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Analysis of the Kazakh tribe Baiuly based on the distribution of the Y-chromosome haplogroups

Abstract. A haplogroup is a group of similar alleles that have a common ancestor in which a mutation has occurred, inherited by all descendants. Haplogroups, particularly from the Y-chromosome (Y-DNA), is widely used in population genetics and genetic genealogy, a science that studies the genetic history of mankind. Recent studies of the Y-chromosome of modern Kazakhs have demonstrated the diversity of the Kazakh gene pool. During the expedition carried out in 2014-2016, clinical material was collected from various regions of Kazakhstan, representing samples of peripheral blood and buccal scrapings. All representatives of Kazakh nationality were familiarized with informed consent. In total 1623 respondents participated in the study, 169 of whom were representatives from Baiuly tribe of Junior zhuz. We analyzed the provided samples and found that the Baiuly is characterized by 10 haplogroups, the most prevailing of which is the C2 haplogroup (85%).

Key words: Y-chromosome, haplogroup, haplotype, kazakh, junior zhuz, Baiuly tribe.

Introduction

Historically, Kazakhs were formed by a combination of previously isolated or differentiated nomadic tribes belonging to the Golden Horde. Kazakh tribes were geographically divided into three tribal associations – Senior, Middle and Junior zhuzes. Each of which is characterized by its own unique tribal composition and occupied territory [1]. Nowadays, many modern representatives of previously nomadic ethnic groups, including Kazakhs, have retained knowledge of their tribal affiliation as part of their tradition and culture.

According to the latest estimates of Professor B. Rakishev, carried out in 2013, the smallest of the three Kazakh zhuzes is the Junior Zhuz, numbering 2,521,900 people. The junior zhuz is made up of three main tribes – Baiuly, Alimuly and Zhetiru. The Baiuly tribe is the most numerous tribe in the Junior Zhuz and with 1,120,000 people, it is the third in size among all Kazakh tribes after the Argyn and Dulat tribes [2].

At the moment, the Baiuly tribe mainly inhabits the western part of Kazakhstan, namely in the Mangistau, Atyrau and West Kazakhstan regions, as well as in the border regions of Russia, such as the Astrakhan region or Orenburg. Due to the small number of sources and inconsistent accounts, many questions remain regarding the origin of Kazakh tribes. The formation of Kazakh zhuzes is also not recorded in written history, which contributed to existence of various versions of the Kazakh origin among historians and ethnographers.

In the encyclopedia of Brockhaus and Efron, published from 1890 to 1916: “Baiuly are one of the three generations that make up the Junior Kazakh Horde. According to Kazakh legend, the ancestor of this generation is Kadyrkazhi (Kadyr-Hajja), the son of Alshin, the founder of the Junior Horde. “Baiuly” (Kadyrkazhi) had 12 sons who were the founders of the 12 tribes of Baiuly dynasty. The names of the tribes of the Baiuly dynasty: Berish, Sherkesh, Maskar, Adai, Zhapas (Zhappas), Ysyk, Essentemir, Baibakty, Alasha, Tana, Kyzylkurt and Taz”. How-

ever, there are other facts in the historical literature [3].

According to the genealogy, Baiuly became a member of the Junior Zhuz and joined to the Alshin union. According to some reports, the Baiuly tribe was once located on the banks of the Bai-Taiga (rich taiga), which covers an area of 600 square kilometers

in the upper reaches of the Alash river (Figure 1). The people of this taiga, with rich wildlife, where hunting is practiced, probably joined the Jochi army under the name of Bai. The name «Baiuly» first appears in historical documents in 1561. On June 23 same year, the bi of Nogai Horde Smail, in a letter to the Russian Tsar Ivan the Terrible, called Baiuly as ulus [4].

