

# HVS1 HAPLOTYPES OF MTDNA IN THE MODERN POPULATION OF YAKUTIA IN COMPARISON WITH ITS ANCIENT POPULATION AND EURASIAN PEOPLES

*D.G. Tikhonov*<sup>1</sup>, *M.V. Golubenko*<sup>2</sup>, *T.M. Sivtseva*<sup>1</sup>, *R.N. Zakharova*<sup>1\*</sup>, *S.I. Semenov*<sup>1</sup>,  
*M.M. Okhotina*<sup>1</sup>, *R.R. Salakhov*<sup>2</sup>, *T.M. Klimova*<sup>1</sup>, *V.L. Osakovsky*<sup>1</sup>

<sup>1</sup> North-Eastern Federal University, NEFU, 27 Oyunskogo St., Yakutsk, 677013, Russia;

<sup>2</sup> Research Institute of Medical Genetics, Tomsk National Research Medical Center, SB RAS, 10 Naberezhnaya Ushaiki, Tomsk, 634050, Russia.

\* Corresponding author: prn.inst@mail.ru

**Abstract.** The study of mitochondrial DNA (mtDNA), particularly the hypervariable segment (HVS1) region, is widely used to reconstruct a population's history, structure, and origin. The origin of the Sakha people living in the north-east of Russia has been discussed for more than 300 years, but up to the present time many aspects of their ethnogenesis remain unclear. In order to clarify the migration routes of the ancient ancestors of the Sakha, we analyzed the mitotypes of 69 unrelated representatives of this ethnic group, whose belonging to the ethnic group was traced to the third generation. In the studied Sakha group, we identified 33 mitotypes, the distribution of which by haplogroups approximately coincides with the data of other studies. The results of a comparative study of Sakha mitotypes according to EMPOP data and literature sources revealed a wide distribution of the identified mtDNA HVS1 haplotypes in many populations of Eurasia. A comparison of the obtained mitotypes with the results of mtDNA sequencing of ancient samples shows that most of the mtDNA lines of modern Sakha have long been located on the territory of Yakutia. West Eurasian and East Asian mtDNA lines were incorporated into the Sakha genome at different times in different ways. The results obtained contribute to a better understanding of the routes of ancient migrations of the ancestors of the Sakha population.

**Keywords:** mitochondrial DNA, HVS1, Yakuts, Sakha, ethnogenesis.

## List of Abbreviations

mtDNA – mitochondrial DNA

HVS1 – hypervariable segment

ybp – year before present

## Introduction

The study of mitochondrial DNA (mtDNA), in particular the hypervariable segment (HVS1) region, is widely used to reconstruct the history, structure, and origin of a population. The origin of the Sakha people (Yakuts), living in the north-east of Russia, has been discussed for more than 300 years (Ides & Brand, 1967), but up to the present time many aspects of their ethnogenesis remain unclear. In the question of the origin of the modern Yakut population, the scientific concept, that assumes the migration of a small group of nomadic shepherds from the extremities of Lake Baikal in the past to the territory of Yakutia, prevails. However, it should be noted that a number of researchers believe that the process of formation of the Sakha

people took a long period of time and began as a result of a mixture of nomads of Xiongnu-Xianbei origin and «ethnic groups of aborigines whose ethnonyms are not known to science» (Alexeev, 2013; Tikhonov *et al.*, 2020).

In order to clarify the migration paths of ancient ancestors, we analyzed the mitotypes of 69 unrelated representatives of the Sakha, whose belonging to the ethnic group was traced to the third generation, and studied the matching mitotypes in the HVS1 region, found throughout the world. To determine the probable time of appearance of mitotypes in the territory of Yakutia, we also compared the obtained mitotypes with the results of mtDNA sequencing data of ancient samples excavated in the territory of Yakutia known in the literature.

## Materials and Methods

The study was carried out in accordance with the standards and the principles of the Helsinki

Declaration of 1975 and its revised version from 2000. Prior to inclusion in the study, written informed consent was obtained from all participants, and the study itself was approved by the Local Ethics Committee of the Medical Institute of the North-Eastern Federal University (Protocol dated June 12, 2021).

