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of the Institute of Plant Biology and Biotechnology

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MtDNA CONTROL REGIONS ANALYSIS AT ETHNIC KAZAKHS

Abstract. The analysis of mitochondrial DNA of modern Kazakhs with a wide geographic localization and tribal affiliation shows that the Kazakh's maternal lines are characterized by great diversity of mtDNA haplogroups, reflecting the historical migration of the population of Eurasia with the predominance of "Asian" and "European" components. The analysis of mutations of hypervariable regions of mtDNA (HVR1, HVR2) regarding affiliation to Zhuz determines the genetic affinity of all Kazakh maternal lines.

Keywords: modern Kazakh, mtDNA, HVR1, HVR2, haplogroup.

Due to intensive studies over the past decades, considerable data on the DNA polymorphism in human populations have been accumulated. A variety of polymorphic markers are used to analyze the gene pool of populations and individual ethnic groups, determine their basic characteristics, dynamics, history and geography. There are high polymorphic loci of coding genes, insertion-deletion polymorphism, micro- and minisatellite, single-nucleotide substitution (SNP), and copy number variants (CNV).

The Y-chromosome and mitochondrial DNA (mtDNA) polymorphisms have become widely used genetic markers in population studies related to the genogeography and Human history because they reflect the inheritance and variability of paternal and maternal lines. The mtDNA polymorphism characterizes the absence of recombinations and the maternal type of inheritance. In this regard, the mitochondrial genome can evolve only through the successive accumulation of mutations in generations. In the mitochondrial genome, two main areas are identified: the coding region and the non-coding region (D-loop). The non-coding region is the most variable region of mtDNA and contains about 23% of the variations in the entire mitochondrial genome. About 30% of the variability of mtDNA associates with interpopulation and intergroup differences. In comparison with the nuclear genome, the mtDNA mutation rate determined the advantages of mitochondrial DNA in front of nuclear genetic markers, which make it possible to use it as a tool for population-genetic studies.

To date, there are huge data on population-based mtDNA polymorphism in many modern and ancient human populations. In this respect, the most studied are the peoples of the New World, Africa, Southeast Asia and Oceania, as well as Western Europe. At the same time, polymorphism of the mitochondrial genome of Turkic-speaking peoples, one of the vast and most ancient ethnic groups in Eurasia, has been little studied.

The ethnogenesis of the Kazakh people is based on historical, demographic, population-genetic and anthropological background, conditioned by the influence of the Aryan, Indo-Iranian and Turkic tribes, from Saks, Sarmatians, Massagets to Huns, Turks, Karluks, Oguzes, Kimaks, Kipchaks and other ethnic groups. Later Turkic-Mongolian tribes later formed a Kazakh ethnos, known since the 15th century.

A few genetic studies of archaeological material and modern populations from Kazakhstan [1-5] testify the genetic homogeneity of modern Kazakhs, which makes it possible to differentiate the Kazakh people from other Eurasian population, despite the wide polymorphism and intrapopulation diversity.

The usage of mtDNA markers indicates a high heterogeneity of mtDNA haplogroups among Kazakhs and suggests that 55% of Kazakh mtDNA haplogroups originated from Eastern Eurasia, while 41% are from Western Eurasia [2-5]. Some authors point to the similarity of mtDNA haplotypes of modern Kazakhs with barrow cultures of the 1st millennium BC [5, 6]. A study of 304 Kazakhs with a wide geographic distribution (10 populations of rural residents) revealed that 58-59% of Kazakhs exhibit Asian mtDNA haplotypes (D, C, G, A, M and F) and 41% European (H, T, J, K, U2, I, U5 and HVR) [3, 7]. Based on the variability of the hypervariable region of mtDNA (HVR1) analysis of the genetic relationships of different Asian populations, which was performed on a large sample size (33 groups, about 3,000 samples), showed the genetic relationship of Altai Kazakhs with other ethnic groups from Altai (Kalmyks, Soyots, Hamnigans, Buryats, Tolengites and other Altai peoples) [7-9]. The distinct differences in the nature of genetic variability between the maternal and paternal genetic lines among Altai Kazakhs (119 individuals) was noticed by Dulik M.S. with co-authors [10]. While on the maternal line the mtDNA haplotypes widespread in the East and West Europe were determined, the paternal type of inheritance demonstrated a low genetic diversity. Population-genetic study of the Y-chromosome and mtDNA of 160 representatives of the Kazakh tribe Naiman showed that maternal lines represent high migration activity and the diversity of mtDNA [11].

So, the most studies were conducted on a representatives of separate tribe or specialized geographically location. To replenish the information about maternal lines of modern Kazakhs we conducted a research of a selected cohort with high tribal affiliation and geographic distribution.

Materials and methods. *Study objects*. The study objects were data of questionnaires, EDTA-treated peripheral blood samples which were taken from 96 modern Kazakhs with different tribal affiliation and geographic localization. Before collection of blood samples we asked people the voluntary consent to participate in genetic research. A detailed questioning was done after obtaining the signed voluntary informed consents. The knowledge of the maternal line history was the main selection criterium for the study. Questionnaires included the information on the paternal and maternal tribal affiliation of the persons (shezhire).

Extraction of DNA. Genomic DNA was extracted from EDTA-treated peripheral blood samples using "Genomic DNA Purification Kit" (Thermo Scientific). Qualitative and quantitative characteristics of the DNA samples were estimated using Eppendorf BioPhotometer plus (Eppendorf, Germany) or NanoDrop 2000 (Thermo Scientific, USA). DNA samples were stored at -20°C.

