



Genetic architecture of a small, isolated Aleut population: Bering Island, Russia

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Introduction

Vitus Bering and his crew discovered the Commander Islands (figure 1) in 1741 on their return journey from the Americas. They were forced to winter on the island after their ship wrecked on its shores, and only 47 of 77 original crew members survived. Bering himself died and was buried on the island. The survivors reached Kamchatka the following summer, and sea otter pelts they brought back with them triggered a rush for furs in the Commander and Aleutian Islands. The Commanders became a provisioning station for Aleutian fur hunting expeditions, which were formalized as the Russian-American Company in 1799.

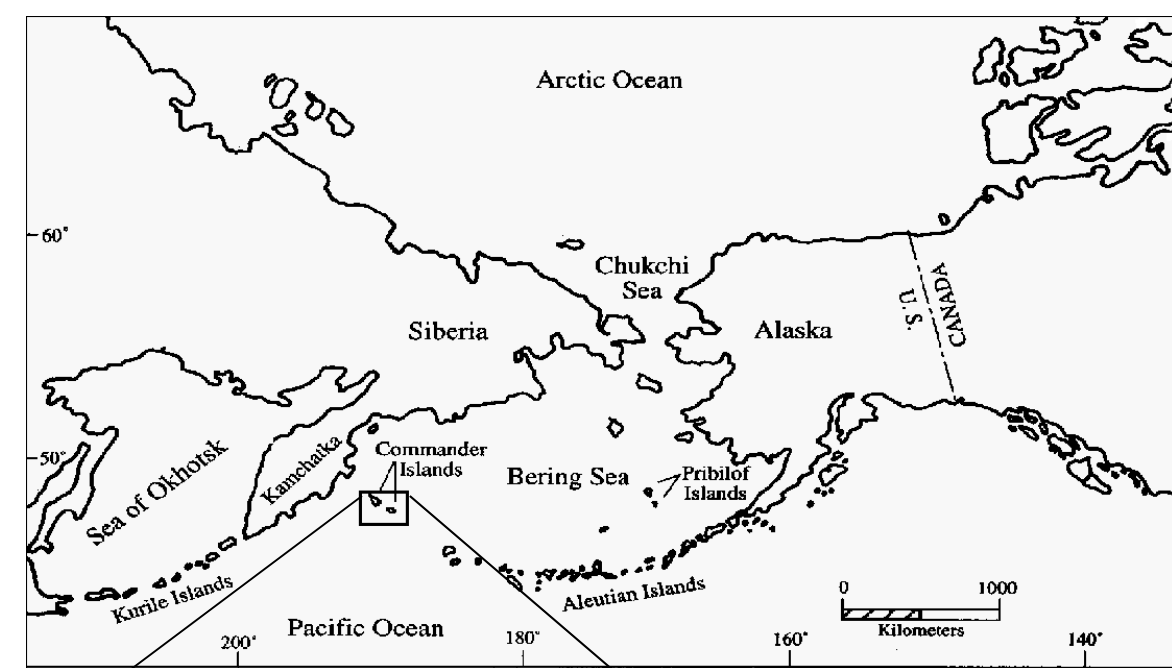
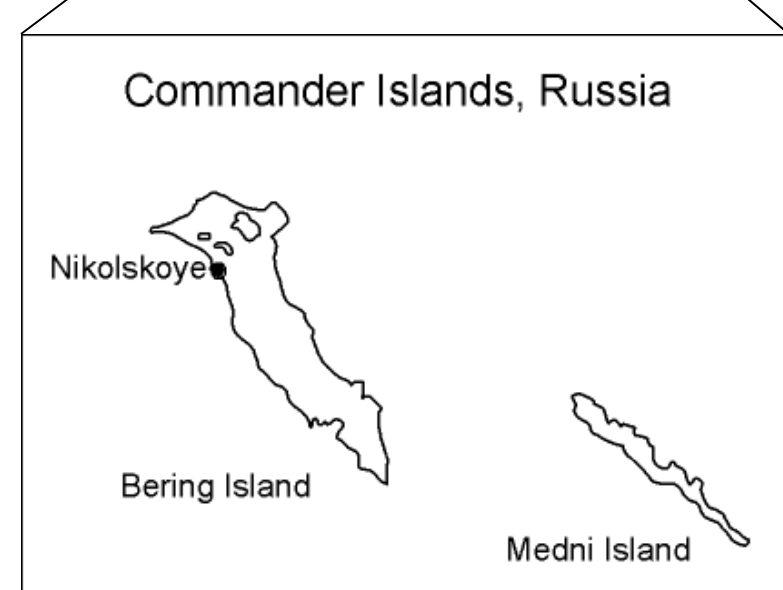


