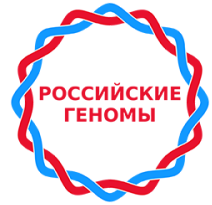


Patterns of maternal and paternal inheritance in Russian populations



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This research is a part of Russian Genome Project conducted with SPbSU and Russian Genome Consortium. Whole genome sequencing was performed for all the samples. Our research is based on these WGS data. We identified mtDNA and Y-chromosomal haplogroups (Macro-haplogroup, Major haplogroup, and Haplogroup) distribution of the 243 individuals (Fig. 1A). Our analyses demonstrate that mtDNA haplogroup composition of three populations, Yakuts, Kalmyks, and Udege was distinct from those of the remaining populations (Fig. 2A). Thus, macro-haplogroup M, which was mainly found in Asia and Siberia, is dominant in Yakuts, Kalmyks and Udege, while macro-haplogroup R, which was mainly found in Europe, is dominant in the remaining populations examined in the study, including ethnic Russians. Interestingly, Udege can be identified by a unique major haplogroup combination (M7, M8, and N9), which is only found in this ethnicity among the 14 populations analysed.

Ethnic Russians, belonging to the East Slavs ethnic group are clustered together with Komi and Seto, the Finno-Ugric peoples, as revealed with the MDS plot (Fig. 1B). Surprisingly, according to our results, Tatars, being a Turkic nation, are closely related to their geographic neighbors, Khanty and Udmurts (the Finno-Ugric peoples), rather than to the members of the same Turkic language family Yakuts and Bashkirs. Adyghe, Kalmyk, and Udege peoples attributed to Circassians, Mongolic, and Tungusic language families, respectively, show similarity between genetic and linguistic distinctness. Overall, mtDNA from non-ethnic Russians is heterogeneous compared to the homogenous ethnic Russians.

The Y-chromosome analyses shows that while subclades of R1a1 and R1b1 haplogroups are common for all ethnic Russian populations, the central and northern Russians also have a significant proportion of N1a haplogroup. Other Y-chromosomal haplogroups in Russians are less-frequent. Komi and Khants both have a typical Finno-Ugric N1a haplogroup (Fig. 2B).