Mitochondrial DNA analysis. Sequencing of the hypervariable regions of mtDNA (HVR1, HVR2), as well as complete mtDNA sequencing, were performed using a next generation sequencing (NGS) analyzer MiSeq (Illumina, San Diego, USA).

PCR amplification, for the creation of four amplicons (nucleotide positions: 29-285, 172-408, 15997-16236 and 16159-16401) representing the 2 hypervariable regions of the ring mitochondrial DNA (HVR1, HVR2), was performed in four separate reactions on a single sample using a kit "Human mtDNA D-Loop Hypervariable Region" (Illumina, San Diego, USA). The quantity and quality of PCR amplicons were determined by electrophoresis and the Quantus ™ Fluorometer. The DNA amplicons were normalized to 0.2 ng/μl and combined in a ratio of 1: 1: 1: 1 in a total volume of 20 μl (5 μl each). The DNA libraries for sequencing were obtained from normalized PCR products (1 ng total input) using the "Next Era XT DNA Sample Preparation" kit (Illumina, San Diego, USA) in accordance with the manufacturer's guidelines. The mtDNA samples were combined with 25% PhiX control and were sequenced at a concentration of 10 pM according to the manufacturer's instructions in 2 x 151 cycles.

Bioinformatic and statistical analysis. The MiSeq data was analyzed using the mtDNA MiSeq Reporter (MSR) plug-in, and interpreted using the BaseSpace® mtDNA Variant Processor v1.0 App (Illumina, San Diego, USA) software. This program allows to align and order the sequenced sequences which are matched to the reference sequence of the full mtDNA genome - Cambridge Reference Sequence (rCRS) [12]. The program allows to analyze any part of the complete genome of the ring mtDNA using a quality and overlapping coverage (Quality for variant call Q30) with the identification of start points. To use this program, the primary sequencing data obtained in the FASTQ format (mtDNA MiSeq Reporter)

were converted into BAM and VCF formats. For identification of mtDNA haplotypes we used the Haplogrep programs (version 2.0, https://haplogrep.uibk.ac.at/), Haplofind (https://haplofind.unibo.it) and mtDNA manager [13]. Mutation frequencies, molecular difference analysis and pairwise genetic distances were calculated using the GeneAlEx 6.2 program. Phylogenetic relationships were defined using the MEGA7 program.

Results and their discussion. The selection a cohort for the study was determined by the availability of personal data on the maternal lines of inheritance. At the same time, we were oriented to the fact that at least 2 generations (mother and grandmother) would belong to the same clan by maternal inheritance. Also we have count the wide geography distributions and tribal affiliation. Thus, 96 persons were selected, who belonging to different families and having different geographical localization. Among them were 55 men and 41 women.

The table S1 (Application 1) presents our data on shezhire, mtDNA control region mutations (HVR1 & HVR2) and sex of investigated individuals. The diversity of mtDNA haplotypes in cohort of modern Kazakhs with different tribal affiliations and geographical localization is shown on figure 1.

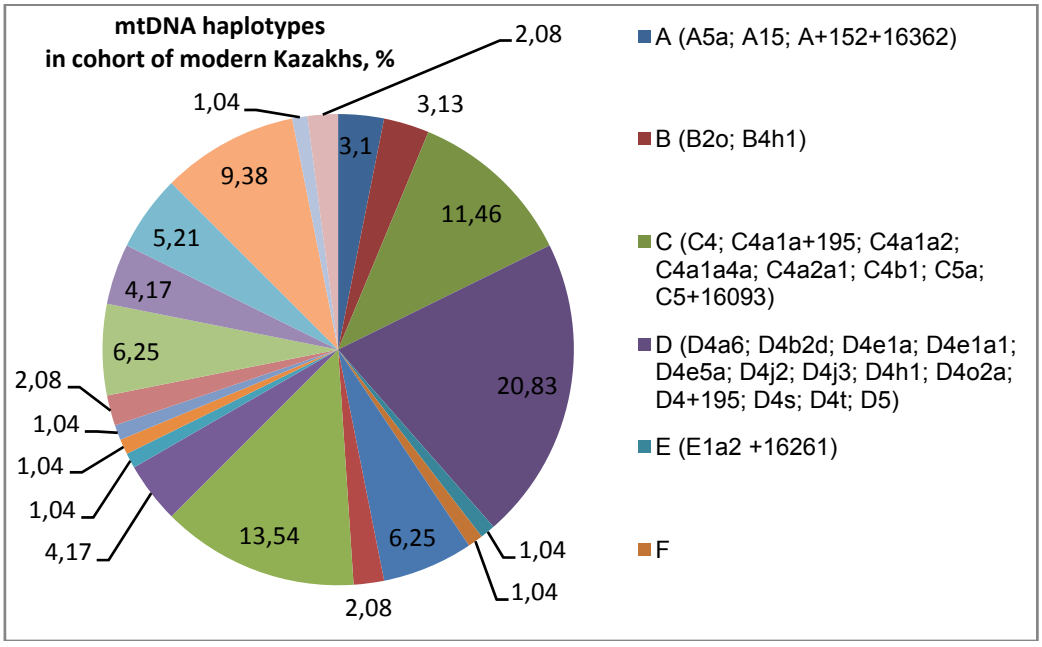

Figure 1 – mtDNA haplotypes in cohort of modern Kazakhs

In total, 81 haplotypes of mtDNA were identified in the studied cohort:

