



Project
MUSE[®]

Today's Research. Tomorrow's Inspiration.

Genetic Architecture of a Small, Recently Aggregated Aleut Population: Bering Island, Russia

Rohina Rubicz
Mark Zlojutro
Guangyun Sun
Victor Spitsyn

[More](#)

Human Biology, Volume 82, Numbers 5-6, October-December
2010, pp. 719-736 (Article)

Published by Wayne State University Press



 For additional information about this article

<http://muse.jhu.edu/journals/hub/summary/v082/82.5-6.rubicz.html>

Genetic Architecture of a Small, Recently Aggregated Aleut Population: Bering Island, Russia

ROHINA RUBICZ,¹ MARK ZLOJUTRO,¹ GUANGYUN SUN,² VICTOR SPITSYN,³ RANJAN DEKA,² KRISTIN L. YOUNG,⁴ AND MICHAEL H. CRAWFORD⁵

Abstract The fishing community of Bering Island, located in the Russian Commander Islands off the Kamchatka Peninsula, was originally founded by a small number of Russian soldiers and merchants, along with Aleuts forcibly relocated from the western region of the Aleutian archipelago. The purpose of this study is to characterize the genetic variation of Bering Island inhabitants for autosomal, mitochondrial, and Y-chromosome DNA and classic genetic markers and to investigate the genetic impact of the 19th-century founding and subsequent demographic events on this heterogeneous community. Our results show a loss of diversity among maternal lineages in the Bering Aleut population, with fixation of mtDNA haplogroup D, as revealed by median-joining network analysis and mismatch differences. Conversely, paternal haplotypes exhibit an increase in diversity and the presence of a substantial number of non-Native lineages. Admixture results, based on autosomal STR data, indicate that parental contributions to the mixed Aleut population of Bering are approximately 60% Aleut and 40% Russian. Classic genetic markers show affinities between the Bering Island Aleuts and the other historically founded Aleut communities of St. Paul and St. George in the Pribilof Islands, Alaska. This study demonstrates that the opposing evolutionary forces of genetic drift and gene flow acted on the maternal and paternal lineages, respectively, to shape the genetic structure of the present-day inhabitants of Bering Island.

To date, a number of studies have used classic genetic and/or molecular markers to investigate the genetic and evolutionary consequences of geographic isolation on small populations, often reporting dramatic decreases in population genetic diversity that are largely attributed to the random loss or fixation of alleles associated with poorly understood founder events or intergenerational drift. Several

¹Department of Genetics, Southwest Foundation for Biomedical Research, San Antonio, TX 78245.

²Department of Environmental Health, University of Cincinnati Medical Center, Cincinnati, OH 45267.

³Research Center for Medical Genetics, Russian Academy of Medical Sciences, Moscow, 117415 Russia.

⁴Department of Family Medicine Research Division, University of Kansas Medical Center, Kansas City, KS 66160.

⁵Department of Anthropology, University of Kansas, Lawrence, KS 66045.

Human Biology, October–December 2010, v. 82, nos. 5–6, pp. 719–736.

Copyright © 2010 Wayne State University Press, Detroit, Michigan 48201–1309

KEY WORDS: ALEUTS, BERING ISLAND, GENETIC DRIFT, ADMIXTURE, Y-CHROMOSOME HAPLOGROUPS, MTDNA HAPLOGROUPS, AUTOSOMAL STRS, CLASSIC GENETIC MARKERS.

examples include the Cres islanders of Croatia (Jeran et al. 2009), the Andaman islanders (Thangaraj et al. 2003), and the island community of Tristan da Cunha in the South Atlantic (Soodyall et al. 1997, 2003). However, with the exception of Tristan da Cunha, few studies have focused on recently aggregated communities with accurate demographic information available on the origins of the founders and population size fluctuations since their initial establishment, which would allow for more detailed examinations of admixture and the sources of genetic differentiation in small populations.

The Russian Commander islands of Bering and Medni were discovered in 1741 by Vitus Bering and his crew when they were shipwrecked on their return journey from the Americas (Jochelson 1933). They were forced to stay the winter on Bering Island; however, conditions were harsh and Captain Bering died along with many of his crew. The survivors sailed to Petropavlovsk, Kamchatka, the following summer and made a small fortune selling sea otter pelts collected during their expedition; these sales triggered a rush for furs in the Commander and Aleutian islands. Within a few years, *promyshlenniki* (fur hunters) from Siberia returned to the Commander Islands, where they set up provisioning stations for Aleutian fur hunting expeditions (Jochelson 1933; Laughlin 1980). By 1799, the Russian-American Company took control of the region, along with the administration of the native inhabitants of the Aleutian archipelago, the Aleuts (Vanstone 1984).