Figure 1 Map of the North Pacific and Commander Islands



Between 1824 and 1828, Aleuts were forcibly relocated from the central and western Aleutian Islands to the Commanders to work for the Russian-American Company. Atka families were relocated to Bering, and Medni was settled by Aleuts from Attu.

In 1825 only 45 Aleuts resided on Bering, in addition to 15-30 original Russian settlers. By 1826 the number of Aleuts increased to 110. The Commander Aleut population sizes fluctuated over time, with an all-time high of 626 (330 on Bering) in 1892. Depopulation due to epidemics and relocations to Kamchatka reduced this number to 364 (204 on Bering) by 1923. In 1969 the Medni Aleuts were relocated to Bering. Today the Bering population consists of approximately 300 individuals.

Project Objectives

1. Compare the genetic makeup of the Aleuts of Bering Island with those from the Aleutian Islands
2. Determine the genetic consequences of unique historic events
3. Characterize the demographic and genetic structures of the Bering Island population

Materials and Methods

Sample collection and Laboratory analysis

Blood samples and questionnaires were collected from 106 individuals residing at the Village of Nikolskoye on Bering Island,

figure 2 (35 Aleut, 41 mixed, 17 Russian, and 13 other), as well as 150 Individuals on Kamchatka (63 Even, 21 Koryak, 49 mixed, 13 Russian, and 4 other) for comparative purposes. DNA was extracted from the samples and characterized using mitochondrial DNA (mtDNA) markers: restriction fragment length polymorphisms (RFLPs) and HVS-I sequencing (see Rubicz et al 2003); Y-chromosome markers: short tandem repeats (STRs) including DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385a/b, DYS438, and DYS439 (using the Y-PLEX 12 Kit from ReliaGene Technologies, Inc); and nuclear markers: STRs, including D3S1358, vWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, D7S820 (using the AmpF/STR Profiler Plus amplification kit from PE Applied Biosystems).



Figure 2 Village of Nikolskoye, Bering Island, Russia

Statistical analysis

mtDNA: 1) To determine the relationship of Aleut haplotypes to each other, gene trees (networks) were generated using reduced median network analysis (Network ver. 4.0, Fluxus Engineering) (Bandelt et al. 1995) from the Aleut HVS-I sequences. 2) In order to look for evidence of population expansion or stability over time, Arlequin ver. 2.00 (Schneider et al. 2000) was used to perform mismatch analysis of the Aleut HVS-I sequences (Rogers and Harpending 1992). **Y-chromosome DNA:** 1) To compare the genetic variation of populations sampled, heterozygosity measures of STR data were generated using Poptree. 2) Population genetic relationships were estimated by converting STR gene frequency data to an R-matrix (Harpending and Jenkins 1973), using the ANTANA program (Harpending and Rogers 1984). **Nuclear DNA:** 1) Heterozygosities were measured (see above). 2) R-matrix analysis was performed (see above). 3) In order to determine parental population contributions to the mixed Bering sample, admixture estimates were generated using Admix 2.0 (Bertorelle and Excoffier 1998).

