

on the northern coast was based on industrial people, mainly people from the eastern regions of the Russian North and the Urals. According to E.A. Strogova, they were the first to reach the lower reaches of the Indigirka River, showing commercial interest in the traditional crafts of local residents, as well as in the collection of mammoth ivory [11].

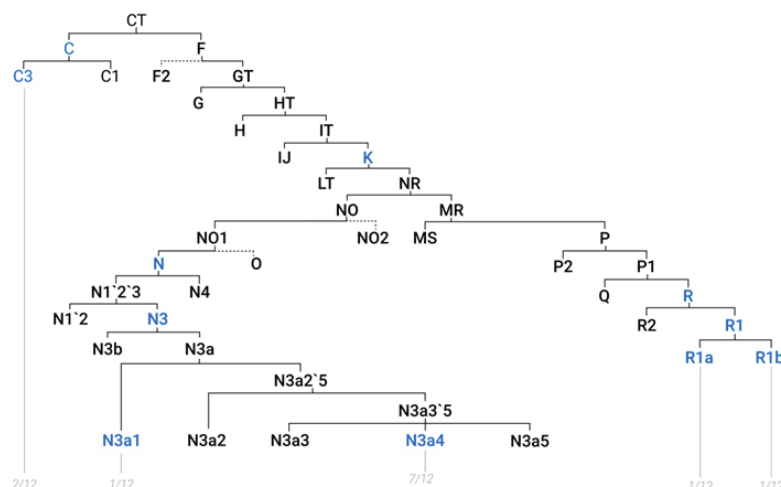
In this work, for the first time, we have analyzed the Y-chromosome lines of men from Russkoye Ustye in order to compare the obtained data with the hypotheses of the origin of the Russkoye Ustye.

**Materials and methods.** The total population of the village of Russkoye Ustye, Allaikovsky Uls of the Republic of Sakha (Yakutia), according to the current archive of the administration was 148 people in 2012-2014 [6]. For research during expeditions in 2018-2019 we collected DNA samples from 12 unrelated men, native russkoustinians, aged 7 to 66 years (average age  $42.4 \pm 17.4$  years).

All DNA samples were tested for Y-chromosome markers C-M130 and K-M9 using real-time PCR. Then, using previously published primer sequences, PCR-RFLP analysis of SNP markers of haplogroups C-M216, R-M207, N-M231, and Q-M242 was performed [26; 23; 17]. Subsequently, in accordance with the defining marker, the following markers were hierarchically typed by PCR-RFLP: R1a-M420 [27], R1b-M343 (original sequence), C1-F3393 [21], C1b-F1370 [21], C2'4-B477 (original sequence), C3b-P39 (original sequence), N2a1-P43 [19], N3-TAT/M46 (original sequence), N3a1-B211 (original sequence), N3a3-VL29 [20], N3a4-Z1936 [21], N3a5\*-F4205 (original sequence), N3a5\*-B202 [20]. The C3-M217 marker [25] was identified using Sanger sequencing. The original primer sequences were selected using FastPCR v.6.7.58 (trial) software.

The haplogroup nomenclature used is based on Karmin et al., 2015 [14], which provides the basic YCC 2002 (The Y Chromosome Consortium 2002) nomenclature with updated data from Jobling & Tyler-Smith, 2003 [21], Karafet et al., 2008 [23] and van Oven et al., 2014 [25].

**Results and discussions.** In the studied sample of Russkoye Ustye residents, five lines of the Y-chromosome



Location of samples from the village of Russkoye Ust'ye on the topology of Y-chromosome haplogroups according to Karmin, 2015 [13]

N3a4-Z1936, N3a1-B211, R1a-M420, R1b-M343 and C3-M217 were identified, which belong to three haplogroups: N3 (8/12; 66.7%), R1 (2/12; 16.6%) and C3 (2/12; 16.6%) (Table 1).

The topological position of the identified Russkoye Ustye lineages on the Y-chromosome phylogenetic tree is shown in Figure 1. Haplogroup designations correspond to the nomenclature proposed by Karmin et al., 2015 [14].