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 Island, and Medni Island was settled by Aleuts from Attu (Lantis 1984). 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, and the population continued to grow as additional Aleuts were brought from the Pribilof Islands (originally settled by Aleuts from the eastern Aleutian community of Unalaska), the Fox Islands, and Sitka (Rychkov and Sheremetyeva 1972). In 1867 Russia sold Alaska, including the Aleutian Islands, to the United States, effectively isolating the Commander Islands Aleuts from their Aleutian relatives.

The size of the Commander Islands Aleut population has fluctuated over time, partly because of emigration resulting from several relocations. The Commander Islands population reached a maximum of 626 (330 on Bering and 296 from Medni) in 1892. Later, the population decreased because of famine brought about by the Civil War period, diseases, including tuberculosis, and chronic alcoholism. Aleuts were relocated to the Kamchatka Peninsula on two separate occasions and also to islands in the Sea of Okhotsk (Rychkov and Sheremetyeva 1972). However, these relocations were unsuccessful because the majority perished, and the survivors were returned to the Commanders. In 1917 Bering and Medni Aleuts became citizens of the Soviet Union, and with the socialist reorganizations their communities grew as a result of an influx of peoples from other regions (Liapunova 1996; Rychkov and

Sheremetyeva 1972). The Commander Islands Aleut communities were consolidated in 1969, when Medni Aleuts relocated to Bering. Today the Aleut population of Bering consists of approximately 300 individuals.

Several studies of the genetics of Bering Island Aleuts have been published previously. Rychkov and Sheremetyeva (1972) presented blood group and serum protein data for 60 Commander Islands Aleut participants. They found that for the ABO and MNS blood groups, the Aleuts were similar to Eskimo populations in having high frequencies of the *ABO*O*, *ABO*A*, and *MNS*Ms* alleles. Like other Native Americans, they lacked the Rh- phenotype and had high frequencies of the *cDE* and *CDe* alleles. The Commander Islands Aleuts had a high frequency of the *DI*A* gene of the Diego blood group system, which although unusual for North Americans, is characteristic of South American populations. Aleuts had nearly equal frequencies of the haptoglobin genes *HPA*1* and *HPA*2*, similar to American Indian groups such as the Haida, Apache, and Assiniboin. Later, in the summer of 2001, Sukernik collected blood samples from 30 individuals on the Island of Bering, and they were analyzed for mtDNA sequence variation (Derbeneva et al. 2002). All the sequences were characterized to the D2 subhaplogroup, with most belonging to the Aleut-specific clade D2a1a (Gilbert et al. 2008; Zlojutro et al. 2009), thus exhibiting a striking absence of genetic diversity.

In our study we use both molecular (mitochondrial, Y-chromosome, and autosomal DNA) markers and classic genetic markers to characterize the genetic structure of the historically established Aleut community of Bering Island, Russia, and to make comparisons with its parental populations. Specific objectives of this study are (1) to use molecular markers to characterize the genetic composition of the Bering Island community; (2) to compare the genetics of the Bering Island Aleut population with other Aleut groups residing in the Aleutian archipelago, their ancestral homeland; and (3) to determine the genetic consequences of unique historical events, including genetic drift and admixture, on this population.

Materials and Methods

During the summer of 2001 researchers from the University of Kansas and a team from the Russian Academy of Medical Sciences, Moscow, collected blood samples and genealogical information from 106 individuals from the village of Nikolskoye, Bering Island (Figure 1). Permissions for this study were obtained from the University of Kansas Advisory Committee on Human Experimentation (ACHE), the Russian Academy of Sciences, and tribal councils for each community, and participants signed informed consent statements. The sample included 35 self-identified Aleuts, 41 admixed individuals, 17 Russians, and 13 individuals of other ethnic backgrounds. An additional 150 samples were collected from the residents of Esso and Anavgai on the Kamchatka Peninsula for comparative purposes. Of these participants, 63 were Even, 21 were Koryak, 49 were mixed, 13 were Russian, and 4 were of other ethnicities.