1) A5a; A15; A+152+16362; B2o; C4; C4a1a2; C4a1a4a; C4b1; C5a; C5+16093; D4a6; D4b2d; D4e1a; D4j2; D4h1; D4o2a; D4s; E1a2 +16261; F; G2a2; G2a5; I1; I1a; H1e1a4; H1+16189; H1ao; H1b; H1e2c; H1e-16129; H2a+152+16311; H2a2a; H2a2a2; H7a1; H13a1d; HV4b; J1c16; J1d; J2b1c1; JT; K1c; L3h1a2a; Z+152 (M8); M6; M10a1a1b; N1a3a; N9a2'4'5'11; R2; R7; R8; R31; T; T1a1; T1a1'3; T2; T2b11; U1a3; U1b2; U2e1h; U2e1b; U2e1'2'3'; U3a3; U4; U4b1b1c; U5a2+1629; X2; W3; W4 – for 1 person from studied individuals (1,04%)

2) B4h1, C4a2a1; D4e1a1; D4e5a; D4j3; D4+195; D5; G2a; G2a+152; – for 2 persons (2,08%);

3) C4a1a+195; D4t; – for 3 persons (3,13%);

4) N9a – for 4 persons (4,17%).

As can be seen from presented data, the highest frequency is typical for the D mtDNA haplogroup, which together with the subclades (D4a6, D4b2d, D4e1a, D4e1a1, D4e5a, D4j2, D4j3, D4h1, D4o2a, D4+195, D4s, D4t, D5) represents 20 individuals (20,83%). Subclades of haplogroup H (H1e1a4, H1+16189, H1ao, H1b, H1e2c, H1e-16129, H2a+152+16311, H2a2a, H2a2a2, H7a1, H13a1d, HV4b) were identified in 13 individuals (13,54%). Another most distribted hapogroup of mtDNA is C (11 persons – 11,46%), which represented in studied cohort by subclades of C4, C4a1a+195, C4a1a2, C4a1a4a, C4a2a1, C4b1, C5a, and C5+16093.

Application 1

Table S1 – Haplotypes of mtDNA of studied cohort regarding the tribal affiliation of modern Kazakh individuals

Maternal line				Haplotype	mtDNA mutations		Gender	No. of ind.
Tribe	**Subtribe**	**Clan**	Subclan		HVR1	HVR1		
Uly zhuz								
Zhalaiyr	Andas	Borte		D4e1a1	16093C, 16176T, 16223T, 16362C	73G, 94A, 194T, 263G, 315.1C	male	1
Dulat	?			H1e-16129	16129A	263G, 309.1C, 315.1C	male	1
Saty	?			U2e1'2'3	16051G, 16129C, 16182d, 16183C, 16189C, 16362C	73G, 152C, 217C, 263G, 315.1C	female	1
Alban	Alaman			D4+195	16218T, 16223T	73G, 195C, 263G, 315.1C	female	2
Kanly	?			J1c16	16069T, 16126C, 16218T	73G, 152C, 185A, 228A, 263G, 309.1C, 315.1C	male	1
Oshakty								
Atalyk	Alimbet	Biguly	Karasirak	N9a	16223T, 16257A, 16261T	73G, 150T, 263G, 315.1C	female	1
Konyr	?			C5a	16223T, 16261T, 16288C, 16298C	73G, 249d, 263G, 315.1C	male	1
Taszhurek	?			J1c2	16126C	73G, 185A, 188G, 228A, 263G, 295T, 309.2C, 315.1C	male	1
Uisun (Sary uisun)	?			D4j3	16184T, 16223T, 16311C, 16362C	73G, 146C, 227T, 263G, 309.2C, 310C, 315.1C	male	1
Zhakyp	?			I1a	16129A, 16172C, 16223T	73G, 195C, 199C, 203A, 204C, 250C, 263G, 309.1C, 315.1C	female	1
Pusyrman	?			T2	16078G, 16126C, 16177G, 16294T, 16296T	73G, 263G, 315.1C	female	1
Suan	Aksham			C4	16223T, 16298C, 16327T	73G, 152C, 195C, 248d, 263G, 315.1C	male	1
Shaprashty								
Ikei	Kosai			C4a1a2	16093C, 16129A, 16223T, 16298C, 16327T	73G, 195C, 248d, 263G, 315.1C	female	1
Eskozha	Baba			C4b1	16223T, 16298C, 16327T	73G, 146C, 234G, 248d, 249A, 263G, 309.1C, 309.2C, 315.1C	male	1
Koshek	Alshan			H7a1	16126C, 16261T	263G, 309.1C, 315.1C	female	1
Ysty	Tilik			U1b2	16111T, 16214A, 16231C, 16249C, 16327T	73G, 146C, 152C, 263G, 285T, 309.1C, 315.1C	male	1
Shanyshkyly								
Arynshi	?			T1a1	16126C, 16163G, 16186T, 16189C, 16294T	73G, 152C, 195C, 263G, 309.1C, 315.1C	male	1
Katagan	?			G2a5	16093C, 16223T, 16227G, 16234T, 16278T, 16309G, 16362C	73G, 152C, 263G, 315.1C	male	1
Mamyt	?			K1c	16224C, 16311C	73G, 146C, 152C, 263G, 315.1C	male	1
Orta zhuz								
Argyn	?			U3a3	16148T, 16189C, 16343G, 16355T, 16390A	73G, 185A, 263G, 315.1C	male	1

