

# Analysis of Polymorphism of Uniparental Markers in Reindeer-Herding Populations: The Tozhu Tuvans of Russia and The Tsaatans Of Mongolia

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## ABSTRACT

We analyzed the data on the variability of the Y chromosome and mitochondrial DNA (mtDNA) in populations of the Tsaatans of Mongolia and the Tozhu Tuvans of Russia. The populations studied are characterized by low genetic diversity for both marker systems. The analysis of Y chromosome haplogroups in the Tsaatan and Tozhu revealed three haplogroups in the Tsaatan and seven haplogroups in the Tozhu. The composition of the haplogroups is coherent to literature data on the Tuvans, which is explained by common origin. According to the data on mitochondrial DNA variability, 12 haplogroups were determined in 46 Tozhus, of which C4b (30.43%) and F1b1b (23.91%) are major haplogroups. According to the HVS-1 (HyperVariable Segment) data, 15 haplotypes were found in the Tozhu Tuvans and the diversity coefficient of 0.8677 turned out to be much lower than among the Torghut of Mongolia (0.9857). In 23 Tsaatans, 14 haplogroups were determined; the most common of which are C4b (22.73%) and C5a1 (18.18%). According to HVS-1, 14 haplotypes were revealed in the Tsaatan, the diversity is 0.9486. The data obtained on uniparental marker systems in the Tozhus and Tsaatans are due to the isolated and inaccessible taiga region and the manifestation of the “founder effect”. The Tsaatans are less polymorphic in terms of the variety of Y chromosome haplogroups, while the Tozhus are less polymorphic in terms of mitochondrial DNA, which is probably a consequence of a high rate of endogamic marriages in the populations studied.

**Key words:** anthropology, reindeer herders, uniparental markers, haplogroups, mitochondrial DNA (mtDNA), Y chromosome

## Introduction

The Tuvans or Tuvinians (endonym Tyva; obsolete names Tannu-Tuvans, Soyots, Uriankhais) are Turkic-speaking Buddhist Lamaists; historically and culturally they are divided into western (mountain-steppe regions of western, central and southern Tuva) and eastern, or Tozhu Tuvans (mountain-taiga part of northeast and southeast Tuva). The total number of Tuvans in Russia is 263,934 people. They live mainly in the Tyva Republic,

where they represent a numerical majority of 82.04% (according to the 2010 census)<sup>1</sup>.

The Tozhu are an ethnic group of Tuvans living compactly in the Tozhu District in north-eastern Tyva. They are engaged in reindeer herding and speak the Tozhu dialect of Tuvan. The Tozhu belong to the indigenous small-numbered peoples. According to the 2010 census, their number is 1856<sup>1</sup>. Their housekeeping practices are similar to those of the Tsaatan, with many families still

living in the taiga in skin tents (chums) and maintaining reindeer herds.

The Tsaatan are a small tribe in north-western Mongolia who live off the reindeer. According to the 2010 census, their number is 282. Their name comes from the Mongolian word *tsaa*, meaning ‘reindeer’, although they call themselves *Tuba* or *Tukha*. They speak the *Tozhu* dialect of *Tuvan*. Due to the small number of Tsaatans and almost inaccessible reindeer pastures, we surveyed those who had left their camps to participate in elections and receive social benefits payable to small-numbered peoples.

The Tuvans have been thoroughly studied in different time periods under all classical anthropological programmes<sup>2–18</sup>.

The ethnic groups of the Altai-Sayan region were selected as model groups in the discussion of fundamental issues of ethnic anthropology of contact zones between representatives of the Central Asian Mongoloid type and carriers of mixed South Siberian complexes. The issue of participation of the ancient Caucasoid population in their formation and the manifestation of borders between the Turkic-speaking and Mongolian-speaking peoples of Asia were considered both by paleoanthropologists and geneticists. The presence of a Caucasoid admixture was confirmed not only by paleoanthropologists<sup>12</sup>, but also by morphologists<sup>19</sup> and geneticists<sup>14</sup>.