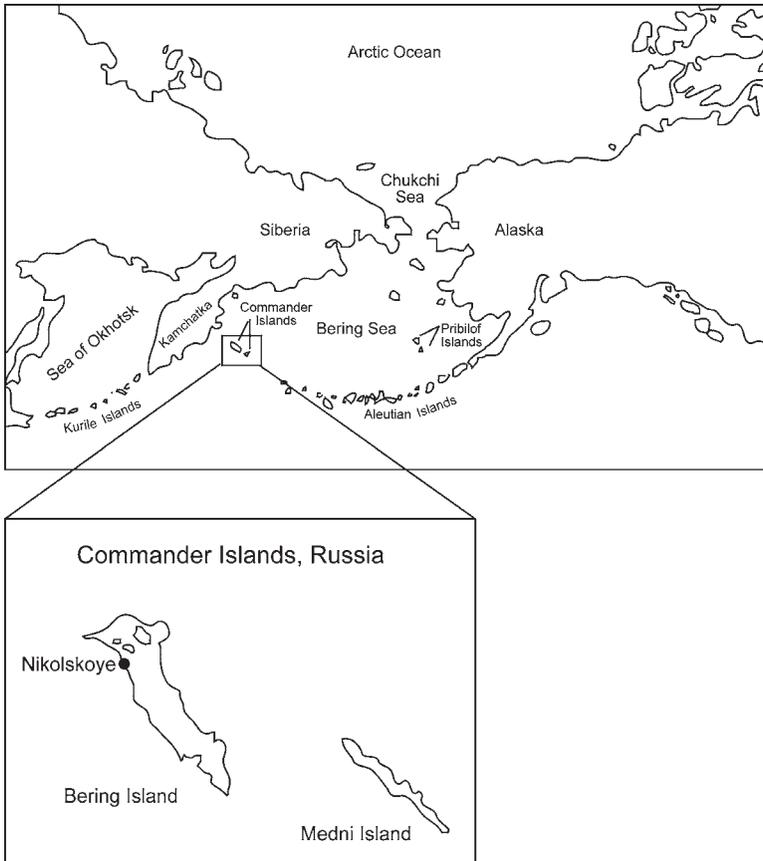


Figure 1. Map of Bering Island, Russia.

DNA Analysis. DNA was extracted from blood samples using the SuperQuik Gene DNA Extraction kit (University of Kansas). Mitochondrial DNA (mtDNA) restriction fragment length polymorphisms (RFLPs) were characterized for markers defining Native American, Siberian, and/or European haplogroups A (+*Hae*III 663), B (COII-tRNA^{Lys} 9-bp deletion), C (–*Hinc*II 13259, +*Alu*I 13262), D (–*Alu*I 5176), G (+*Hae*II 4830, +*Hha*4831), H (–*Alu*I 7025), K (–*Hae*II 9052), and U (+*Hinf*I 12308). In addition, the mtDNA first hypervariable region (HVS-1) was sequenced for np 16000–16400 [sequence results published by Zlojutro et al. (2006)].

Y-chromosome analysis was performed on 11 commonly used short tandem repeats (STRs): *DYS19*, *DYS389I*, *DYS389II*, *DYS390*, *DYS391*, *DYS392*, *DYS393*, *DYS385A*, *DYS385B*, *DYS438*, and *DYS439*, available in the Y-PLEX 12 Kit from ReliaGene Technologies Inc. (Rubicz et al. 2010). The following

single nucleotide polymorphisms (SNPs), defining Y-chromosome haplogroups, were characterized for the male samples: P39 (haplogroup C), P2 (haplogroup E), M170 (haplogroup I), 12f2 (haplogroup J), M231 (haplogroup N), P36 (haplogroup Q), and M269 and SRY 10381b (haplogroup R).

Autosomal STRs analyzed for the samples included *D3S1358*, *VWA*, *FGA*, *D8S1179*, *D21S11*, *D18S51*, *D5S818*, *D13S317*, and *D7S820*, using the AmpFI-STR Profiler Plus amplification kit and corresponding protocol designed by PE Applied Biosystems (Rubicz et al. 2010).