	Saidaly			N1a3a	16201T, 16223T, 16265G	73G, 189G, 195C, 204C, 207A, 210G, 263G, 315.1C	female	1
	Tolengit			M6	16223T, 16362C	73G, 195T, 263G 315.1C	male	1
Meiram	Suindik	Karzhas		H33c	16188T,	263G, 315.1C	female	1
				D4b2d	16223T, 16274A, 16278T 16287T, 16362C	73G, 151T, 152C, 237G, 263G, 315.1C	male	1
				U2e1h	16051G, 16129C, 16183C, 16193.1C	73G, 195C, 217C, 228A, 263G, 309.1C, 309.2C, 315.1C, 340T	male	1
			Khangeldi-Kushik	H13a1d	16234T	152C, 263G, 309.1C, 315.1C	male	1
		Kulboldy	Aidabol-Tulpar	G2a+152	16169T, 16223T, 16227G 16265C, 16278T	73G, 146C, 152C, 263G, 315.1C	female	1
			Aidabol	R31	16362C	73G, 263G, 309.1C, 315.1C	male	1
				C4a1a+195	16129A, 16223T, 16298C, 16327T	73G, 195C, 248A, 263G, 309.1C, 315.1C	male	1
				G2a+152	16169T, 16223T, 16227G, 16265C, 16278T	73G, 146C, 152C, 263G, 315.1C	male	1
			Kulik	JT	16126C	73G, 146C, 315.1C	male	1
		?		C5+16093	16093C, 16223T, 16288C, 16298C, 16327T	73G, 248A, 263G, 315.1C	male	1
		Orman-shy		J1d	16069T, 16126C, 16153A, 16193T	44.1C, 73G, 146C, 152C 263G, 295T, 309.1C, 315.1C	female	1
	Karakesek	Kernei	Daua	D4t	16129A, 16213A, 16223T 16298C	73G, 195C, 198T, 247d, 309.1C, 315.1C	male	1
		Baibori		N9a	16189C ,16193.1C, 16223T, 16257A, 16261T	73G, 150T, 263G, 309.1C, 315.1C	female	1
		?		A15	16093C, 16223T, 16290T, 16319A, 16362C	73G, 152C, 207A, 235G, 309.2C, 315.1C	male	1
		Aksha	Boshan-Moshai-Koyanshi	D4e5a	16223T, 16274A, 16362C	73G, 152C, 263G, 309.1C, 315.1C	female	1
		Kara		H2a2a2	16184G, 16264T, 16295T, 16390A	60.1T, 64T, 152C, 309.1C, 309.2C, 315.1C	female	1
		Bolat-kozha	Tuiten-Tanas-Botei-Batshor-Shekshek	M10a1a1b	16129A, 16193.1T, 16223T, 16311C, 16357C	73G, 146C	male	1
			Tuiten-Tanas-Botei-Batshor-Shashty	C4a2a1	16167T, 16171G, 16213A, 16223T, 16298C, 16327T, 16344T, 16357C	47A, 73G, 152C, 248A, 263G, 309.1C, 315.1C	male	1
		?		T1a1'3	16126C, 16163G, 16186T, 16189C, 16192T, 16294T	73G, 152C, 195C, 263G, 315.1C	male	1
		?		N9a	16189C, 16223T, 16257A, 16261T	73G, 150T, 263G, 309.1C, 315.1C	male	1
	Begendik	Kozgan		L3h1a2a	16093C, 16193.1T, 16223T, 16311C, 16357C, 16399G	73G, 146C, 199C, 263G, 309.1C, 315.1C	male	1
	Kuandyk	Karpyk	Toka-Kulumbet	B4h1	16129A, 16182C, 16183C, 16189C,	73G,263G, 315.1C	female	2