Analysis of mtDNA haplotypes was carried out using Sanger sequencing at the Research Institute of Medical Genetics of the Tomsk Research Center of the Siberian Branch of the Russian Academy of Sciences. In total, sequencing of 80 samples of the population of Yakutia from different districts (uluses) was carried out. Of these, 11 samples were excluded due the clarification of family ties on the maternal side and origin from mixed interethnic families as a result of studying the pedigree. In total, the results of HVS1 sequencing were analyzed in 69 individuals whose belonging to the Sakha ethnic group was traced to the third generation. The analyzed group consisted of 32 men and 37 women, originating from 17 uluses of the Republic of Sakha (Yakutia) and the city of Yakutsk by place of birth.

The search for matching mitotypes was carried out in the EMPOP database (Parson and

Dür, 2007) and according to published sources. The haplogroup was determined manually according to the HVS1 mitotypes, in accordance with the PhyloTree 17 nomenclature (February 18, 2016). Mitotypes were considered matched if the mtDNA sequences were completely identical to the HVS1 sequence and their haplogroup determined from the HVS1 mitotypes. In addition, in order to determine the probable time of the appearance of mitotypes in the territory of Yakutia, we compared the obtained mitotypes with the results of mtDNA sequencing data of ancient samples excavated in the territory of Yakutia known in the literature.

### Results

In the studied Sakha group, 33 mitotypes were identified, which are presented in Table 1. The most common haplogroups in Yakut samples were C with frequency 45.0%, the second place is occupied by haplogroup D – 32.0%. The most frequent subclades of mtDNA haplogroup C and D were: D5a2a – 20.3% and C (C4b) – 18.8%.

Comparison these data with other mtDNA studies of the Yakuts shows the similarity of the distribution of identified haplogroups (Table 2).

Table 1

**HVS1 mitotypes and mtDNA haplogroups  
in the examined representatives of the Sakha population**

No	Profile HVS1 (items 16024-16365)	Number of observations	Haplogroup
1	179-223-290-311-319-362*	1	A
2	066-223-242-290-319	1	A8a1
3	189-217	1	B4
4	093-129-223-298-327	1	C4a1
5	129-223-298-327	1	C4a1
6	093-129-223-327	7	C4a1a3d
7	129-150-223-298-327	1	C4a1a4a
8	223-298-327-344-357	3	C4a2a
9	171-223-298-327-344-357	3	C4a2a1
10	223-298- 327	10	C
11	189-223-298-327	1	C
12	223-298-311-327	2	C
13	093-223-261-288-298	1	C5a1
14	148-164-223-288-298-327	1	C5b1b
15	223-294-362	2	D4
16	223-319-320-362	1	D4b1

End of table. 1

No	Profile HVS1 (items 16024-16365)	Number of observations	Haplogroup
17	223-319-362	2	D
18	174-223-362	1	D4h1
19	093-192-223-232-362	1	D4o2a
20	093-223-232-362	1	D4o2a
21	092-172-182-183-189-223-266-362	13	D5a2a
22	172-189-223-266-362	1	D5a2a
23	189-232CA-249-304-311	1	F1b
24	223-227-274-278-362	1	G2a
25	223-227-278-362	1	G2a
26	189-356	1	H1
27	067-260-355	2	HV1a1
28	067-189	1	HV1b2
29	069-126-241	2	J2a2b
30	129-192-223-297	1	M7b1a1
31	145-223-295-304	1	M7c1a1b1
32	356	1	U4
33	185-223-260-274-298	1	Z
		<b>69</b>	

Note: \* The HVS1 profile of an individual is a list of positions of nucleotide substitutions (transitions), minus 16000 (for example, 145 is a transition at position 16145 of mtDNA). In the case of transversions, specific substitutions are indicated (for example, 232CA is 16232C→A).