The last decades of the twentieth century are characterized by a rapid development of population research of highly polymorphic genetic systems in solving ethnogenetic problems. The uniparentally inherited genome components, mitochondrial DNA (mtDNA) and Y chromosome, provide individual analysis of maternal and paternal components. In recent years, mtDNA diversity was extensively studied in the ethnic groups from South Siberia<sup>20–23</sup>. It was demonstrated that mitochondrial gene pools of the populations studied were characterized by different ratios between the Mongoloid and Caucasoid mtDNA lineages<sup>21</sup>. The frequency of Caucasoid lineages declined in the direction from the south to the north, and from the west to the east, which was generally consistent with the anthropological data. Most of these lineages have south Caucasoid origin, while the others belong to east Caucasoid lineages<sup>24</sup>.

Studies of Y chromosome in ethnic groups from Altai–Sayan region also showed the presence of profound paleo-Caucasoid component in their gene pools<sup>25, 26</sup>. These data are consistent with those obtained earlier upon the analysis of mtDNA: the ethnic groups of South Siberia, compared to other indigenous populations of Siberia, possessed the most variable haplogroup composition. Furthermore, Altaians were characterized by a high frequency of haplogroup R1a1, which marked the Caucasoid gene pool component<sup>27–29</sup>.

## Materials and Methods

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. Informed consent was obtained from all individual participants included in the study. This genetic and epidemiological study was approved by the Research Ethics Committee of the Research Centre for Medical Genetics (protocol No. 5/2 dated 9 February 2015).

We studied mitochondrial DNA (mtDNA) and Y chromosome in 23 Tsaatans and 46 *Tozhu* Tuvans. The sample is representative because of very small size of total population. All respondents included into the study were interviewed about unrelated relatives up to 3 generations. DNA was extracted from peripheral blood leukocytes by the standard chlorophenol method. The haplogroup composition was studied using markers from the nonrecombining portion of Y chromosome: M9, M130, M217, M77, F6379, B469, B80, B90, L1373, M407, M217, M48, M401, M207, page07, Z93, Z95, Z2125, M343, M269, M412, Z2105, M458, M558, M478, M124, M175, M122, M119, M268, M95, P203, P201, M7, M231, P43, TAT, M128, F2930, F4205, CTS6967, B478, B525, B187 M2118, YAP, M170, M423, 12f2, M410, Page 08, M35, M174, M201, P15, M242, M25, M346. Genotyping was performed using PCR and subsequent fragment analysis by electrophoretic separation or Sanger sequencing. Mitochondrial diversity was studied using HVS-1, and then analyzed to confirm haplogroups: HVS-2, 663, 827, 1005, 1048, 1107, 1119, 1382, 1391, 1719.1G, 1719, 1811, 3010, 3206, 3552, 3759, 3816, 3826, 3970, 4491, 4646, 4715, 4833, 4850, 4904, 5021, 5093, 5108, 5153.5178, 5231, 5360, 5442, 5465, 5601, 5821, 6023, 6047, 6024, 6212, 6392, 6962, 7028, 7100, 7476, 7768, 7828, 8020, 8200, 8273, 8414, 8479, 8485, 8563, 8701, 8703, 8762, 8793, 8964, 8994, 9120, 9181, 9449, 10084, 10208, 10238, 10398, 10400, 10427, 11215, 11467, 11696, 12672, 12705, 12879, 13104, 13942, 14233, 14766, 15071, 15080, 15204, 15326, 15475, 15487, 15884.

### Statistics

Excel 2016 was used to visualize the results. For cluster analysis and multivariate scaling, the Statistica software version 12.6 was used.

We used R to perform the principal component analysis (PCA). AMOVA analysis, values of Tajima's D and Fu's Fs tests for neutrality were calculated in Arlequin 3.5.

## Results

The analysis of Y-haplogroups in the Tsaatan and *Tozhu* was previously studied during the analysis of the *Oirat* ethnogenesis<sup>30</sup>. The distribution of haplogroups is presented in Table 1 and Figure 1. The low diversity of