					16261T			
		Temesh		W4	16129A, 16223T, 16284G, 16287T, 16304C, 16362C	73G, 143A, 189G, 194T, 195C, 196C, 204C, 207A, 263G, 309.1C, 315.1C	male	1
	Basentiyin	Syrym		X2	16182C, 16183C, 16189C, 16223T, 16278T	73G, 146C, 153G, 195C, 263G, 309.1C, 315.1C	female	1
		Karpyk		HV4b	16069T	263G, 315.1C	male	1
Momyn	Tobykty	?		D4e1a	16092C, 16111T, 16213A, 16223T, 16362C	73G, 94A, 146C, 263G, 309.1C, 315.1C	male	1
		Koybas	Zhaush	R8	16357C	73G, 195C, 263G, 309.1C, 315.1C	female	1
				G2a2a	16223T, 16227G, 16278T	195C, 207A, 263G	female	1
		Zhuan-tayak		R7	16189C, 16193.1C, 162223T, 16261T, 16319A, 16362C	73G, 183G, 263G, 309.1C, 315.1C	female	1
				D4t	16223T, 16362C	73G, 309.1C, 315.1C	female	1
	Atygai			D4a6	16093C, 16129A, 16223T, 16362C	73G, 152C, 204C, 217C, 263G, 315.1C	male	1
	Kanzhigaly			U4b1b1c	16235G, 16311C, 16356C	73G, 146C, 152C, 195C, 263G, 309.1C, 315.1C	male	1
				U5a2+16294	16192T, 16270T, 16294T	73G, 263G, 315.1C	male	1
Kerei	?			N9a	16223T, 16257A, 16261T	73G, 150T, 263G, 315.1C	female	1
Ashamaily	?			D4j3	16184T, 16223T, 16265C, 16311C, 16362C	73G, 152C, 263G, 309.1C, 315.1C	female	1
	Iteli	Asha-maily		D4o2a	16093C 16223T 16232T 16290T	73G 195C 210G 263G 309.1C 315.1C	female	1
?	Kosai-batyr			D4s	16173T, 16223T, 16362C	73G, 146C, 199C, 263G, 315.1C	male	1
?	Zhantekei	Bodes		W3	16093C, 16223T, 16270T, 16292T, 16311C	73G, 189G, 194T, 195C, 196C, 204C, 207A, 263G, 309.1C, 310C, 315C	male	1
Konyrat	?			D4e5a	16223T, 16274A, 16362C	73G, 152C, 263G, 309.1C, 315.1C	male	1
	Kylshash			U2e1b	16051G, 16129C, 16182d, 16183C, 16189C, 16256T, 16362C	73G, 152C, 200G, 217C, 263G, 315.1C, 340T	male	1
Koktinuly	Kulshy-gash			C4a1a+195	16129A 16223T 16298C	73G 195C 249d 263G 315.1C	female	1
Kypshak	?			G2a	16223T, 16227G, 16278T, 16362C	73G, 263G, 315.1C	female	1
Zhambai	?			H1+16189	16189C, 16193.2C	263G, 309.2C, 315.1C	female	1
Naiman	?			D4h1	16174T, 16223T, 16362C	73G, 263G, 309.1C	male	1
	Medet			D5	16189C 16223T	73G 150T 152C 263G 309.1C 315.C	female	1
	Tumatai			T	16126C 16294T	73G 152C 263G 309.1C 315.1C	female	1
	?			C4a1+195	16093C 16129A 16223T 16298C	73G 195C 247d 263G 315.1C	female	1
Okiresh	Belgibai	Suinshi	Tolegetai baba-Kytai bi-Karakerei	A5a	16187T, 16223T, 16290T, 16319A	73G, 150T, 235G, 263G, 309.1C, 315.1C	female	1
Tolegetai	Matai	Togyz		Z+152	16185T, 16189.Del(T), 16223T, 16260T,	73G, 152C, 246T, 247.Del(-G), 263G,	female	1

					16288C, 16298C	309.1C, 315.1C		
Uak	?			D5	16189C 16223T	73G 150T 152C 263G 309.1C 315.C	female	1
	?			G2a	16223T, 16227G, 16278T, 16362C	73G, 152C, 263G, 315.1C	male	1
	?			D4j2	16223T, 16291T, 16362C	73G, 263G, 315.1C	female	1
Kishi zhuz								
Baiuly								
Adai	Tazike	Kabakai	Alke	D4t	16129A 16213A 16223T 16298C	73G 195C 198T 247d 309.1C 315.1C	female	1
	?			I1	16129A, 16223T, 16311C, 16391A	73G, 199C, 204C, 250C, 263G, 309.1C, 315.1C	male	1
Salik	?			C4a2a1	16167T, 16171G, 16223T, 16298C, 16327T, 16344T, 16357C	47A, 73G, 249d, 263G, 309.1C, 315.1C	male	1
Alimuly								
Kete (Ak-kete)	?			H1b	16189C, 16193.1C, 16356C, 16363C	249G, 263G, 315.1C	male	1
Shomekei	Kozha-keldi			H1ao	16278T	93G, 143A, 263G, 308.1CT, 315.1C	female	1
Tortkara	?			A+152+16362	16086C, 16223T, 16256T, 16290T, 16319A, 16362C	73G, 152C, 235G, 263G, 309.1C, 315.1C	male	1
Zhetiru								
Zhagalbaily	?			H2a+152+16311	16311C, 16324C	152C, 207A, 263G, 309.1C, 315.1C	male	1
Tama	?			H1e2c	-	73G, 263G, 309.1C, 315.1C	male	1
	?			J2b1c1	16069T, 16126C, 16148T, 16193T, 16261T, 16301T, 16311C, 16319A, 16355T, 16356C, 16362C, 16368C, 16390A, 16399G	73G, 150T, 152C, 263G, 315.1C	male	1
Tabyn	?			D4t	16223T, 16362C	73G, 194T, 279G, 315.1C	male	1
	?			E1a2+(16261)	16147A, 16172C, 16189C, 16223T, 16261T, 16301T, 16311C, 16319A, 16355T, 16356C, 16362C, 16368C, 16390A, 16399G	63C , 73G, 152C, 199C, 204C, 263G, 315.1C	male	1
Non belong to any zhuz								
Tore	?							
	?			D4e1a1	16223T, 16362C	73G, 94A, 263G, 315.1C	male	1
Zhadik auleity	?			U1a3	16182d, 16183C, 16189C, 16249C	73G, 146C, 263G, 285T, 309.1C, 315.1C	male	1
Kozha	?			R2	16037G, 16071T, 16172C	73G, 152C, 263G, 309.1C, 315.1C	female	1
	Aksuiek			B2o	16092C 16182C 16183C 16189C 16217C	73G 263G 315.1C	female	1
Kereyit kozha	?			C4a1a4a	16129A, 16150T, 16223T, 16298C, 16301T, 16311C, 16327T, 16355T, 16356C, 16368C,	73G, 195C, 248d, 263G, 315.1C	female	1