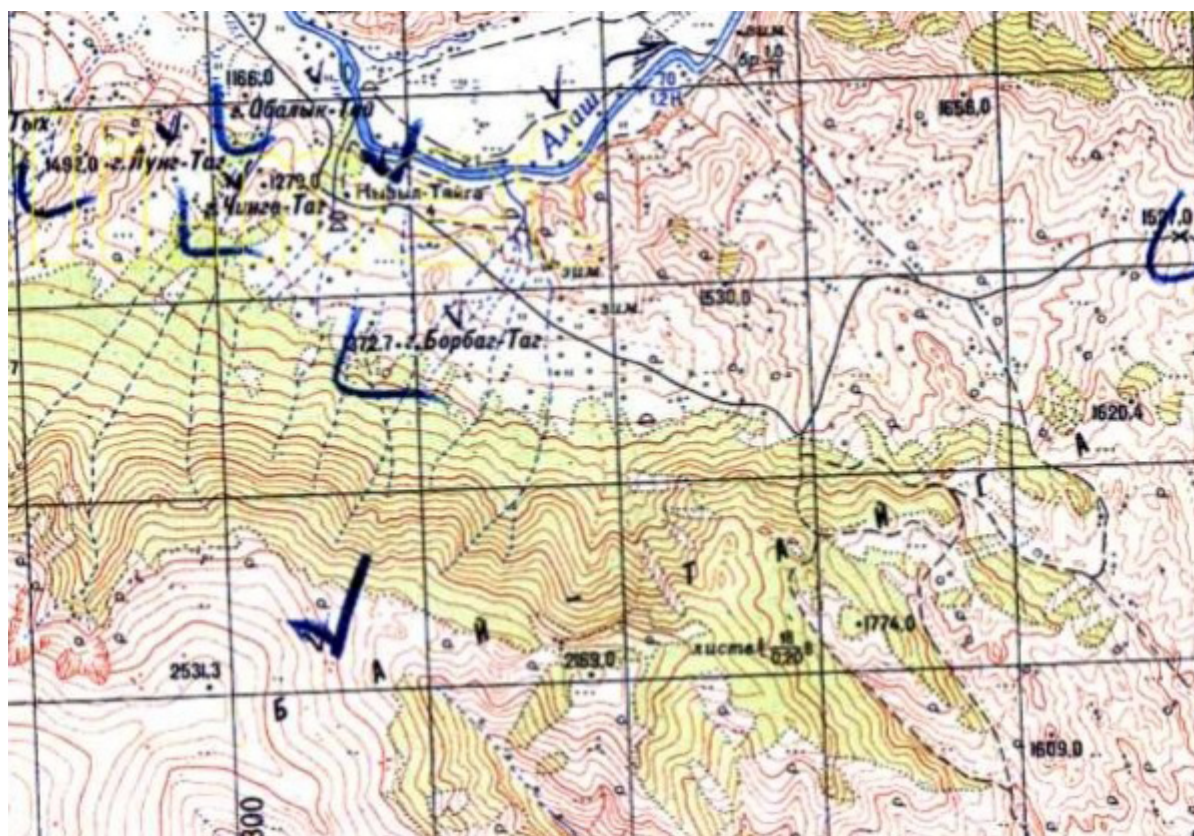


Figure 1 – Bay-taiga region in the upper reaches of the Alash River, which in ancient times was the habitat of the Bayuly tribe [5]

In addition to the study of historical text, another important approach to study the formation of any ethnic groups, tribes or tribes is through population genetic research of the ancient and modern population using modern methods of physical anthropology and molecular genetics.

Research on the Y-chromosome is the most relevant to the current study here. Because of haploidy, Y-chromosome is transmitted strictly through the paternal line and does not undergo recombination. As a result, the Y chromosome haplogroups are very effective markers that can be used to study migration events in the history of certain peoples and the formation of an ethnic group as a whole. Such studies have been carried out in many populations around the world. At this time, there is an active accumula-

tion of information on various markers of the Y-chromosome in Kazakh tribes [6-8]. However, the haplogroup of the Baiuly tribe has not been sufficiently studied.

Therefore, the purpose of this work was to study the composition of the gene pool of the Baiuly tribe using materials from the Population Genetics Laboratory of the Institute of General Genetics and Cytology.

Materials and methods

The study used biomaterials of the modern population of Kazakhstan (DNA, blood and buccal scrapings) stored in the Genbank of the Laboratory of Population Genetics of the Institute of General Genetics and Cytology. In total, 1623 volunteers participated

in the study, of which 169 people are representatives of the Baiuly tribe. The main biomaterial was collected during the expedition in 2014-2016 from Kazakh nationals with voluntary informed consent across various regions of the Republic of Kazakhstan. The ethnic and tribal affiliation of the volunteers was determined by a detailed individual questionnaire.

DNA was extracted from buccal scrapings and frozen (-20 °C) peripheral blood samples containing EDTA as an anticoagulant agent. The QIAamp DNA Mini Kit (Qiagen, Germany) and the GeneJET Genomic DNA Purification Kit (Thermo Scientific, USA) were used for extracting genomic DNA.

Quantitative and qualitative assessment of the extracted DNA was performed using a DNA photometer (Biofotometer Plus, Eppendorf, Germany) and electrophoretic analysis using EV265 Power Supply (Consort, India).