**TABLE 1**  
DISTRIBUTION OF Y CHROMOSOME HAPLOGROUP FREQUENCIES IN THE POPULATIONS STUDIED

Sample / haplogroup	Mongols Torghuts (N=47)	Mongols Khoshuts (N=18)	Mongols Derbets (N=40)	Mongols Tsaatans (N=23)	Tozhu Tuvans (N=46)
C3 (XC3c1, M407)	2.1	0.0	2.5	0.0	0.0
C3c-M48	4.3	11.1	2.5	0.0	0.0
C3c1a-Z40439	0.0	0.0	0.0	0.0	4.3
C3c1b-F6379	29.8	66.7	50.0	4.3	0.0
C3f1-M407	14.9	0.0	7.5	0.0	0.0
C3h-F914 (M401)	14.9	0.0	7.5	0.0	4.3
C3c1a-B469	0.0	0.0	2.5	0.0	0.0
G2a-P15	2.1	0.0	0.0	0.0	0.0
J2a-M410	4.3	5.6	2.5	0.0	0.0
N2a1-B478	2.1	0.0	0.0	0.0	13.0
N2a1-B525	2.1	0.0	0.0	0.0	0.0
N3a5a-vF4205	10.6	5.6	12.5	52.2	15.2
N3b-B187	0.0	0.0	0.0	0.0	2.2
O-M175	2.1	0.0	0.0	0.0	0.0
O2a2-P201	2.1	5.6	7.5	0.0	0.0
O3`7-M122	4.3	0.0	0.0	0.0	0.0
Q-M242	0.0	5.6	0.0	0.0	0.0
Q2a-M25	0.0	0.0	0.0	43.5	50.0
R1a1c-M558	0.0	0.0	0.0	0.0	2.2
R1a2-Z2125	2.1	0.0	0.0	0.0	0.0
R1a2-Z93	2.1	0.0	0.0	0.0	8.7
R2a-M124	0.0	0.0	5.0	0.0	0.0

haplogroups in the populations studied is explained by the geographical isolation of these groups. The “founder effect” is clearly represented in the Tsaatan population, since the genealogy is traced along two male lineages, haplogroups N3a5-F4205 (52.2%) and Q1a1b-M25 (43.5%) constitute the main diversity. According to the analysis of marriage records, the Tsaatans show a high endogamy rate since marriage relationships with Tozhu Tuvans ceased after

the state border between Russia and Mongolia had been established. In Tozhu, the most common haplogroup is Q1a1b-25 (50.0%), the other haplogroups are represented with the following frequencies: N3a5-F4205 (15.2%), N2a1-B478 (13.0%), R1a2-Z93 (8.7%), C2b1b1-M77 (4.3%), C3-M217 (4.3%), N3b-B187 (2.2%).

According to the data on mitochondrial DNA variability, in 46 Tozhus, 12 haplogroups were determined (C4a1a,

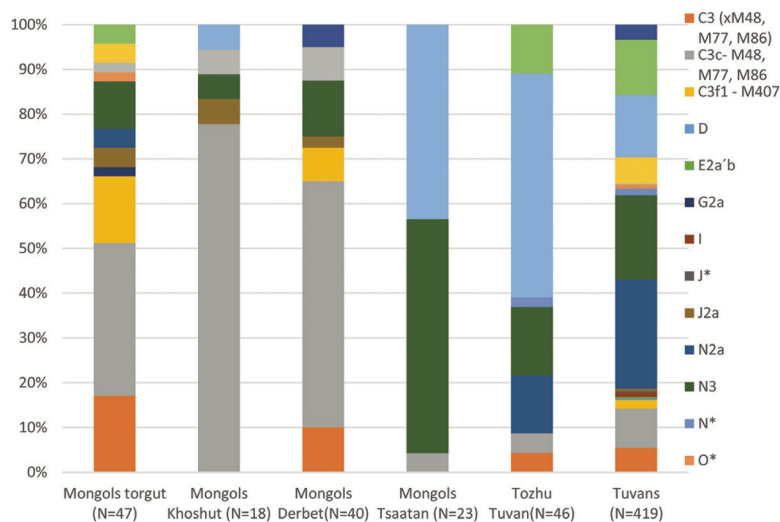


Fig. 1. Bar chart of Y-chromosomal haplogroup distribution among the studied populations.

**TABLE 2**

MITOCHONDRIAL DNA HAPLOGROUPS IN THE POPULATIONS OF THE TSAATANS OF MONGOLIA AND THE TOZHUS OF TUVA.