Table 2

**Comparative analysis of the frequency of mitotypes of mtDNA haplogroups in Sakha according to the HVS data in different studies**

mtDNA haplogroups	This study	Fedorova S.A., 2008 (Fedorova, 2008)	Tarskaya L.A., 2009 (Тарская et al., 2009)	Keyser C. et al., 2015 (Keyser et al., 2015)	Pyzurev V.P. et al., 2003 (Puzyrev et al., 2003)
Total Observations, n (%)	69 (100)	423 (100)	140 (100)	164 (100)	83 (100)
East Asian haplogroups					
Total East Asian haplogroups, n (%)	62 (89,86)	383 (90,54)	116 (82,86)	151 (92,07)	78 (93,98)
A	2	8	2	4	1
B	1	5	11	3	3
C	31 (44,93)	175 (41,37)	47 (33,57)	77 (46,95)	40 (48,19)
C4b (223-298-327)	10 (14,49)	26 (6,15)	6 (4,29)	13 (7,93)	5 (6,02)
C4a1a3d	7 (10,14)	29 (6,86)	10 (7,14)	16 (9,76)	7 (8,43)
C4a2a1	3 (4,35)	23 (5,44)	4 (2,86)	11 (6,71)	3 (3,61)
D	22 (31,88)	125 (29,55)	41 (29,29)	50 (30,49)	27 (32,53)
D5a2a	13 (18,84)	47 (11,11)	17 (12,14)	14 (8,53)	15 (18,07)
F	1	19	5	6	3
G	2	22	3	–	2
R	–	2	3	4	–
Y	–	5	–	1	–
Z	1	4	–	1	–

End of table. 2

mtDNA haplogroups	This study	Fedorova S.A., 2008 (Fedorova, 2008)	Tarskaya L.A., 2009 (Тарская <i>et al.</i> , 2009)	Keyser C. <i>et al.</i> , 2015 (Keyser <i>et al.</i> , 2015)	Pyzurev V.P. <i>et al.</i> , 2003 (Puzyrev <i>et al.</i> , 2003)
East Asian haplogroups					
M	2	18	4	5	2
West Eurasian haplogroups					
Total West Eurasian haplogroups, n (%)	7 (10,14)	40 (9,46)	24 (17,14)	13 (7,93)	5 (6,02)
H и HV	4	21	3	2	2
J	2	6	7	4	1
K	–	–	1	1	–
T	–	5	7	3	–
U	1	2	1	3	2
W	–	6	5	–	–

Note: \* data are presented as number of observations n and frequency in %. The frequency is calculated for the most common haplogroups.

### Discussion

It should be noted, that the mitotype of the C4b haplogroup found in the Sakha (16223T-16298C-16327T) is widespread in the world and was already found in Yakutia in ancient times. Thus, haplogroups C4b, C4b1 with mitotype 16223T-16298C-16327T were found in the Pomazkino burial of the Middle Kolyma district with an age of ~3385 year before present (ybp), in the remains of the Kamenka triple burial of the Lower Kolyma district with an age of ~3880-3940 ybp (Keyser *et al.*, 2015) and in the Rodinka burial of the Lower Kolyma district (Kilinç *et al.*, 2018). The oldest mtDNA HVS1 polymorphism in our observations is 16223T-16319A-16362C, defined as haplogroup D. This mitotype was found in the Onnyuos burial of the Amga district with an age of ~6340 ybp (Kilinç *et al.*, 2018). Haplogroup C4a1 with the motif 16129A-16223T-16298C-16327T, coinciding with haplotype 5 (Table 1), was identified in bone remains found in the Buor Yurekh area, whose age corresponded to approximately 1080 AD (Lee *et al.*, 2018).

The results of a comparative study of Sakha mitotypes according to EMPOP data and literature sources revealed a wide

distribution of the identified HVS1 mtDNA motifs in the populations of the world, especially in Eurasia. The obtained data are reflected in Fig. 1.