Analytical Methods. Median-joining network analysis (Network, version 4.0, Fluxus Engineering) was performed using 35 Bering Aleut mtDNA HVS-1 sequences and an additional 198 sequences from Aleuts residing in the Aleutians, Pribilofs, and Anchorage (Bandelt et al. 1999; Rubicz et al. 2003; Zlojutro et al. 2006). This was done to determine the evolutionary relationships of Aleut haplotypes to each other and to identify mtDNA differences between Bering Aleut haplotypes and those of other Aleuts from Alaska. This analysis generates gene trees or networks of mutation pathways, with different haplotypes represented by individual nodes that are proportional in size to the number of carriers. The median-joining algorithm is capable of resolving mutation parallelisms and reversals between haplogroups and incorporates any parallelisms that are not resolved as reticulations, thereby generating networks that subsume the most parsimonious pathways. Networks for haplogroup A and D sequences were generated separately and then joined together based on reported phylogenies of East Asian HVS-1 sequences (Kivisild et al. 2002; Zlojutro et al. 2006). The Aleut HVS-1 sequences were also used to construct a distribution of pairwise sequence differences (i.e., mismatch distribution) using Arlequin, version 2.00 (Schneider et al. 2000), which was used to identify signatures of population expansion or stability over time (Rogers and Harpending 1992).

To further compare the genetic variation among the populations sampled, including Bering Aleuts, mixed Aleuts, Russians, Evens, and Koryaks, we computed heterozygosity levels for autosomal STRs using the program DISPAN (Ota 1993). The genetic relationships among the study populations were visualized using the *R* matrix method (Harpending and Jenkins 1973); autosomal and Y-chromosome STR frequency data were converted to variance-covariance matrices of genetic similarity and dissimilarity between populations using the ANTANA program (Harpending and Rogers 1984), and the matrices were projected into two-dimensional space by means of principal components analysis. *R* matrix analysis was also performed on classic genetic data obtained from the literature (Crawford and Enisco 1982; Crawford et al. 1981; Majumder et al. 1988; Rychkov and Sheremeteyeva 1972) for Commander Island Aleuts (both Bering and Medni communities) and comparative populations (including Aleuts from St. Paul and St. George in the Pribilofs). A plot of allele scores for the classic genetic data was created to assess the relative contribution of allele variables to the *R* matrix ordination results of the populations.

Table 1. Mitochondrial DNA Haplogroups

Population	mtDNA Haplogroup								Other (%)
	N	A (%)	C (%)	D (%)	G (%)	H (%)	K (%)	U (%)	
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	–	–	9.5
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

To determine whether any of the populations in this study had experienced gene flow or genetic drift, we regressed the population mean heterozygosities on genetic distances from the centroid (r_{ii}) using autosomal STR data. According to Harpending and Ward (1982), heterozygosity and distance from the centroid are expected to have a linear relationship, and deviations from this can be explained by the operation of one of the two previously mentioned forces of evolution. Finally, to quantify the relative contributions of parental populations (Aleut and Russian) to the mixed Bering sample, we calculated admixture estimates using the Admix 2.0 program (Bertorelle and Excoffier 1998) for autosomal STR haplotypes.

Results

Mitochondrial DNA. The results of the mtDNA RFLP analysis are presented in Table 1. Of the five major founding mtDNA lineages identified among Native American populations (haplogroups A, B, C, D, and X), most of the Aleut populations carry only a subset of these: A and D. Furthermore, the Aleuts of Bering Island exhibit only a single mtDNA lineage, fixed for haplogroup D. Our findings are consistent with those from a study by Derbeneva et al. (2002) that examined the Commander Islands Aleut population. The Bering Aleuts differ from Aleut populations in the Aleutian Islands, where haplogroups D and A are present at frequencies of 72% and 28%, respectively, among the inhabitants of the central and western communities (Rubicz et al. 2003). In contrast, eastern Aleut communities exhibit high frequencies of haplogroup A, ranging from 49% to 73%, in addition to low frequencies of other haplogroups, indicating a small amount of non-Aleut maternal gene flow into these communities (Zlojutro et al. 2009).

The Aleut haplogroup patterns are unusual for Native populations of North America, where haplogroup D is normally absent and haplogroup A is usually present at high frequencies. For example, most Eskimo populations have low frequencies of haplogroup D and high frequencies of haplogroup A mtDNA types, with haplogroup A reaching near fixation in the Eskimos of Greenland (Helgason

et al. 2006; Saillard et al. 2000). Likewise, Athapaskan populations also have high frequencies of haplogroup A.