Results

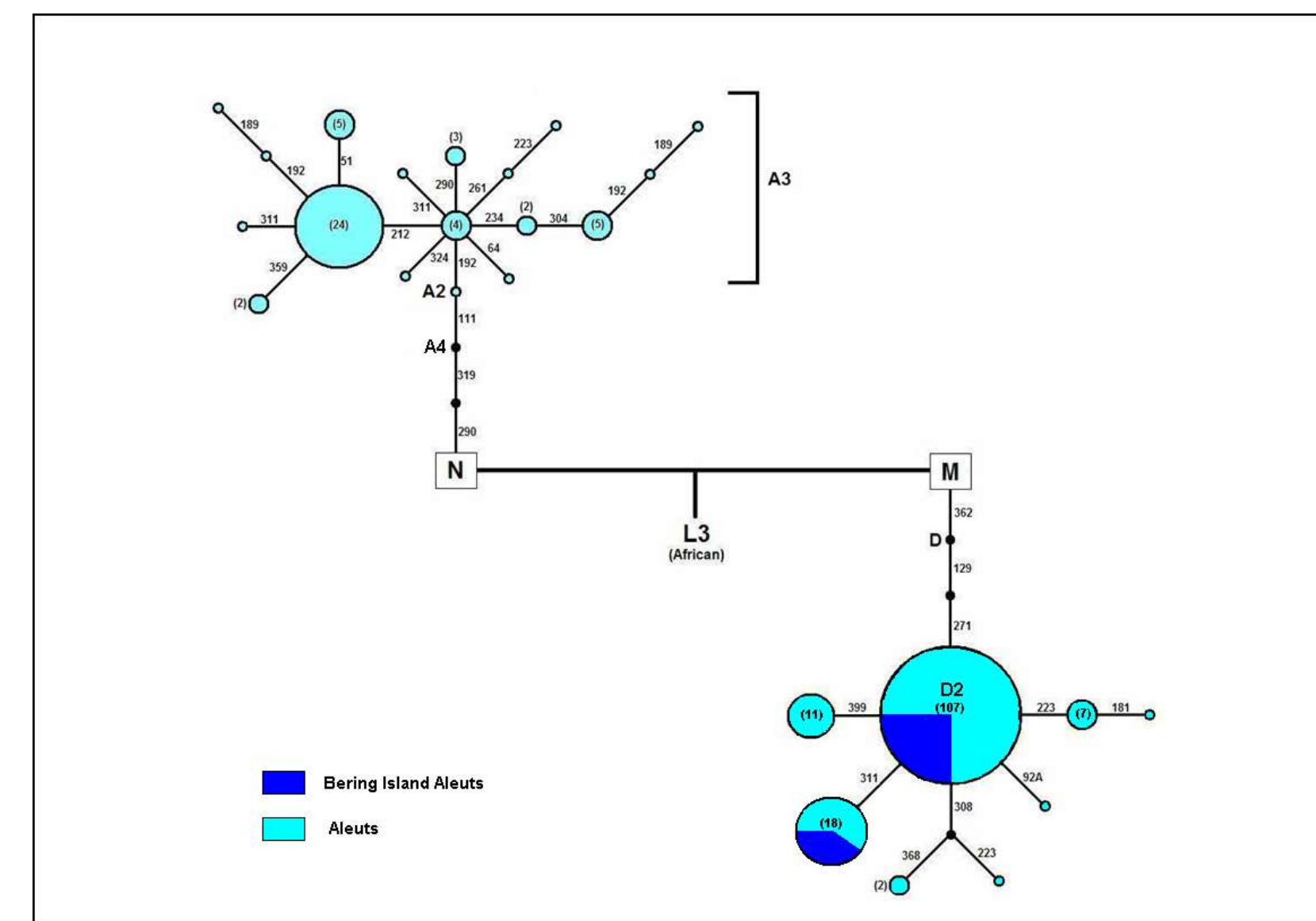
Mitochondrial DNA

Table 1 Mitochondrial DNA haplogroup frequencies

Population	N	Mitochondrial DNA Haplogroups							
		A	C	D	G	H	K	U	Other
Aleut	35	--	--	100%	--	--	--	--	--
Bering Mixed	41	--	7.32%	63.41%	2.44%	4.88%	7.32%	--	14.63%
Russian	30	--	--	--	--	43.33%	26.67%	3.33%	26.67%
Even	63	--	31.75%	19.05%	9.52%	--	--	--	39.68%
Koryak	21	42.86%	38.10%	4.76%	--	4.76%	--	--	9.52%
Kamchatka Mixed	49	14.29%	30.61%	16.33%	6.12%	2.04%	--	--	30.61%
Other	17	5.88%	11.76%	41.18%	--	29.41%	--	--	11.76%