					16390A, 16399G			
Seyit	?			H1e1a4	16311C	263G, 309.1C, 309.2C, 315.1C	female	1
Sunak	?			H2a2a	-	263G, 315.1C	male	1
Nogai-kazakh	?			N9a2'4'5'11	16166G, 16172C, 16223T, 16257A, 16261T, 16304C	73G, 150T, 263G, 309.1C, 315.1C	male	1
	?			U4	16356C	73G, 195C, 263G, 309.1C, 315.1C	male	1

Corresponding to the Cambridge Reference Sequence (CRS) [12, 13], with which all other Human mtDNAs were compared, the D haplogroup was originated from haplogroup HVR D haplogroup has an East Asian origin [14]. It is believed, that mtDNA D haplogroup appeared in Asia 48000 B.C. [15]. The subclade D4 are the principle branch of D. D4 is the most common mtDNA haplogroup among modern populations of northern East Asia, such as Japanese, Okinawans, Koreans, and Mongolic- or Tungusic-speaking populations of northern China [7, 16]. D4 is also the most frequently occurring haplogroup among the Buryats, Khamnigans, Kalmyks, and the Telenghits and Kazakhs of the Altai region [7, 17]. D4 mtDNA branch spread also all over China, Southeast Asia, Siberia, Central Asia, and indigenous peoples of the Americas [18, 19].

H haplogroup is believed to have arisen in eastern Europe or western Asia [20]. There is an opinion, that origin of H clade is associated with Southwest Asia [21], around 20000 to 25000 years ago. Nowaday, the mtDNA haplogroup H is predominantly found in Europe, and is believed to have evolved before the Last Glacial Maximum (LGM). Firstly it expanded in the northern Near East and Southern Caucasus between 33000 and 26000 years ago. Later migrations from Iberia allows to suggest, that the clade reached Europe before the LGM. mtDNA H haplogroup has also spread to parts of Africa, Siberia and inner Asia. Today, around 40% of all maternal lineages in Europe belong to haplogroup H.

It is believed, that mtDNA C haplogroup to have originated somewhere between the Caspian Sea and Lake Baikal some 24000 years B.C. Haplogroup C is spread in Northeast Asia [7], including Siberia. Rissian scientists showed, that, in Eurasia, mtDNA C haplogroup was most frequent among populations of arctic Siberia, such as Yukaghirs and Nganasans [19]. At indigenous peoples of the Americas, the haplogroup C (with subclades C1b, C1c, C1d, and C4c) is one of five mtDNA haplogroups, the others are A, B, D, and X haplogroups [22]. The subclade C1a is found only in population of Asia.

For most mtDNA haplotypes, regional and racial specificity have been established. For an example, in the gene pools of the peoples of Europe and Western Asia, there are mainly 10 major mtDNA haplotypes (HVR *, H, V, J, T, U, K, I, W, X). In the gene pools of Mongoloid populations of Eastern, Central and Nothern Asia another 10 mtDNA haplogroups found (A, B, C, D, E, F, G, Y, Z, M *), and in Negroids - mtDNA of macro group L [2, 4, 7, 23].

Known data about modern Kazakh mtDNA types are contradictory, which may be due to the insufficient amount of collected genetic material for reliable generalization. According to the studies of two different research groups [2, 4], 31.7-45% of mitochondrial haplotypes of the Kazakh ethnos have an "West Eurasian" origin, 50-63.4% have an "Eastern Eurasian" origin, and 0-4.9% are characteristic of the population of India. We should note that in the work of D. Comas and others [2] mtDNA haplotypes of 232 individuals from 12 populations were investigated, among others only 20 were Kazakhs, which is an unrepresentative sample for any conclusions.

More representative population of modern Kazakh (246 individuals with different geographical localization) has been studied by Berezina G.M. with co-authors [3]. According to the results, 58% of Kazakh mtDNAs were haplogroup D (17.9%), C (16%), G (16%), A (3.25%), F (2.44%), presumably Eastern Eurasian. And 41.46% were haplogroups H (13%), T (4.07%), J (4.07%), K (4.07%), U5 (3.25%), I (0.41%), V (0.81%), W (1.63%), presumably of Western Eurasian origin.

In general, in the studied cohort of modern Kazakh, the presence of typically "Asian" mtDNA haplotypes (A5a, A15, A+152+16362, B2o, B4h1, C4, C4a1a+195, C4a1a2, C4a1a4a, C4a2a1, C4b1, C5a, C5+16093, D4a6, D4b2d, D4e1a, D4e1a1, D4e5a, D4j2, D4j3, D4s, D4t, D4+195, D4o2a, D5, E1a2 +16261, F, G2a, G2a+152, G2a2, G2a5, Z+152(M8), M6, M10a1a1b) is estimated as 50%, "European" (H1+16189, H1ao, H1b, H1e1a4, H1e2c, H1e-16129, H2a+152+16311, H2a2a, H2a2a2, H7a1, H13a1d,

HV4b, I1, I1a, J1c16, J1d, J2b1c1, JT, K1c, T1a1, T1a1'3, T2, T, T2b11, U1a3, U1b2, U2e1h, U2e1b, U2e1'2'3', U3a3, U4, U4b1b1c, U5a2+1629, X2, W3, W4) - 38.54%; and ancient groups traced from "Middle East", but originated in Central and South-East Asia (N1a, N1a3a, N1b, N9a N9a1'3, N9a2'4'5'11, R2, R7, R8) - 7.29%; there is even 1 person with an ancient "African" group - L3h1a2a (1.04%).