Haplogroups	Tsaatan N = 23	Tozhu N = 46
C4a1	4.55	2.17
C4a1a	4.55	0.00
C4a2a1	0.00	8.70
C4b	22.73	30.43
C5a1	18.18	6.52
C5b1a1	4.55	6.52
C5d1	0.00	2.17
D4	9.09	2.17
D4o2a	0.00	2.17
F1a1	4.55	0.00
F1b1b	4.55	23.91
G1a1	0.00	4.35
G2	4.55	8.70
G2a1e	0.00	2.17
H20a	4.55	0.00
M7c1	4.55	0.00
T2	4.55	0.00
Y1	9.09	0.00

C4a2a1, C4b, C5a1, C5b1a1, C5d1, D4, D4o2a, F1b1b, G1a1, G2, G2a1e), of which C4b (30.43%) and F1b1b (23.91%) are major haplogroups (Table 2, Figure 2). According to the HVS-1 data, 15 haplotypes were found and the diversity coefficient of 0.8677 turned out to be much lower than among the Torghut of Mongolia (0.9857). In 23 Tsaatans, 14 haplogroups were determined (C4a1, C4a1a, C4b, C5a1, C5b1a1, D4, F1a1, F1b1b, G2, H20a, M7c1, T2, Y1), with C4b (22.73%) and C5a1 (18.18%) being the most common. According to HVS-1, 14 haplotypes were revealed, and the diversity is 0.9486.

**Discussion**

The presented diversity indicates the similarity of the gene pools of the Tsaatans and Tozhus with the results obtained for Tuva populations, where a greater number of haplogroups were found, and the most frequent haplogroups are N1b (N3), N1c (N2), Q1a3 (Q1a1b)<sup>31</sup>. The phylogeography of haplogroup N is well structured: the sub-clade N3b-B187 being specific to southern Siberia and Mongolia is found in Kazakhs, Altaians, and Tuvans; N3a5 is restricted to eastern Eurasia, N3a5-F4205 is prominent around Lake Baikal among Mongolic-speaking Buryats and Mongols; the N2a1-B478 group covers the western and southern parts of Siberia, the Taimyr Peninsula and the Volga-Uralic region and does not extend to eastern Siberia<sup>32</sup>.

Haplogroup Q1a1b-M25 was frequent in Turkic-speaking populations, and its time estimates were at approximately 3–5 Thousand Years Ago<sup>33</sup>. It had spread from Central Asia to Western Asia and Central Europe. The results coincided with the Turkic nomadic migrations from Southern Siberia and Mongolia to Central and Western Asia, the Caucasus and Eastern Europe<sup>34</sup>. The high level of this contribution to the gene pool of the Tsaatans and Tozhus is evidence of an ancient component that has been preserved in remote taiga regions.

The low frequency of haplogroups C3 and C2, that are widespread in the steppe area of Mongolia, indicates that waves of population displacement associated with nomadic migrations of Mongols penetrated the taiga zone insignificantly (Figure 1).

The haplogroup R1a in southern Siberia demonstrates the geographical variation in occurrence, reaching a maximum of more than 50% in the south of Altai, and up to 18% in Tuva populations<sup>31,35</sup>.

The genetic diversity was assessed based on the frequencies of diallelic haplogroups in populations. The minimum diversity in the frequencies of haplogroups was found in the Tozhus of Tuva (0.66) and the Tsaatans of Mongolia (0.56), which is apparently due to the inaccessibility and isolation of small mountain populations.

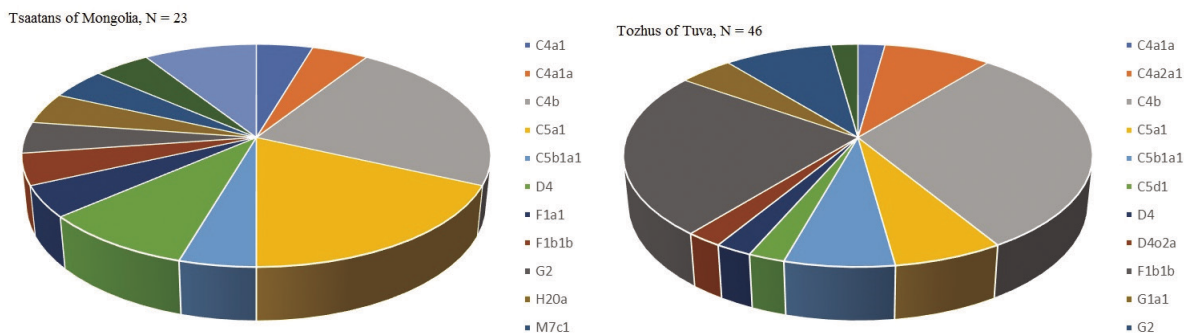


Fig. 2. Mitochondrial DNA haplogroups (%) in the populations of the Tsaatans of Mongolia (a) and the Tozhus of Tuva (b).