The most frequent coincidences of DNA mitotypes are territorially localized in Uzbekistan (37)<sup>1</sup>, Buryatia (37), Krasnoyarsk Territory (23), Altai Republic (18), China (16), Pakistan (13), South Korea (11). It should be noted that the analyzed literary sources are not complete, so the above data are not exhaustive. In ethnic terms, the largest number of common mitotypes was with the Buryats (24), Evenks<sup>2</sup> (23), Tatars (20), Uzbeks, Karakalpaks, Kazakhs, Kirghiz and Uzbeks of Uzbekistan (37), Han (16), Tuva (15), Tubalars (12), Koreans (11), Hamnigans (9). A significant number of coincidences of the Sakha mitotypes with the Turkic peoples throughout Eurasia indicates a period of common culture of life and language, which were inherited by a certain part of the Sakha ancestors even before migration to the North, while the data of modern research cannot yet establish the exact time of these migrations.

<sup>1</sup> Number of cases in brackets.

<sup>2</sup> Evenks living outside the territory of Yakutia.



**Fig. 1.** The mtDNA mitotypes of the modern world populations, as well as of the ancient population on the territory of Yakutia, which coincide with the HVS1 mitotypes of Sakha obtained by us (provided that the mtDNA haplogroups match). The map was compiled using Google Map and the Yandex Map constructor. The list of sources used in compiling the map can be provided by the authors upon request

*Note:* \* red circles are our data; blue circles are the matching mitotypes outside of Yakutia; yellow circles are the ancient mitotypes on the territory of Yakutia

As can be seen from Fig. 1 coinciding motifs of HVS1 mtDNA Sakha were not detected only in Australia and Oceania, rare cases in Africa and South America. Of some interesting coincidences, it should be noted some Sakha mitotypes that are common with the Hazaras of Pakistan (5), Iraqis of Iraq (3), Hungarians of Transylvania (5), the indigenous population of America (2) and the population of California (5), which may be associated with the peculiarities of migration ancient and modern population.

It is interesting to find out the time of appearance of the West Eurasian mtDNA haplogroups on the territory of Yakutia. According to our data, 10.1% of the examined Yakut mtDNA samples are represented by European haplogroups, which is slightly more compared to the data obtained by other authors. In the Sakha we

found 4 haplogroups of the seven European haplogroups designated as 7 daughters of Eve by Brian Sachs (Bryan, 2001).

At present, according to the results of the International Sakha-French Archaeological Expedition (headed by Professor E. Crubézy), it is generally accepted that the West Eurasian mtDNA haplogroups J, K, H and W were present on the territory of Yakutia before the appearance of the Russian population here. It should be noted that the mtDNA haplogroup J2a2b was probably one of the most common in ancient times. So, it was found in the burial of Sytygan sihe in Khangalassky district with an age earlier than the 17th century (Ricaud *et al.*, 2006), the burial of Dyrlyaa sayulug of the Christian period in the Churapcha district and the Uorai locality in the 17th century in the Amga district (Crubézy *et al.*, 2010).

## Conclusion

Although informative, this study of a single region of the mtDNA D-loop has limitations. The true connections of the Sakha population with representatives of other modern peoples, the time of divergence and the phylogeny of mtDNA lines can be clarified by studying the whole genomes of mitochondrial DNA. Nevertheless, it is possible to draw a preliminary conclusion and outline ways for further research to elucidate the path of ancient migrations of the ancestors of the Sakha population. At present, we can assume that most of the mtDNA lines of modern Sakha have long been located on the territory of Yakutia, and the West Eurasian and

East Asian mtDNA lines were incorporated into the Sakha genome at different times in different ways. It should be noted that some of the Western Eurasian haplogroups have ancient roots, and some were acquired as a result of contacts with the European population after the 18th century.

## Acknowledgements

This work was supported by the Ministry of Science and Higher Education of the Russian Federation (Project FSRG-2020-0016 «Wide-genome studies of the gene pool of the indigenous population of the Arctic coast of Yakutia» 2020–2022 years).