The presence of such a variety of maternal lines for a small cohort of representatives of the same tribe testifies the Kazakh history full of migrations. That's why we should to mention the malestones in Kazakh ethnic history.

Ethnic history of the Kazakh people is rooted in the ancient period of settling the territory of modern Kazakhstan. The first archaeological finds in the territory of Kazakhstan belong to the Paleolithic period. According to archaeological and paleoanthropological data, the ancient tribes spread on the territory of Kazakhstan since the Bronze Age. During the Bronze Age the ancient population of Kazakhstan was concentrated at the center of a large ethno-cultural region of Eurasia and was one of the representatives of extensive anthropological formation of steppe type proto-European trunk [1]. Nomadic animal husbandry becomes the predominant type of activity to the I millennium BC. The irrigated agriculture, the production of iron and metal manufacture have been developed. Stratification occurred among the nomads, the tribal leaders were indicated, new association of tribes and tribal units appeared, among which the most widely known was Saka Union of tribes. During this period, Savromats tribes settled in western Kazakhstan. At the end of I millennium BC in Kazakhstan there are new tribes: Usuns (from Balkhash to Issyk-Kul lakes), Kangyuy (foothills of Karatau mountains and Syrdarya river basin), Alans (between Aral and Caspian seas).

The statehood on the territory of Kazakhstan established in the I millennium AD. There were areas of sedentary culture: Turk Kaganat (VI cent.), Turgesh and Karluk kaganats (VIII cent.). At the same period the great migration of tribes began from the Altai, southern Siberia and Central Asia to the territory of present-day Kazakhstan and further to the west. The Hun period of "Great Migration" is dated by IV-VII centuries. However, the large-scale movement of the Hun tribes, which led to significant changes in the ethnic and political map of Eurasia, began in I-II centuries. The Huns ancestors - Turkic people "hunnu" lived in the territory of modern Mongolia, Buryatia, and North China.

In X-XI centuries a significant territory of Semirechye was a part of Karakhanid State. Nomadic tribes from south-west and western Kazakhstan entered into an Union of Oguz tribes. In the north-eastern Kazakhstan territory tribal Union of Kimaks was formed. The vast territory of Central Kazakhstan became known as the Kipchaks country or Dasht-i-Kipchak. Start of the XIII century marked by the Mongol invasion and the establishment of the Mongol districts (ulus – Mongolian) in the territory of Kazakhstan. The first Kazakh Khanate (XV-XVI centuries, Semirechye) was a tribal union and included the tribes Uisuns, Kangly, Dulats, Zhalair, Naimans, Argyns and others. These tribes formed a single ethnic array.

Interesting data on paleo-mtDNAs have been presented by C. Lalueza-Fox with co-authors [4]. They studied human teeth 36 of 15 graves located in Kazakhstan, radiocarbon dates vary from XIV-XI centuries BC to the III-V centuries BC. The 21 teeth revealed Caucasoid (Western Eurasian) mtDNA haplotypes, and 6 teeth – the Eastern Eurasian. The authors noticed the dynamics of changes in the haplogroups of the ancient population of Kazakhstan in the direction to the East Eurasia.

Territorially all Kazakh tribes were divided into three major groups ("Zhuzes"): Elder Zhuz (Uly zhuz, 12 tribes), Middle Zhuz (Orta zhuz, 6 tribes), and Junior Zhuz (Kishi zhuz, 3 tribes). Elder Zhuz tribes were mostly occupied South and South-East Kazakhstan, the Middle Zhuz tribes lived in Eastern, Northern and Central Kazakhstan, and the Junior Zhuz tribes traditionally lived in Western Kazakhstan. Also, there are 5 (or 7) tribes which not included to any zhuz.

Obtained data revealed that the modern descendants of ancient Kazakh tribes demonstrate a high degree of heterogeneity in relation to the maternal lines. Because the our studied cohort represents the vast variety of mtDNA haplogroups reflecting the history of ancient nomadic tribes migration we have tried to look after genetic distances between representatives of different tribes combining the data fo tribes unions – Zhuzes.

For this analyses we took into account the mutations in total number of 135 loci in HVR1 and HVR2 regions of mtDNA. Genetic heterogeneity of each locus and cohort (Table 1), the genetic distances and identity (Nei) were calculated in the program GenAlEx 6.2 (table 2).

Table 1 – The heterogeneity characteristics of studied cohorts on HVR1 and HVR2 mtDNA loci

Indexes*	N	Na	Ne	I	h	uh	Polymorphic Loci, %
Cohort	Grand Mean and SE over Loci for each Cohorts						
Uly zhuz	19,000±0	0,889±0,085	1,104±0,016	0,132±0,015	0,075±0,010	0,080±0,010	43,70%
Orta zhuz	55,000±0	1,541±0,073	1,097±0,014	0,144±0,012	0,075±0,008	0,076±0,008	77,04%
Kishi zhuz	11,000±0	0,837±0,085	1,142±0,019	0,164±0,018	0,099±0,012	0,109±0,013	41,48%
Non zhuz	9,000±0	0,533±0,076	1,090±0,015	0,106±0,016	0,064±0,010	0,072±0,011	25,93%
	Grand Mean and SE over Loci						
Total Cohort	23,500±0,80	0,950±0,043	1,108±0,008	0,136±0,008	0,078±0,005	0,084±0,005	47,04±10,75

*Na - number of different alleles; Ne - number of effective alleles; I - Shannon's information index; h – diversity; uh - unbiased Diversity.