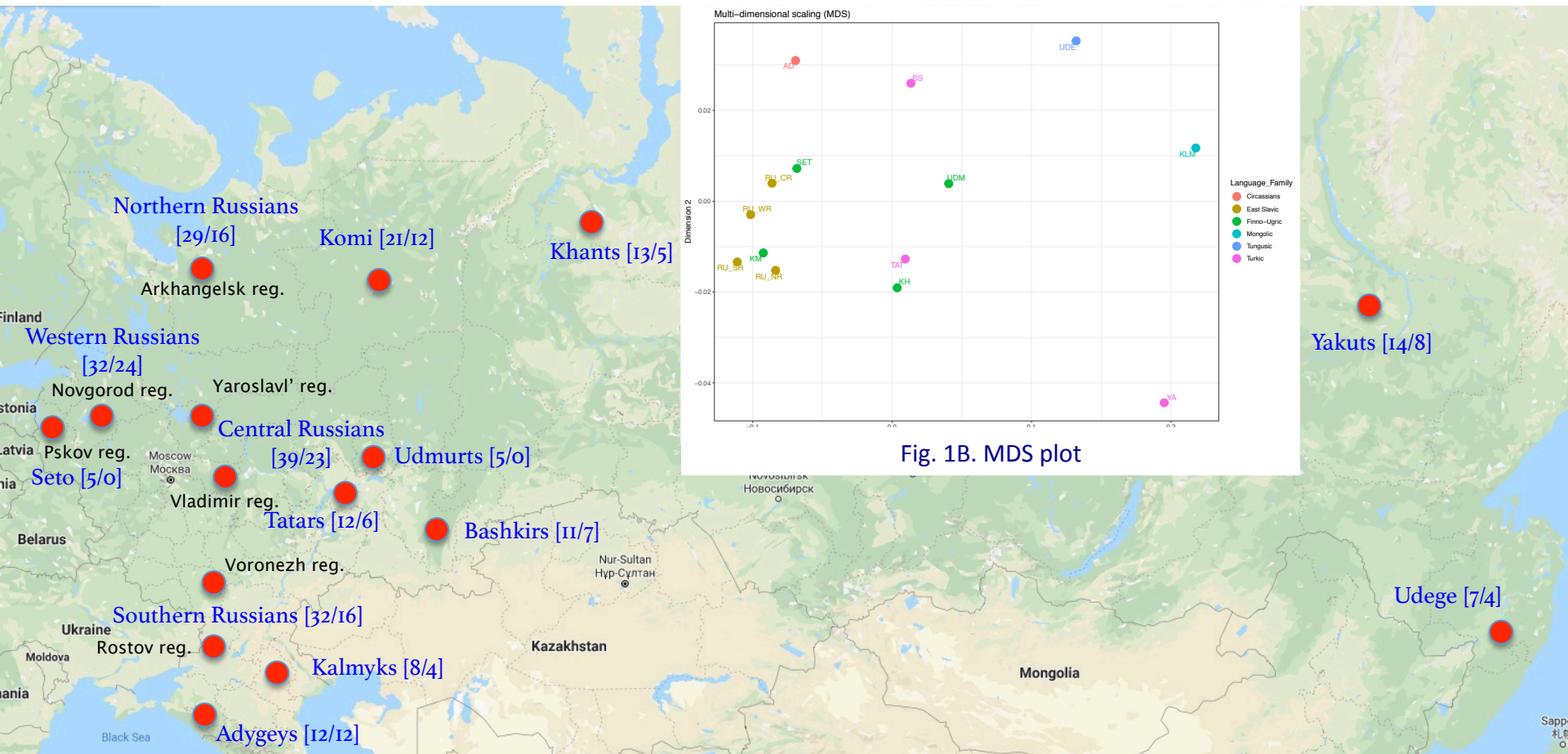


Fig. 1B. MDS plot

Fig. 1A. Investigated populations and regions of samples collection for WGS. In the square brackets numbers of sequenced: mitochondrial DNAs (before slash) and Y-chromosomes (after slash)

Adygeys people have different subclade of G2a Y-chromosomal haplogroup commonly encountered in the Caucasus. Haplogroup frequency patterns revealed in Bashkirs show equal distribution of J1, J2a, E1b and R1b1. We also noticed that Y-chromosomal haplogroups diversity in our dataset is higher in populations of ethnic Russians and Bashkirs as compared to Adyghe, Kalmyks, Komi and Udeges peoples. Khants and Yakuts shown the smallest haplogroup diversity in our dataset.