## References

- ALEXEEV A.N. (2013): Rannie Kochevniki v Yakutii [Early Nomads in Yakutia]. *Bulletin of the North-Eastern Federal University M.K. Ammosova* **10**(5), 62–69.
- BRYAN S. (2001): *Seven Daughters of Eve*. London: Sorig Books, 368 pp.
- CRUBÉZY E., AMORY S., KEYSER C., BOUAKAZE C., BODNER M., GIBERT M., RÖCK A., PARSON W., ALEXEEV A. & LUDES B. (2010): Human Evolution in Siberia: From Frozen Bodies to Ancient DNA. *BMC Evolutionary Biology* **10**(1). <https://doi.org/10.1186/1471-2148-10-25>.
- FEDOROVA S.A. (2008): *Geneticheskie Portrety` Narodov Respubliki Saxa (Yakutiya): Analiz Linij Mitochondrial`noj DNK i Y-Xromosomy` [Genetic Portraits of the Peoples of the Sakha Republic (Yakutia): Analysis of Mitochondrial DNA and Y-Chromosome Lines]*. Yakutsk: Publishing House of the YSC SB RAS.
- IDES I. & BRAND A. (1967): *Zapiski o Russkom Posol`stve v Kitaj (1692-1695 Gg.) [Notes on the Russian Embassy to China (1692-1695)]*. Translation from German. Translated from German by M.I. Kazanin. Moscow: Glavnaya redakciya Vostochnoj literatury.
- KEYSER C., HOLLARD C., GONZALEZ A., FAUSSER J.L., RIVALS E., ALEXEEV A.N., RIBERON A., CRUBÉZY E. & LUDES B. (2015): The Ancient Yakuts: A Population Genetic Enigma. *Philosophical Transactions of the Royal Society B: Biological Sciences* **370**(1660). <https://doi.org/10.1098/rstb.2013.0385>.
- KILINÇ G.M., KASHUBA N., YAKA R., SÜMER A.P., YÜNCÜ E., SHERGIN D., IVANOV G.L. ET AL. (2018): Investigating Holocene Human Population History in North Asia Using Ancient Mitogenomes. *Scientific Reports* **8**(1): 1–10. <https://doi.org/10.1038/s41598-018-27325-0>.
- LEE E.J, MERRIWETHER D.A., KASPAROV A.L., KHARTANOVICH V.I., NIKOLSKIY P.A., SHIDLOVSKIY F.K., GROMOV A.V. ET AL. (2018): A Genetic Perspective of Prehistoric Hunter-Gatherers in the Siberian Arctic: Mitochondrial DNA Analysis of Human Remains from 8000years Ago. *Journal of Archaeological Science: Reports* **17**(Complete), 943–49. <https://doi.org/10.1016/j.jasrep.2016.06.001>.
- PARSON W. & DÜR A. (2007): EMPOP–A Forensic MtDNA Database. *Forensic Science International. Genetics* **1**(2), 88–92. <https://doi.org/10.1016/j.fsigen.2007.01.018>.
- PUZYREV V.P., STEPANOV V.A., GOLUBENKO M.V., PUZYREV K.V., MAXIMOVA N.R., KHARKOV V.N., SPIRIDONOVA M.G. & NOGOVITSINA A.N. (2003): MtDNA and Y-Chromosome Lineages in the Yakut Population. *Russian Journal of Genetics* **39**(7), 816–22. <https://doi.org/10.1023/A:1024761305958>.
- RICAUT F.X., KOLODESNIKOV S., KEYSER-TRACQUI C., ALEKSEEV A.N., CRUBÉZY E. & LUDES B. (2006): Molecular Genetic Analysis of 400-Year-Old Human Remains Found in Two Yakut Burial Sites. *American Journal of Physical Anthropology* **129**(1), 55–63. <https://doi.org/10.1002/ajpa.20195>.

- TARSKAYA L.A., GOGOLEV A.I., ELCHINOVA G.I, EGOROVA A.G. & LIMBORSKAYA S.A. (2009): *E`tnicheskaya Genomika Yakutov (Naroda Saxa): Geneticheskie Osobennosti i Populyacionnaya Istoriya: (Monografiya) [Ethnic Genomics of the Yakuts (Sakha People): Genetic Features and Population History: (Monograph)]*. Moskow: Nauka.
- TIKHONOV D., ZAKHAROVA A., POARCH E., NEUSTROEVA T. & SLEPTSOVA A. (2020): On the Issue of Dating Olonkho. *Siberian Research* **4**(2), 65–76. <https://doi.org/10.33384/26587270.2020.04.02.08e>.