Genotyping of 17 polymorphic STR loci (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385a, DYS385b, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS448,

DYS456, DYS458, DYS635, multi-GATA format) by PCR using the enzymatic amplification system of the AmpFlSTR Yfiler™ kit (Life Technologies, USA). Amplification products were separated and determined using an ABI PRISM 310 genetic analyzer (Applied Biosystems, USA). Alleles were identified using the GeneMapperID ID-X v1.4 software (Thermo Scientific, USA) based on the allelic ladders included in the sets.

Previously, haplogroups were identified using Whit Athey's online predictors (<http://www.hprg.com>) and NEVGEN (<http://www.nevgen.org/>) based on microsatellite haplotype data. Then the samples were tested for 15 single nucleotide polymorphisms (M231, M217, M215, M17, M285, M242, M343, M172, M122, P37.2, M61, P15, M253, M479 and M267) to clarify the haplogroups of the Y chromosome. Analysis of polymorphisms was performed using PCR with subsequent restriction and analysis of the length of restriction fragments (PCR-RFLP). The primer sequences and reaction conditions are shown in Table 1.

Table 1 – The primer sequences and reaction conditions

| Haplogroup | Marker(1) | Primer sequence 5'-3' | Fragment Size (bp) | Mutation | Mutated position (nt) | Enzyme | Allele Fragments (bp) |
|------------|-----------|--|--------------------|----------|-----------------------|-----------|-----------------------|
| C2 | M217 | f tgaaggagaatgaaaaagtgggtggc r agcaaaagataaattgtccagggt | 352 | A→C | 327 | BanI | (A)352 (C)327/25 |
| N | M231 | f attatcctggaaaatgtgggctcg r tccgattcctagtcacttggtt | 326 | G→A | 326 | TaqI | (G)221/105 (A)326 |
| E1b1b | M215 | f gtaaaactcagatatatacatcccatg r aaaaaaaaaagaatcactatcttaacg | 386 | A→G | 222 | BseI1 | (A)386 (G)222/164 |
| R1a1 | M17 | f ctggtcataaactggaaatc f tgaacctacaatgtgaaact | 168 | 4G→3G | | DdeI | (4G)168 (3G)24/144 |
| Q | M242 | f aactcttgataaacctgtgctg r tccaatctcaattcatgcctc | 366 | C→T | 180 | Alw21I | (C)187/179 (T)366 |
| G2a | P15 | f gagtttctaacagggegtaaca r caacttcatctgccttcagac | 155 | C→T | 133 | HpyCH4IV | (C)134/23 (T)157 |
| I1 | M253 | f gcaacaatgagggttttttg r cagctccacctctatgcagttt | 400 | C→T | 283 | HincII | (C)280/120 (T)400 |
| I2 | P37.2 | f cgtctatggccttgaaga r tccgaaaatgcagacttt | 447 | T→C | 135 | HpyCH4III | (T)447 (C)311/136 |
| J2 | M172 | f aaataggagccagatgacc r aataataaftgaagacctttgagt | 176 | T→G | 151 | HinfI | (T)176 (G)151/25 |
| L | M61 | f attggattgattcagccttc r attttatttctgtgtccttgc | 190 | C→T | 98 | TaqI | (C)97/93 (T)190 |
| R1b | M343 | f gcagagtgcctctggtt r acctggaaccagtgcctct | | A→G | | Hpy8I | |

Continuation of table 1

| Haplogroup | Marker(1) | Primer sequence 5'-3' | Fragment Size (bp) | Mutation | Mutated position (nt) | Enzyme | Allele Fragments (bp) |
|------------|-----------|---|--------------------|----------|-----------------------|---------------------|--------------------------------|
| G1 | M285 | f ggatggacaaggaagactttca r ctgttgcccaggctagtgtc | 371 | G→C | 371 | RsaI | (G)175/196 (C)371 |
| O2 | M122 | f tagaaaagcaattgagataactca r gcgatgctgatatgctagttcag | 122 | A→G | 73 | Hin1III (NlaIII) | (A)100/22 (G)122 |
| J1 | M267 | f acgtcacccatctcaacatc r aaagaatgtctcccattgagg | 306 | T→C | 189 | DdeI | (T)158/69/49/3 (C)207/69/30 |
| R2 | M479 | f gatactttatcaggettacttc r cgattctgagagatttggt | 323 | C→T | 107 | HphI | |

Results and discussion

The study of the haplotype diversity of the Y chromosome in the Kazakh population belonging to the Baiuly tribe (Junior Zhuz) revealed 10 haplogroups, of which 8 were identified as a result of testing single nucleotide polymorphisms, and 2 haplogroups (O1b2 and T) were determined by the NevGene predictor using the microsatellite haplogroup. Analysis of the spectrum of Y-chromosome haplogroups in the Baiuly tribe showed the presence of a major haplogroup C-M217, which makes up 85% of their gene pool. The remaining 15% of the Baiuly tribe gene pool is represented by the sector of 9 identified haplogroups,

but their percentage is extremely small, where each of them is less than 5% of the total composition of haplogroups (Figure 2).