**TABLE 3**  
 AMOVA RESULTS FOR STUDIED POPULATIONS BASED ON  
 Y-CHROMOSOME HAPLOGROUP FREQUENCIES

Source of variation	%variation	F-statistics	P
Among geographic groups <sup>a</sup>	1.77	$F_{CT}=0.01771$	0.13196
Among populations within groups	12.89	$F_{SC}=0.13119$	0
Within populations	85.34	$F_{ST}=0.14658$	0
Among groups according to CA <sup>b</sup>	15.67	$F_{CT}=0.15670$	0.00293
Among populations within groups	5.20	$F_{SC}=0.06168$	0
Within populations	79.13	$F_{ST}=0.20872$	0

a – Two groups: Europe (Kalmyk Derbet, Kalmyk Khoshut, Kalmyk Torghut, Kalmyk Buzava, Xinjiang Kalmyk) and Asia (Mongol Derbet, Mongol Khoshut, Mongol Torghut, Sart Kalmak, Mongol Tsaatan, Tozhu Tuvan).

b – Three groups according to correspondence analysis results based on haplogroup frequencies: (1) Kalmyk Derbet, Kalmyk Khoshut, Kalmyk Torghut, Kalmyk Buzava, Mongol Derbet, Mongol Khoshut, Mongol Torghut, Xinjiang Kalmyk, (2) Mongol Tsaatan, Tozhu Tuvan and (3) Sart Kalmak.

Position of the studied populations among the geographic neighbors shows that the Tsaatans of Mongolia and Tozhu are close to the populations of reindeer herders who are at a considerable distance, which indicates a common origin (Table 3).

That is why for a comparative analysis, literature data on geographically neighboring and Northern reindeer herders were taken<sup>36,37</sup>. Cluster analysis was carried out for a complex of haplogroup frequencies in each studied sample. Haplogroups with the frequency of at least 25% in all cohorts (19 in total) was selected. For clustering, the Ward's method was used. An important property of the Ward procedure is the robustness to the influence of various kinds of statistically random interference. As a result, clusters are obtained for which the internal variability of features is close to the least<sup>38</sup>. An interesting fact is the unification of the Tsaatans of Mongolia with the Khants (Figure 4), which are located at a significant geo-

graphic distance. We can assume that N3a5a and Q2a haplogroups were associated with the resettlement of reindeer herders and further with the Finno-Ugric peoples. The subclade N1a1a1a1a-L392 (N3a) was determined in ancient samples BOO002 and BOO004 (aged about 3500 years ago) from the Oleneostrovsky burial ground in the Murmansk region (Bolshoi Oleny Island, Kola Bay)<sup>39</sup>. The oldest sample N1a (subclade N1a1a1a1a-L1026 / Z1973, L392 or N3a3'5) from the Estonian Iron Age dated back to 770–430 BC<sup>40</sup>. Q1a1b-YP1500 / B143 > YP1500 \* was determined for sample Kolyma1 from the Duvanny Yar site (9769 years ago)<sup>41</sup>.

Based on the data on the nucleotide variability of mtDNA HVS1, in samples from populations of Torghut, Derbet, Tozhu and Tsaatan, we determined the following indices: gene diversity (H), absolute number of detected haplotypes (K), number of polymorphic sites (S), average number of pairwise nucleotide differences (Pi), as well as Tajima's D and Fu's  $F_s$  neutrality tests (Table 4). The highest level of genetic diversity in the populations studied is observed in the Torghuts of Mongolia (0.9857), which also corresponds to the high diversity of haplotypes found in them (43 at N=60). The lowest value is found in the Tsaatan population (0.8677), the mitochondrial diversity of which is represented by haplogroups C4b (22.73%) and C5a1 (18.18%). Statistically significant Fu's  $F_s$  values were obtained for all populations studied. The high reliably negative value of the Fu's  $F_s$  test for selective neutrality<sup>42</sup> in populations indicates exponential growth in population numbers. Negative values of Tajima's neutrality tests<sup>43</sup> were also obtained for all populations except the Tozhu population, where the value is close to 0, which indicates selective neutrality and a constant population size. Inconsistent neutrality test values for the Tozhu population may be a result of heterogeneous mutation rates in the mtDNA control region, which could mask the signals of population expansion.