Haplogroup B is either absent or present at low frequencies among many North American populations, with exceptions including populations from the Southwest, Great Basin, and the Columbia Plateau. Haplogroup C is present at low frequencies among some North American groups and is found at substantial frequencies among the populations of Kamchatka. In the Americas, haplogroup X is restricted to northern populations, where it is present at low frequencies.

Overall, the Native American haplogroups are considered a subset of the mtDNA variation present in Asia and can be further broken down into American-specific subhaplogroups (A2, B2, C2, and D1) and several minor lineages (C4c, D2a, D3, and X2a) (Achilli et al. 2008; Bandelt et al. 2003; Brown et al. 1998; Fagundes et al. 2008; Lorenz and Smith 1996; Merriwether et al. 1995; Schurr and Wallace 1999; Tamm et al 2007; Torroni et al. 1993, 1994).

The Bering Island mixed sample has haplogroups that are 63% haplogroup D, suggesting that their maternal lineages are mainly of Aleut ancestry; 7% haplogroup C; 3% haplogroup G, which may be the result of admixture with Kamchatkan groups (Koryaks and Even); and approximately 27% haplogroups H, K, and "other" haplogroups that likely represent Russian maternal gene flow into the Aleut population. The Russian mtDNAs are characterized by high frequencies of haplogroup H (43%), haplogroup K (27%), and other as yet undetermined haplogroups (27%), with a small frequency of haplogroup U (3%). The Koryaks and Evens have elevated frequencies of haplogroup C mtDNAs (38% and 32%, respectively), with the Koryaks also displaying a high frequency of haplogroup A (43%).

The network based on mtDNA sequences (Zlojutro et al. 2006) presented in Figure 2 demonstrates that Bering Aleuts have greatly reduced mtDNA sequence variation in comparison to the Aleut population as a whole. Not only do Bering Aleuts completely lack haplogroup A sequences, but their D haplotypes consist of only two sequence motifs (16129A-16223T-16271C-16362C and 16129A-16223T-16271C-16311C-16362C), both of which belong to subhaplogroup D2 defined by Forster et al. (1996) and Saillard et al. (2000) as 16129A-16223T-16271C-16362C. Aside from Aleuts, subhaplogroup D2 is mainly found in Eskimo and Chukchi populations, although it was also reported among the Apache (Torroni et al. 1993). In contrast to the reduced mtDNA diversity observed in the Bering Aleuts, 26 mtDNA haplotypes (18 different haplogroup A sequences and 8 different haplogroup D sequences) were observed in the Aleut sample ($n = 163$) from the Aleutian and Pribilof islands (Rubicz et al. 2003). As is evident from the network, all these sequences belong to subhaplogroups A2 and D2 (Zlojutro et al. 2006). Subhaplogroup A2 is defined by the mutation motif 16111T-16223T-16290T-16319A-16362C and is considered a founder lineage of Native Americans that is also present among the Chukchi and Siberian Eskimos of Chukotka (Shields et al. 1993; Starikovskaya et al. 1998). Within subhaplogroup A2, most Aleut sequences also have the 16192T mutation, which defines the A2a1