Results show that Bering Aleut mtDNAs exclusively belong to haplogroup D (table 1), similar to the findings of Derbeneva et al 2002. The Bering mixed were 63% D (mainly Aleut); 7% C and 2.5% G, which may be the result of admixture with Kamchatkan groups; and approximately 27% H, K and "other" haplogroups likely representing Russian maternal gene flow into the Aleut population.

Figure 3 Network of Aleut mitochondrial DNA haplotypes



The network (figure 3) demonstrates Bering Aleuts have greatly reduced mtDNA sequence variation in comparison to the Aleut population as a whole. Bering Aleuts completely lack A sequences, which are present at 28% throughout the Aleutians and Pribilofs (Rubicz et al 2003). The Bering D sequences have only one of two sequence motifs, 129A-223T-271C-362C and 129A-223T-271C-311C-362C, which belong to the D2 subhaplogroup defined by Forster et al 1996 and Saillard et al 2000. The lack of diversity among Bering sequences is further demonstrated by the mismatch distribution (figure 4). Sequences from Aleutian and Pribilof Aleuts are bimodally distributed, representing nucleotide differences between A and D haplogroups, and two population expansion events (Zlojutro et al *in press*).

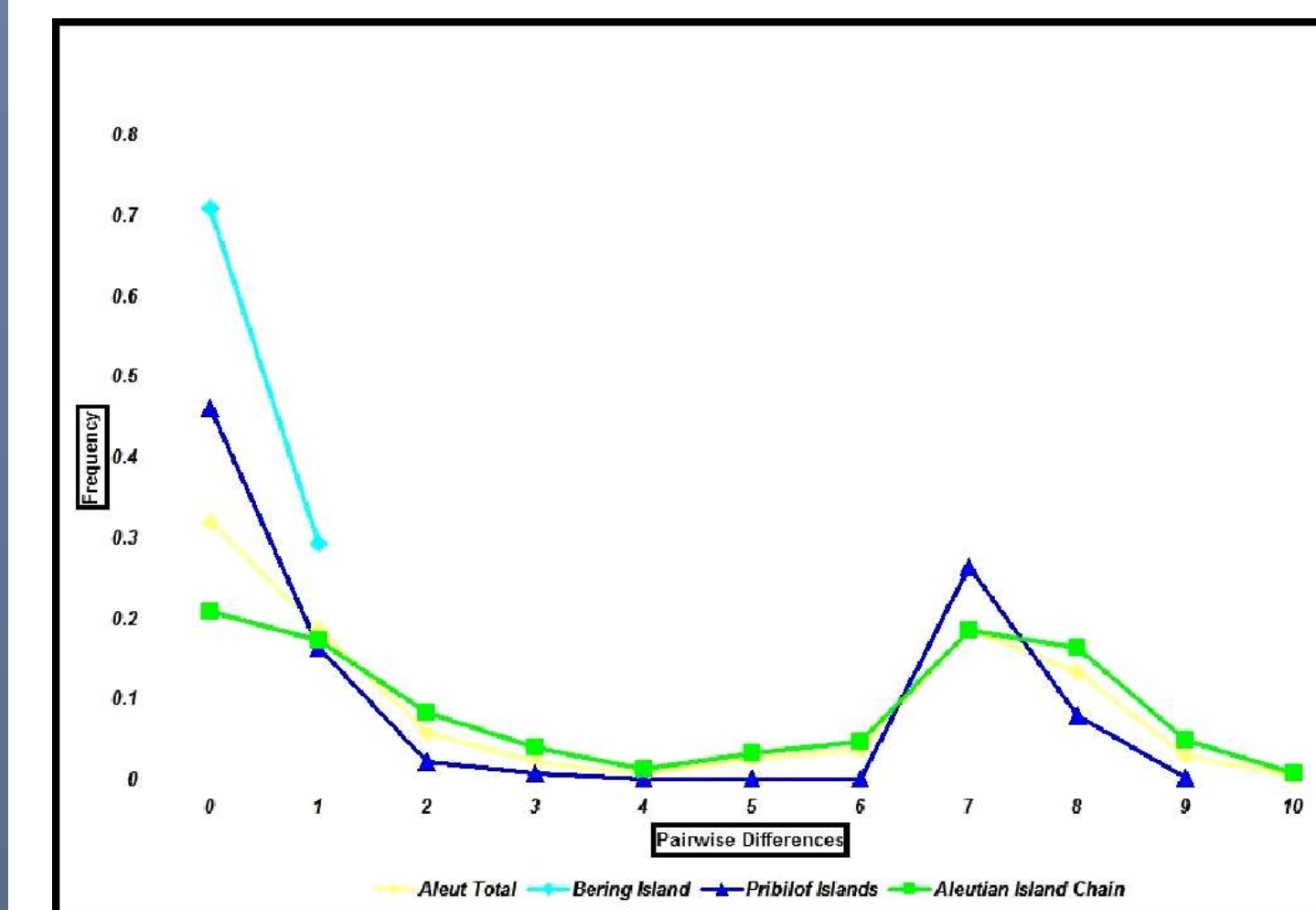


Figure 4 Mismatch distribution of Aleut HVS-I sequences

Y-chromosome DNA

Bering males claiming Aleut ancestry on the paternal side (n=11), had 10 different Y STR haplotypes, one of which was shared with Russians. The Russian males (n=27) had 22 Y haplotypes. Y diversity measures were highest for the Aleuts (H=.68), followed by

Russians (H=.62), Koryaks (H=.55), and Even (H=.32). R-matrix analysis (figure 5) shows the Aleut sample clustering near Russians, rather than native Kamchatkans, likely due to extensive Russian paternal gene flow. This is consistent with preliminary Y-chromosome single nucleotide polymorphism (SNP) analysis, which has identified only one Bering paternal lineage belonging to the Native American Q-M3 lineage.