Haplogroup N3 (8/12; 66.7%) dominating in this sample is distributed throughout Northern Eurasia, from Japan to Scandinavia [20]. A more detailed analysis of the N3-lines of the russkoustinians showed that most (~58.4%) of the samples belong to the N3a4 subhaplogroup (Table 1), which was found mainly in North-Eastern Europe among northern Russians, Sami, Veps, Karelians, and Finns [20] and is not found in the gene pool of the neighboring peoples of Yakutia and Chukotka [16]. It should be noted that the N3a4 subhaplogroup and the N3a3 branch phylogenetically close to it are the most informative for the differentiation of the two so-called "poles" of the Russian gene pool, northern and southern Russians [3]. In the populations of northern Russians, the haplogroup N3a4 is associated with the ancient pre-Slavic (Finno-Ugric) component of the gene pool [3; 10].

One sample (8.3%) belongs to the N3a1 subhaplogroup, which is more

typical for the Finno-Ugric populations of the Udmurts, Mari Komi-Zyryans and Komi-Permyaks [20], neighboring with the populations of northern Russians.

The Eurasian haplogroup R1 is found in more than half of men in Europe [15]. In the studied sample of Russkoye Ustye residents, haplogroup R1 is represented by two lines: R1a (~8.3%) and R1b (~8.3%), which are widespread in the Slavic populations of Eastern Europe (Russians, Ukrainians, Belarusians, Poles) [2; 3]. In the gene pool of the Yukaghirs, Evens, Evenks and Yakuts, these lines were found in small numbers and their origin is associated with the processes of miscegenation with Russians and, possibly, other Eastern European ethnic groups that have settled the territory of Yakutia since the time of joining the Russian Empire in the 17th century [16].

The frequency of the Asian haplogroup C3 among the russkoustinians was ~16.6%. This haplogroup is one of the most widespread in East Asia [24], including Siberia [16,19]. In Yakutia, haplogroup C3 occurs with high frequencies in the populations of Yukaghirs (46%), Evens (60%), and Evenks (54%) adjacent to the Russkoye Ustye people [16].

Thus, the frequency distribution of the Y-chromosome haplogroups in the studied sample of the Russkoye Ustye residents shows the dominance of lines characteristic of European populations (83.4%), of which more than half is the subhaplogroup N3a4. In the populations of the indigenous peoples of the northeast of Eurasia (Yukaghirs, Evens, Evenks, Yakuts, Dolgans, Chukchi, Eskimos), the N3a4 subhaplogroup was not found [16; 20]. The gene pool of the indigenous peoples of Yakutia and Chukotka is characterized by an insignificant

The frequency of haplogroups (%) of the Y-chromosome in residents of the village Russkoye Ustye

Haplogroup	C3		N3		R1
	C3-M217	N3a1-B211	N3a4-Z1936	R1a-M420	R1b-M343
n	1	7	1	1	2
%	8.3%	58.4%	8.3%	8.3%	16.7%

contribution of haplogroups of Western Eurasian origin (less than 10%) [16].

If we consider the distribution of haplogroups of the Russkoye Ustye people from the point of view of their origin, then we can assume that the dominance of the lines characteristic of the populations of the northeast of Europe most likely reflects the connection with the Russian North. It is known that the northern populations of Russians are characterized by similarities with the Finno-Ugric and Baltic peoples [3], while the central and southern populations of Russians are genetically closer to other Slavic populations [2; 22; 26]. The line N3a4 (58.7%), which dominates among the Russkoye Ustye people, is considered not typical for the Slavic populations in general, however, the highest frequencies of this subhaplogroup were found in the northern Russians of the Arkhangelsk and Vologda regions and much lower frequencies – among the Novgorodians [3].

**Conclusions.** As a result of the analysis of Y-chromosome lines, 83.4% of haplogroups (N3a4, N3a1, R1a, R1b), more characteristic of Western Eurasian populations, and 16.6% of East Eurasian lines (C3) were found in residents of the village of Russkoye Ustye. The paternal lines of the inhabitants of the village of Russkoye Ustye are represented mainly by Y-chromosome haplogroups, common in the populations of northeastern Europe. The dominant subhaplogroup is N3a4 (58.7%), which is absent in the gene pool of the neighboring peoples of Yakutia and Chukotka and occurs with high frequencies in the populations of the Russian North and among the Finnish-speaking peoples of Finland and Karelia. The results obtained are more in line with the Pomor hypothesis of the origin of the russkoustinians.