Haplogroup C-M217, also known as C2 (and previously as C3), is the most frequently occurring branch of the wider Y-chromosome DNA haplogroup C (M130) [9]. It is found mostly in central Asia, Eastern Siberia and at is present at significant frequencies in part of East Asia and Southeast Asia including some populations in the Caucasus and Middle East. It is assumed that haplogroup C-M217 originated approximately 7,100 – 16,700 years ago in eastern or central Asia. The closest phylogenetic relatives are found in the vicinity of South Asia, East Asia, or Oceania.

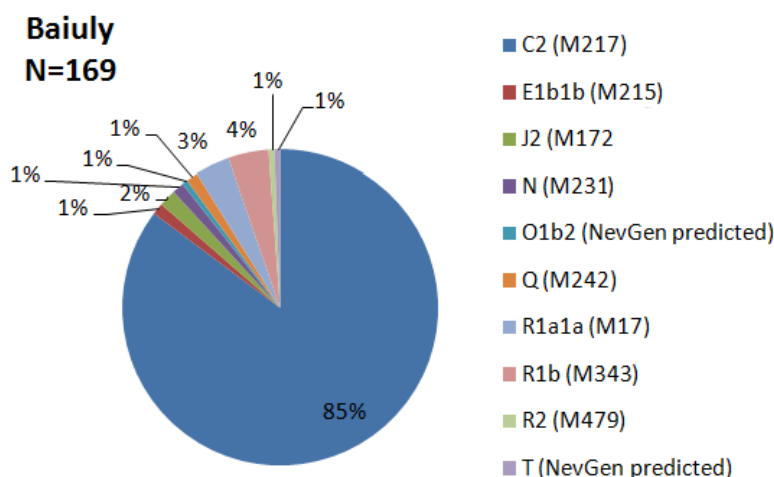


Figure 2 – Diagram of distribution of the Y-chromosome haplogroups in the Kazakh tribe Baiuly

The haplogroup C-M217 is now found at high frequencies among Central Asian peoples, indigenous Siberians, and some Native peoples of North America. In particular, males belonging to peoples

such as the Buryats, Evenks, Kalmyks, Kazakhs, Mongolians, and others have high levels of M-217 [10-17]. Among Kazakhs from different regions of Kazakhstan, the total occurrence of variants of hap-

logroup C2 on average reaches 80% [18-22]. One particular haplotype within Haplogroup C-M217 (star-cluster C2*(C2*-ST)) has received a great deal of attention, because of the possibility that it may represent direct patrilineal descent from Genghis Khan [23], though that hypothesis is controversial. In a recent study [24] of Y-chromosomes of more than 800 people, carriers of the “star-cluster” C2 *, showed that its origin and distribution in Eurasia is more likely associated with the ancient Mongol-speaking tribes than with Genghis Khan. According to the data, the estimated age of the C2 * – ST star cluster was 2576 years, preceding the rise of the Great Mongol Empire [25]. The authors hypothesize that the C2 * -ST line originated in the northern region of the Greater Khingan and spread during the dispersal of ancient Mongolian-speaking populations. Thus, this is the initially dominant haplogroup of the “proto-Mongols”. The highest frequency of C2 * -ST was noted in several populations of Kazakhs in South-East Kazakhstan, followed by populations of North-West China, Mongolia, Buryatia and Uzbekistan. In Kazakhstan, with a frequency of more than 50%, C2 * -ST occurs in tribal groups of the Elder Zhuz and the Kerey tribe of the Middle Zhuz [14], [20].

After haplogroup C2-M217, the second most prevalent component of the Y-chromosome gene pool of the Kazakh tribe Baiuly (about 8%) is represented by three variants of haplogroup R: R1b (M-343) – 4.1%, R1a1a (M-17) – 3.6% and R2 (M-479) – 0,6%.

