].

Slovens. The effect of Pskov Krivichi in the western part of the oblast also cannot be excluded.

## MATERIALS AND METHODS

Biological material (whole blood samples) of Russian individuals was obtained from the Novgorod blood transfusion center. Two population samples were formed, representing the individuals born in Velikii Novgorod ( $n = 81$ ) and Volot ( $n = 79$ ), respectively. Judging by the questionnaire data, all individuals examined were unrelated Russians at least in two generations down the maternal lineage.

Genomic DNA for the analysis was extracted from the blood samples using standard techniques, including cell lysis by proteinase K (Sigma, United States) in the presence of 1% sodium dodecylsulphate, purification of DNA samples by use of phenol/chloroform, and precipitation of DNA with ethanol.

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 [15, 16]. 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. The mtDNA types were identified based on classification of the haplogroups of mtDNA types in the populations of Eurasia [12, 13]. Mitochondrial mtDNA haplogroups and subhaplogroups are defined as monophyletic clusters marked by certain polymorphisms. According to this classification, the haplogroups of mtDNA types are denoted by single Roman letters (excluding haplogroup HV), and subhaplogroups within the haplogroups, by digits added to the letter code of the group (Table 1). The mtDNA types that belonged to a particular haplogroup, but could not be attributed to any of known subhaplogroups, were designated by asterisks. For example, HV = H + pre-V + HV\*, where HV\* is a paraphyletic group, since, according to currently available data, it does not contain the markers for individual monophyletic groups.

In addition to the polymorphisms mentioned in Table 1, screening for the markers distinguishing haplogroup Z, subhaplogroups D5, U5b-16144, and U8 markers was conducted. To identify haplogroup Z,