**Acknowledgements.** The work was carried out within the framework of the State Assignment of the Ministry of Science and Higher Education of the Russian Federation (FSRG-2020-0016), as well as with the support of RFR grant No. 19-34-60023

## Reference

- Alekseev, A.N. Pervye russkie poseleniya XVII-XVIII vv. na severo-vostoke YAKutii [The first Russian settlements of the 17th-18th centuries. in the north-east of Yakutia]. RAS Siberian Branch Institute of Archeology and Ethnography; executive editor V.E. Larichev. Novosibirsk, 1996: 151 (In Russ.). ISBN 5-7803-0011-9.
- Balanovsky O.P. Genofond Evropy [Gene pool of Europe]. Moscow: Association of Scientific. ed. KMK, 2015: 354 (In Russ.).
- Balanovskaya EV, Agdzhoyan AT, Skhalyakho RA, et al. Genofond novgorodcev: mezhdru Severom i Yugom [Gene pool of Novgorodians: between North and South]. Genetics. 2017; 53 (11): 1338-1348 (In Russ.). DOI 10.7868/S0016675817110029.
- Zenzinov V.M. Starinnye lyudi u holodnogo okeana: Russkoe Ust'e YAKutskoj oblasti Verhoyanskogo okruga [Ancient people near the cold ocean: Russian Mouth of the Yakutsk region of the Verkhoyansk district]. Printing house P.P. Ryabushinsky, 1914: 122 (In Russ.).
- Itogi Vserossiyskoj perepisi naseleniya 2010 goda: v 11 tomah: oficial'noe izdanie / Rossijskaya Federaciya, Federal'naya sluzhba gos. statistiki; (podgot.: E.S. Nabiullina - ruk. rabochej gruppy i dr.). [Results of the All-Russian population census of 2010: in 11 volumes: official publication. Russian Federation, Federal State Service. statistics; (prepar.: E.S. Nabiullina – manuscript of the working group, etc.). Moscow: Statistics of Russia, 2012: 30 (In Russ.).] ISBN 978-5-4269-0002-8.
- Nikitina, SE. Russkie arkticheskie starozhily Respubliki Saha (YAKutii): problemy sohraneniya unikal'noj kul'tury. Russkie arkticheskie starozhily YAKutii: Sbornik nauchnyh statej [Russian Arctic old-timers of the Republic of Sakha (Yakutia): problems of preserving a unique culture. Russian Arctic old-timers of Yakutia: Collection of scientific articles. Yakutsk: IGIIPMNS, 2019: 16-33 (In Russ.).]
- Okladnikov AP, Gogolev ZV, Ashchepkov EA. Drevnij Zashiversk. Drevnerusskij zapolarnyj gorod [Ancient Zashiversk. Old Russian polar city. Moscow: Nauka, 1977: 212 (In Russ.).]
- Skvortsov EF. V pribrezhnyh tundrah YAKutii: Dnevnik astronoma Lensko Kolymskoj ekspedicii. Trudy komissii po izucheniyu YAKutii. T. 15: Lensko Kolymskaya ekspediciya 1909 goda pod nachal'stvom K.A. Vollosovicha [In the coastal tundra of Yakutia: Diary of an astronomer of the Lensko-Kolyma expedition. Proceedings of the commission for the study of Yakutia]. Lensko-Kolyma expedition of 1909 under the command of K.A. Vollosovich, 1930; 15: 1–244 (In Russ.).]
- Skvortsov EF. Russkie na Indigirke. Topografo-geodezicheskij zhurnal [Russians on the Indigirka. Topographic and geodetic journal]. 1910: 10 (In Russ.).]
- Chukhryaeva MI, Pavlova ES, Napolskih VV, et al. Sohranilis' li sledy finno-ugorskogo vliyaniya v genofonde russkogo naseleniya YARoslavskoj oblasti? Svidetel'stva Y-hromosomy [Are traces of Finno-Ugric influence preserved in the gene pool of the Russian population of the Yaroslavl region? Evidence of the Y-chromosome. Genetics. 2017; 53 (3): 378-389 (In Russ.).] DOI 10.7868/S0016675817030043.