Table 2 – Nei Genetic distance and identity between representatives of modern Kazakh based on mutations of variable regions (HVR1 and HVR2) of mtDNA

Pairwise Population Matrix of Nei Genetic Distance // Identity				
Scoring				
0,000 // 1,000				*Uly zhuz*
0,003 // 0,997	0,000 // 1,000			*Orta zhuz*
0,009 // 0,991	0,006 // 0,994	0,000 // 1,000		*Kishi zhuz*
0,006 // 0,994	0,006 // 0,994	0,008 // 0,992	0,000 // 1,000	*Non zhuz*
Uly zhuz	*Orta zhuz*	*Kishi zhuz*	*Non huz*	

Evolutionary connections were defined by the Neighbor-Joining method [24]. The phylogenetic tree was inferred using the UPGMA method and constructed in the MEGA7 program [25]. The figure 3 presents obtained phylogenetic tree. The optimal tree with the sum of branch length = 0.01216667 is shown.

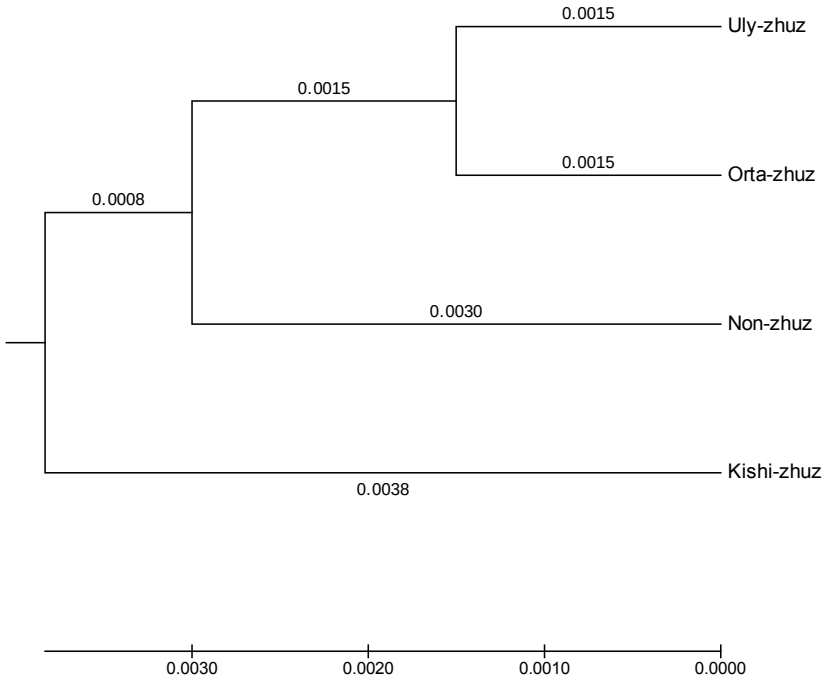

Figure 3 – Evolution relationships of modern Kazakh

The presented data show that the maternal lines of representatives of different zhuzes are characterized by genetic proximity despite of great genetic diversity. The Kishi zhuz maternal lines are more distant from representatives of other tribe unions (GD=0,0038). We suggest that this is due to the lower migration activity of the population of Western Kazakhstan in comparison with other regions of the Republic.

Thus, the analysis of mitochondrial DNA of modern Kazakhs with a wide geographic localization and tribal affiliation shows that the Kazakh's maternal lines are characterized by great diversity of mtDNA haplogroups, reflecting the historical migration of the population of Eurasia with the predominance of "Asian" and "European" components. The analysis of mutations of hypervariable regions of mtDNA regarding Zhuz-affiliation determines the genetic affinity of all Kazakh maternal lines.

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ЭТНИКАЛЫҚ ҚАЗАҚТАРДЫҢ мтДНК БАҚЫЛАУ АЙМАҚТАРДЫҢ ТАЛДАУЫ

Аннотация. Географиялық орналасуына және руына қарай қазіргі заманғы қазақтардың митохондриялық ДНҚ-на жасалған талдау барлық аналар линияларының "азиялық" және "европалық" топтарына қатысты Евразия халықтарының тарихи қоныс аударуын көрсететін мтДНҚ гаплотоптарының әртүрлілігімен сипатталады. Жүзге қатысты мтДНҚ (HVR1, HVR2) гипервариабелді аудандарының мутациясын талдау қазақтардың барлық аналар линияларының генетикалық туыстығын анықтайды.

Түйін сөздер: қазіргі заманғы қазақтар, мтДНҚ, HVR1, HVR2, гаплотоптар.

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АНАЛИЗ КОНТРОЛЬНЫХ РЕГИОНОВ мтДНК У ЭТНИЧЕСКИХ КАЗАХОВ

Аннотация. Проведенный анализ митохондриальных ДНК современных казахов с широкой географической локализацией и родовой принадлежностью свидетельствует, что материнские линии казахов характеризуются большим разнообразием гаплогрупп мтДНК, отражающим исторические миграции населения Евразии с преобладанием «азиатского» и «европейского» компонентов. Анализ мутаций гипервариабельных районов мтДНК (HVR1, HVR2) в отношении жузовой принадлежности определяет генетическую родственность всех материнских линий казахов.

Ключевые слова: современные казахи, мтДНК, HVR1, HVR2, гаплогруппа.

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