Haplogroup R1b arose from a mutation of the haplogroup R1 that occurred in a man who lived about 22,800 years to the present day (the date was determined from SNPs by YFull [26]). The last common ancestor of R1b carriers lived 20.4 thousand years ago [21].

A number of modern geneticists believe that R1b originated in Central [27] or Western Asia [28] presumably 16,000 years ago [29]. At first, the hypothesis was put forward that R1b is indigenous to Western Europe, the geographical location where the haplogroup is most prevalent. Subsequently, it was shown that R1b haplotypes have greater variety of small side branches in Anatolia and the Caucasus than in Europe [30].

As was mentioned R1b is the most common haplogroup in Western Europe, reaching over 80% of the population in Ireland, the Scottish Highlands, western Wales, the Atlantic fringe of France, the Basque country and Catalonia. It is also common in Anatolia and around the Caucasus, in parts of Russia and in Central and South Asia. Besides the Atlantic and North Sea coast of Europe, hotspots include the

Po valley in north-central Italy (over 70%), Armenia (35%), the Bashkirs of the Urals region of Russia (50%), Turkmenistan (over 35%), the Hazara people of Afghanistan (35%), the Uyghurs of North-West China (20%) and the Newars of Nepal (11%). R1b-V88, a subclade specific to sub-Saharan Africa, is found in 60 to 95% of men in northern Cameroon [31]. It is also found in Central Asia, Eastern Europe, North Africa, Western Asia. After the migration of Europeans to America and Australia, it makes up a significant share there as well.

R1b is found in almost all kazakh tribes, with predominance in the Kypshak tribe of the Middle Zhuz [32].

The haplogroup R1a is a Y-chromosomal haplogroup common in Central and Eastern Europe, Central and South Asia, South Siberia and Scandinavia [33,34].

R1a arose about 22 thousand years ago [35] (according to other sources – about 25 thousand years ago [29]) from a mutation of the haplogroup R1 that occurred in a man who lived about 22 800 years ago (the date was determined from snips by YFull [36]) presumably in Asia.

While R1a arose about 22,000 to 25,000 years ago, its subclass (R1a1a1) diversified around 5800 years ago [29]. The place of origin of the subclade plays a role in the debate about the origin of the Proto-Indo-Europeans.

There are different hypotheses about timing and origin of haplogroup R1a. According to recent studies [37], R1 haplogroup and its subclade R1a resulted from a series of mutations on the root R haplogroup. The time of occurrence is approximately after the last glacial maximum. The exact place of origin of the haplogroup R1a is currently unknown, but could include geographic regions such as Pakistan, North-west India, the Balkans as these regions have the greatest genetic diversity of this haplogroup. An alternative hypothesis is that the haplogroup R1a came to the Balkans from migratory flows coming from the Eurasian steppes, and due to the fact that migrations occurred in waves, a diversity of mutations was provided. In South Asia, for 10,000 years, the density and number of population are the largest on the planet, and therefore the diversity of the haplogroup is also great. Based on this, geneticists suggest that the R1a haplogroup could have arisen either in Central Asia or in southern Russia – in Siberia [33].

In Kazakhstan R1a also is found in representatives of almost all kazakh tribes, but with a predominance in the Kozha tribe [16]

The rest of the haplogroups of the Y-chromosome were found in the sample of representatives of

the Baiuly tribe with a frequency of less than 2%. Thus, the genetic portrait of the Kazakh tribe Baiuly, at least through the male lineage, is determined by a very high contribution of the Central Asian (proto-Mongolian) component (haplogroup C-M217) and a small presence of a “paleo-European” substrate (variants of haplogroup R), which prevailed in Central Asia during the Scythian-Sarmatian period.

Conclusion

In this study, we have characterized for the first time the gene pool of the Kazakh tribe Baiuly. Our study was based on a single extensive panel (17 STR loci, 15 SNP) of Y-chromosome markers in a sample of 169 people. For representatives of this tribe, the major haplogroups are C2-M217, as well as R1b – M343, typical for the peoples of the countries of Western and Central Europe [38]. In general, this tribe has a fairly high level of genetic diversity.

The information we have uncovered about the ethnogenesis of the Baiuly tribe is important for its people given the increasing ethnic self-awareness, the strengthening of national culture and language, and the growing interest in the history of their people. In addition, the results are important for understanding the ethnogenesis of Kazakhs, which are relevant for anthropologists, archaeologists, linguists, historians, or ethnographers interested in the reconstruction of the history of the people of Kazakhstan. This study lays the foundation for subsequent large-scale studies of the Baiuly tribes with its unique history, including genome-wide analysis.

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