- Strogova, E. A. Obrazovanie etnicheskoj territorii i formirovanie postoyannogo russkogo naseleniya na Severe YAKutii v XVII-XVIII vv. [Formation of an ethnic territory and the formation of a permanent Russian population in the North of Yakutia in the 17th-18th centuries]. Russkie arkticheskie starozhily YAKutii: Sbornik nauchnyh statej [Russian Arctic old settlers of Yakutia: Collection of scientific articles. Yakutsk: IGIIPMNS, 2019: 7-15 (In Russ.).]
- Chikachev AG. Russkoe serdce Arktiki [Russian heart of the Arctic. Yakutsk: Literary Fund, 2010: 478 (In Russ.).]
- Rootsi S, Zhivotovsky LA, Baldovic M, et al. A counter-clockwise northern route of the Y-chromosome haplogroup N from Southeast Asia towards Europe. Eur. J. Hum. Genet. 2007;15:204–211
- Karmin M, Saag L, Vicente M, et al. A recent bottleneck of Y chromosome diversity coincides with a global change in culture. Genome Res. 2015; 25: 459–466 (In Russ.).]
- Myres N, Rootsi M, Lin S, et al. A major Y-chromosome haplogroup R1b Holocene era founder effect in Central and Western Europe. European journal of human genetics: EJHG, 2011; 19(1), 95–101. <https://doi.org/10.1038/ejhg.2010.146>
- Fedorova SA, Reidla M, Metspalu E, et al. Autosomal and uniparental portraits of the native populations of Sakha (Yakutia): implications for the peopling of Northeast Eurasia. BMC Evol. Biol. 2013;13:127.
- Cinnioglu C, King R, Kivisild T, et al. Excavating Y-chromosome haplotype strata in Anatolia. Hum Genet. 2004 Jan;114(2):127-48. doi: 10.1007/s00439-003-1031-4.
- Zhong H, Shi H, X.B. Qi, et al. Global distribution of Y-chromosome haplogroup C reveals the prehistoric migration routes of African exodus and early settlement in East Asia. J Hum Genet; 2010; 55, 428–435. <https://doi.org/10.1038/jhg.2010.40>
- Karafet TM, Osipova LP, Gubina MA, et al. High levels of Y-chromosome differentiation among native Siberian populations and the genetic signature of a boreal hunter-gatherer way of life. Hum. Biol. 2002;74:761–789
- Illumäe AM, Reidla M, Chukhryaeva M, et al. Human Y Chromosome Haplogroup N: A Non-trivial Time-Resolved Phylogeography that Cuts across Language Families. Am J Hum Genet. 2016;99(1):163-173. doi:10.1016/j.ajhg.2016.05.025
- International Society of Genetic Genealogy. Y-DNA Haplogroup Tree 2019, Version: [15.73], Date: [11 July 2020], <http://www.isogg.org/tree/> [Date of access: 10.06.2021].
- Jobling M.A., Tyler-Smith C. The human Y chromosome: an evolutionary marker comes of age. Nat Rev Genet. 2003 Aug;4(8):598-612. doi: 10.1038/nrg1124. PMID: 12897772.
- Karafet TM, Mendez FL, Meilerman MB, et al. New binary polymorphisms reshape and increase resolution of the human Y chromosomal haplogroup tree. Genome Res. 2008;18(5):830-838. doi:10.1101/gr.7172008
- Malyarchuk B, Derenko M, Denisova G, et al. Phylogeography of the Y-chromosome haplogroup C in northern Eurasia. Annals of human genetics, 2010; 74(6), 539–546. <https://doi.org/10.1111/j.1469-1809.2010.00601.x>
- Van Oven M, Van Geystelen A, Kayser M, et al. Seeing the wood for the trees: a minimal reference phylogeny for the human Y chromosome. Hum Mutat. 2014 Feb;35(2):187-91. doi:10.1002/humu.22468.
- Underhill PA, Passarino G, Lin AA, et al. The phylogeography of Y chromosome binary haplotypes and the origins of modern human populations. Annals of Human Genetics, 2001. 65(1): 43–62. <https://doi.org/10.1046/j.1469-1809.2001.6510043.x>
- Hinds DA, Stuve LL, Nilsen GB, et al. Whole-genome patterns of common DNA variation in three human populations. Science. 2005; 307:1072–1079.