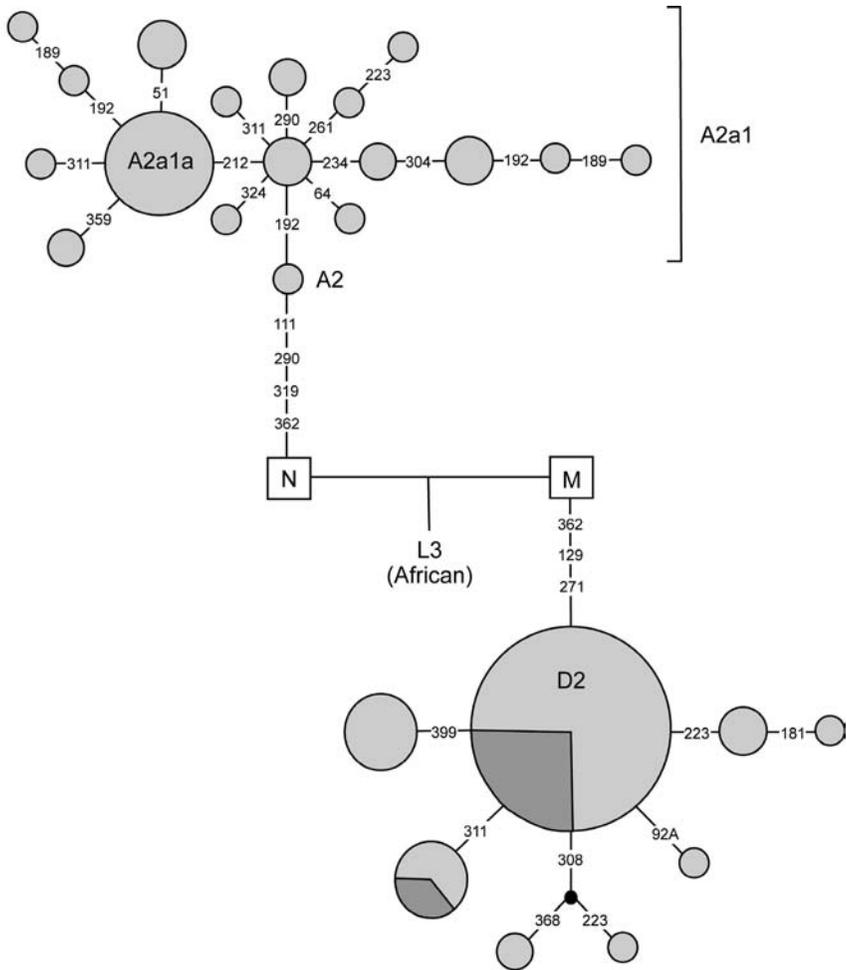


Figure 2. Network analysis of mtDNA HVS-1 sequences. Dark gray, Bering Island Aleuts; light gray, Aleuts from the Aleutian Islands, Pribilof Islands, and Alaska Peninsula.

subhaplogroup of Beringian and Na-Dene populations. The network of Aleut sequences shows three starlike clusters, one centered over the D2 root, one over the A2a1 root, and the last within the Aleut-specific subclade A2a1a defined by the mutation 16212A. According to Zlojutro et al. (2006), the A2a1 cluster represents an earlier expansion event of Beringian and/or northern North American populations and the D2 and Aleut-specific A2a1a mutation clusters derive from a later expansion event that involved ancestral populations of modern-day Aleuts.

The mismatch analysis (Figure 3) underscores the low levels of mtDNA diversity in Bering Aleuts compared to the larger Aleut population. The distribution

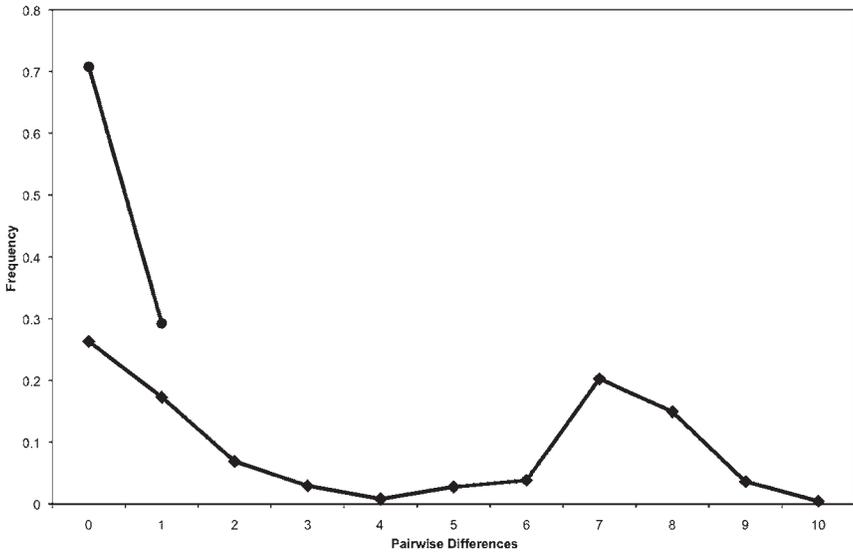


Figure 3. Mismatch analysis based on mtDNA HVS-1 sequences. Circles, Bering Island Aleuts; diamonds, non-Bering Island Aleuts.

of sequence mismatches for non-Bering Aleuts is bimodal, with peaks at 0 and 7 pairwise differences. Although multimodality can be interpreted as population stability over time (Rogers and Harpending 1992; Slatkin and Hudson 1991), Zlotjutro et al. (2006) argued that when interpreted in conjunction with the three star-like clusters identified in the network analysis, the distribution contains signatures of two separate expansion events. Dating of the Aleut lineages provides expansion times of approximately 5400 BP for the A2a1a and D2 lineages and 19,900 BP for the A2a1 cluster. In contrast, the Bering Aleut sequences have only a single peak positioned at 0 pairwise differences, representing the difference between the two D2 sequence motifs characterized for that community.