The results of the analysis of the Y-chromosome haplogroups suggest that the Tsaatans of Mongolia and the

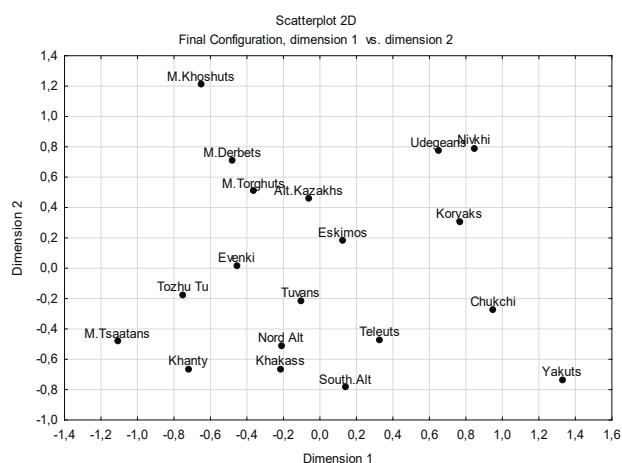


Fig. 3. Position of the studied populations among geographical neighbors.



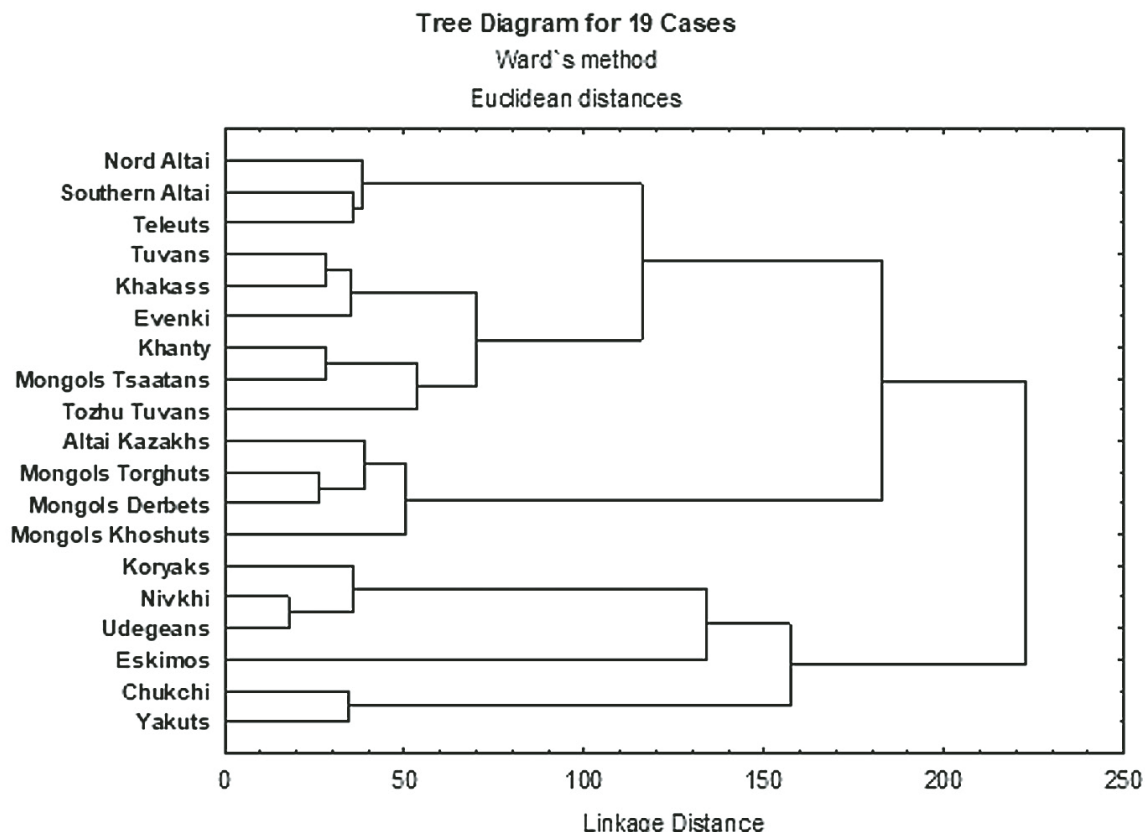


Fig. 4. Cluster analysis of the studied populations with geographic neighbors and Northern reindeer herders.