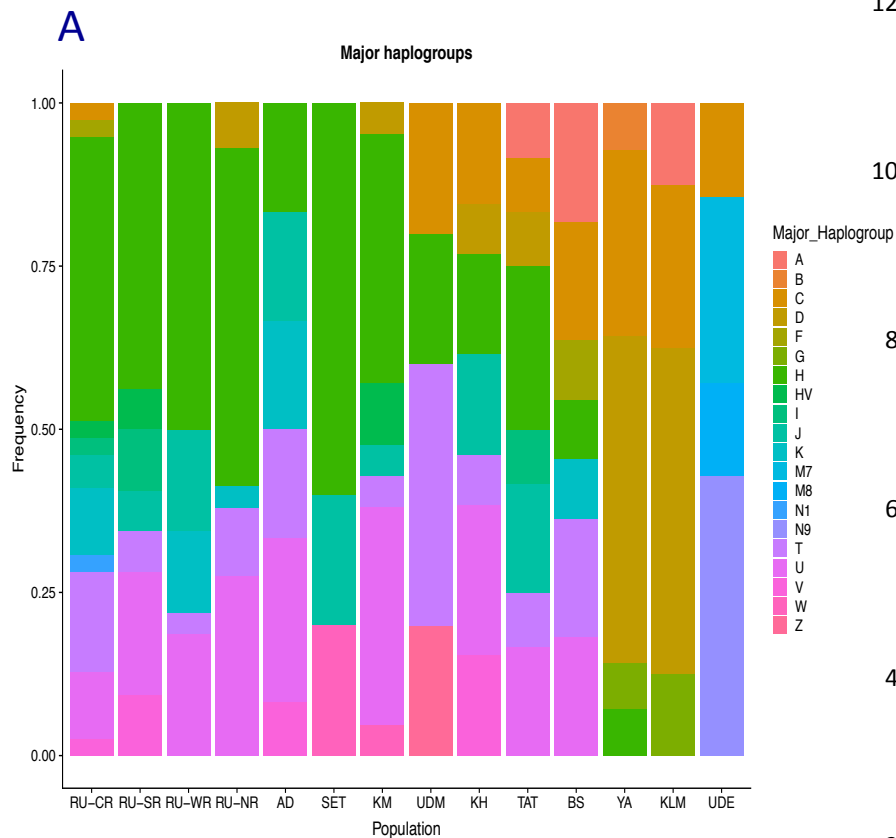
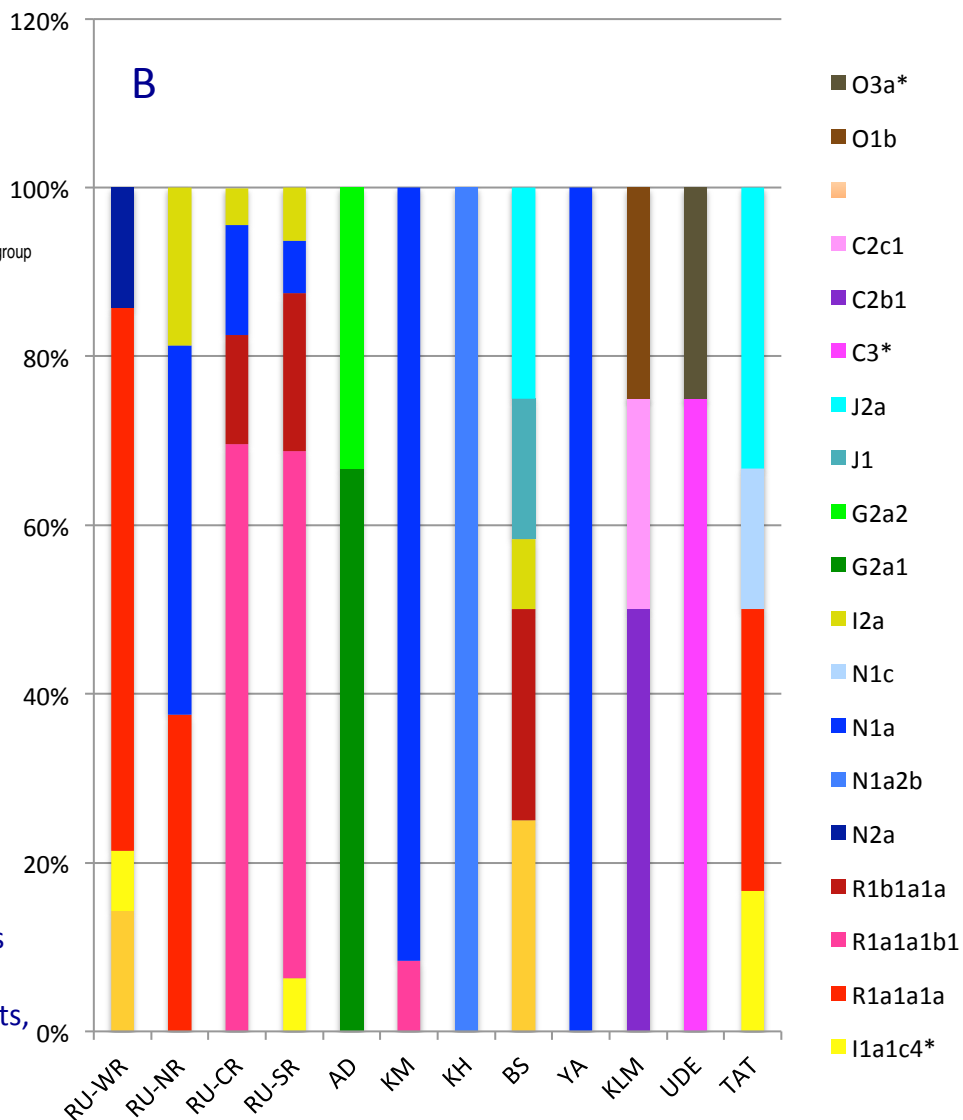


Fig. 2. Distribution of major haplogroups of mtDNA (A) and Y-chromosome (B) across investigated populations: RU – russians (CR – central, WR – western, NR – northern, SR – southern), AD – adygeys, SET – seto, KM – komi, UDM – udmurts, KH – khants, TAT – tatars, BS – bashkirs, YA – yakuts, KLM – kalmyks, UDE - udeges



In conclusion, we say that Russian Genome Project is continued and expanding the numbers of investigated populations across Russian Federation for WGS and consequently for further and deeper analysis of maternal and paternal inheritance pattern of indigenous peoples.