Figure 5 Y STR R-matrix

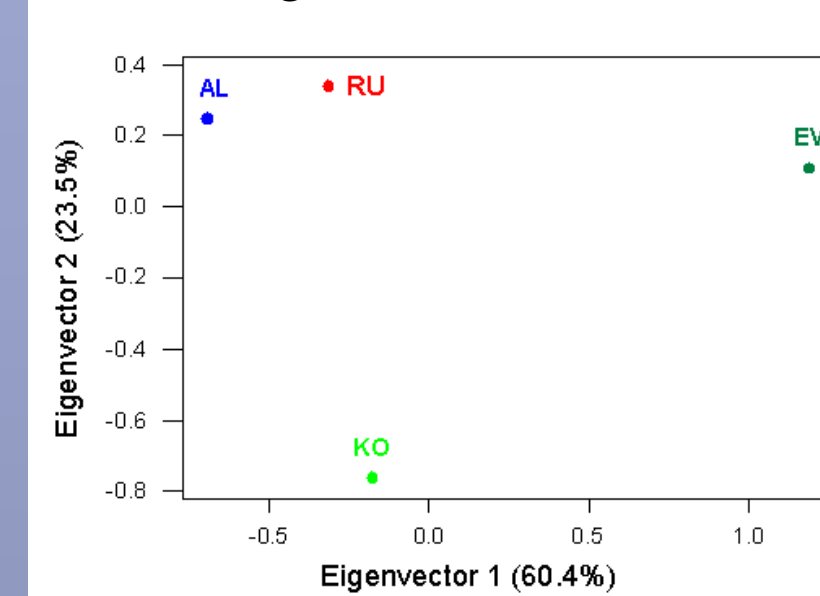
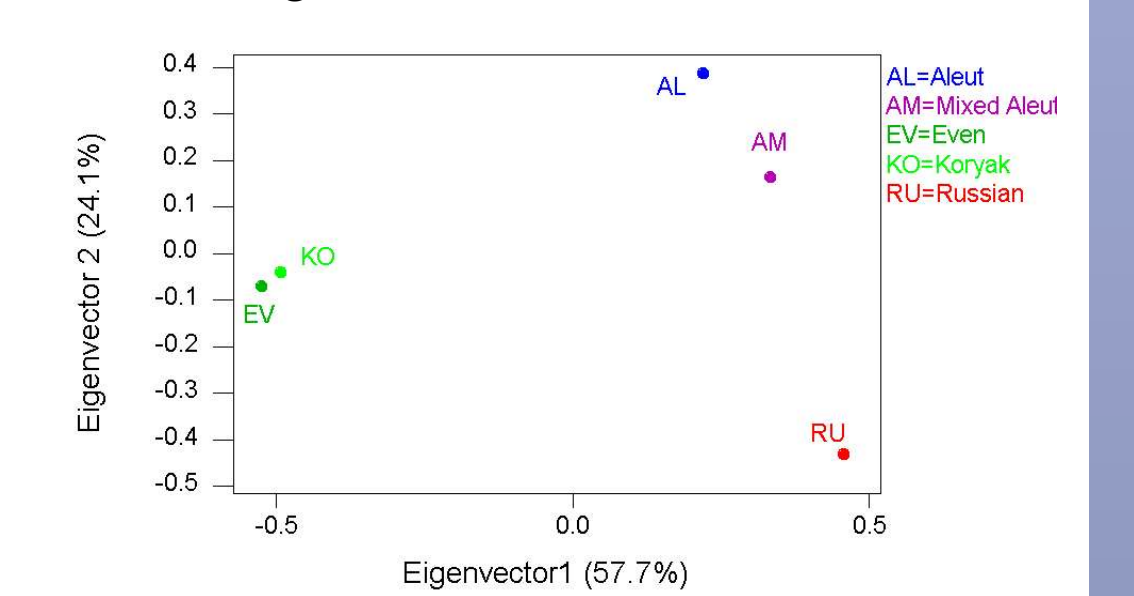


Figure 6 Autosomal STR R-matrix



Autosomal DNA

Analysis of nuclear STRs demonstrated that Bering Aleuts had lower variability (H=0.77) than Russians (H=0.81), or mixed Aleuts (H=0.79), but higher variability than Kamchatkans (Even H=.74, Koryak H=.72). The mixed Aleuts (n=33) were mainly Aleut-Russian. The contribution of parental populations to the mixed Aleuts was estimated at 60.1% Aleut and 39.9% Russian. An R-matrix analysis of these data (figure 6) places the Aleuts closer to Russians than to native Kamchatkan groups (Koryaks and Even). The mixed Aleuts are located between Aleuts and Russians.

Discussion

Analysis of mitochondrial, Y-chromosome, and nuclear markers among Bering Aleuts demonstrates the genetic consequences of the founding of this small, relatively isolated island community. Reduced genetic variability on the maternal side is seen in the fixation of mtDNA D2 lineages in this population. Elevated Bering Aleut Y STR heterozygosity measures, compared to those of surrounding groups, is likely due to substantial non-Aleut paternal gene flow. This is supported by the variation present among nuclear markers, which is lower in Aleuts and increased in mixed Aleuts. The Bering Aleut admixed population is mainly the result of Russian males marrying Aleut females, a policy at one time mandated by the governor. According to Derbeneva et al (2002), the absence of haplogroup A mtDNA lineages among Bering Aleuts is due to the genocide of the natives. However, A lineages are present in approximately 28% of the Aleut population residing in the Aleutian chain and Pribilof islands. It is unlikely that genetic drift would cause the loss of all A lineages over the relatively short time frame of 175 years. A more likely explanation is that absence of Bering A lineages is the result of founder effect, where the original Aleut families brought from Atka and Attu islands had mainly the D2 mtDNA haplotypes.

Acknowledgements

We thank the Aleuts, Russians, Koryaks, and Even for their participation in the project. This research was supported by NSF grants OPP-9905090 and OPP-0327676