**TABLE 4**  
GENETIC DIVERSITY AND NEUTRALITY TEST VALUES BASED ON HVS-1 DATA

Ethnic groups	N <sup>1</sup>	H <sup>1</sup>	K <sup>1</sup>	S <sup>1</sup>	Pi <sup>1</sup>	Tajima's D <sup>1</sup>	Fu's Fs <sup>1</sup>	East Eurasian mtDNA types, %	West Eurasian mtDNA types, %
Mongols Torghuts	60	0.9857	43	73	8.46060	-1.55987	-24.78814	0.8	0.2
Mongols Derbets	38	0.9832	30	56	7.02091	-1.63832	-25.06715	0.74	0.26
Tozhus of Tuva	47	0.8677	13	32	6.69843	-0.25480	-25.11964	100	0
Tsaatans of Mongolia	22	0.9526	15	33	5.78656	-1.35028	-22.32441	0.91	0.09

1 N — sample size; H — genetic diversity; K — number of revealed haplotypes; S — number of polymorphic sites; Pi — average number of nucleotide differences based on paired alignments; Tajima's D — Tajima's D test for neutrality (p<0.05); Fu's Fs — Fu's Fs test for neutrality (p<0.02).

Tozhus of Tuva, engaged in reindeer herding, appeared to have common paternal lines with other reindeer breeders (Khanty). Close ethnocultural contacts with Tuvans demonstrate some paternal genetic similarity, which could indicate a common origin. The anthropological research program conducted by us came to similar conclusions<sup>44</sup>. The data obtained on the variability of the Y chromosome and mtDNA in the Tozhus and Tsaatans are due to the isolated and inaccessible taiga region and the manifestation of the “founder effect”. The Tsaatans are less polymorphic in terms of the variety of Y chromosome haplogroups, while the Tozhus are less diverse in terms of mitochondri-

al DNA HVS-1 haplotypes, which is probably a consequence of a high rate of endogamic marriages in the populations studied.

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## **ANALIZA POLIMORFIZMA UNIPARENTALNIH BILJEGA U POPULACIJAMA KOJE SE BAVE UZGOJEM SOBOVA: TOZHU TUVANCI IZ RUSIJE I TSAATANI IZ MONGOLIJE**

### **SAŽETAK**

Analizirali smo podatke o varijabilnosti Y kromosoma i mitohondrijske DNK (mtDNA) u populaciji Tsaatan u Mongoliji i populaciji Tozhu Tuvana u Rusiji. Ispitane populacije karakterizira niska genska raznolikost za oba markera. Analiza haplogrupa Y kromosoma u otkrila je tri haplogrupe u u populaciji Tsaatan i sedam haplogrupa u skupini Tuvanaca. Sastav haplogrupa podudara se s podacima o Tuvancima iz literature, što se objašnjava zajedničkim podrijetlom. Prema podacima o varijabilnosti mitohondrijske DNK, u 46 Tuvanaca utvrđeno je 12 haplogrupa od kojih su glavne C4b (30,43%) i F1b1b (23,91%). Prema podacima HVS (HyperVariable Segment) –1, 15 haplotipova pronađeno je kod Tozhu Tuvanaca, a koeficijent raznolikosti od 0,8677 pokazao se mnogo nižim nego kod Torghuta iz Mongolije (0,9857). U 23 Tsaatana određeno je 14 haplogrupa; od kojih su najčešći C4b (22,73%) i C5a1 (18,18%). Prema HVS–1, kod Tsaatana je otkriveno 14 haplotipova čija raznolikost je 0,9486. Podaci dobiveni o uniparentalnim markerskim sustavima Tuvanaca i Tsaatana posljedica su izolirane i nepristupačne regije tajge i manifestacije efekta utemeljitelja. Tsaatani su manje polimorfni u pogledu raznolikosti haplogrupa Y kromosoma, dok su Tuvanci manje polimorfni u pogledu mitohondrijske DNK, što je vjerojatno posljedica visoke stope endogamnih brakova u proučavanim populacijama.