Y-Chromosome Markers. Bering males claiming Aleut ancestry on the paternal side ($n = 11$) had 10 different Y-chromosome STR haplotypes, one of which was observed among the Russian samples (Rubicz et al. 2010). Y-chromosome SNP analysis (Table 2) indicates that these STR lineages belong to the following Y chromosome haplogroups: 27% to Native American haplogroup Q; 45% to European haplogroups I, J, and R; 18% to Eurasian haplogroup N; and 9% to other as yet undetermined haplogroup categories.

The Russian males ($n = 27$) exhibit 22 Y-chromosome haplotypes, belonging to haplogroups I and R (53%), haplogroup N (33%), the (predominantly) Asian haplogroup C (7%), and other lineages (7%). The Even males ($n = 10$) harbor six different haplotypes, belonging to haplogroups C (90%) and N (10%). Likewise,

Table 2. Y-Chromosome Haplogroups

Population	N	Y-Chromosome Haplogroup							
		C (%)	E (%)	I (%)	J (%)	N (%)	Q (%)	R (%)	Other (%)
Aleut	11	–	–	9.09	27.27	18.18	27.27	9.09	9.09
Mixed Aleut	6	–	33.33	16.67	–	–	–	33.33	16.67
Russian	15	6.67	–	20.00	–	33.33	–	33.33	6.67
Even	10	90.00	–	–	–	10.00	–	–	–
Koryak	11	27.27	–	9.09	–	45.46	–	9.09	9.09
Other	6	–	16.67	16.67	–	–	16.67	33.33	16.67

six different Y-chromosome STR haplotypes were identified in the Koryaks ($n = 11$), of which 27% belong to haplogroup C, 18% to haplogroups I and R, 45% to haplogroup N, and 9% representing other patriline. Males of other ethnicities ($n = 6$) had Y chromosomes belonging to haplogroups I and R (50%), European and African haplogroup E (17%), Native American haplogroup Q (17%), and other haplogroups (17%). These results indicate that there was substantial non-Aleut paternal gene flow into the Bering community.

The R matrix analysis of the Y-chromosome STRs indicates a closer relationship of the Bering Aleuts to Russians rather than to the native Kamchatkans. Figure 4 is a plot of the first two principal components based on an R matrix of the four populations. The first eigenvector (representing 60% of the total variation) separates the Evens from the Aleuts, Russians, and Koryaks, and the second eigenvector (accounting for 24% of the variation) separates the Aleuts and Russians from the Koryaks. This grouping of the Bering Aleuts with Russians is likely due to extensive Russian paternal gene flow into the Bering community that may have been less substantial among the Evens and Koryaks.

Autosomal STRs. The analysis of autosomal STRs (see Rubicz et al. 2010) demonstrates that Bering Aleuts have lower genetic variability ($H = 0.77$) than the Russians ($H = 0.81$) or the Bering mixed Aleuts ($H = 0.79$) but higher variability than the Kamchatkans (Even $H = 0.74$ and Koryak $H = 0.72$). The mixed Aleut sample ($n = 33$) is mainly of Aleut-Russian ancestry. Using the Admix 2.0 program on the autosomal STR data, we estimate that the contribution of the parental populations to the mixed Aleut nuclear gene pool is 60.1% Aleut and 39.9% Russian. This result differs from previous admixture estimates obtained for an Aleut sample collected from the historically established community of St. Paul in the Pribilof Islands, Alaska. Based on the analysis of Merriwether et al. (1995), in addition to haplogroups A and D, which are characteristic of Aleuts, haplogroup C and other undetermined mtDNA lineages can be identified in the St. Paul sample. Rubicz et al. (2003) argued that the C haplogroups were likely the result of maternal gene flow from Athapaskans (with an estimated frequency of 10.5%), with the unclassified mtDNAs most likely being of European ancestry (0.07%). However,

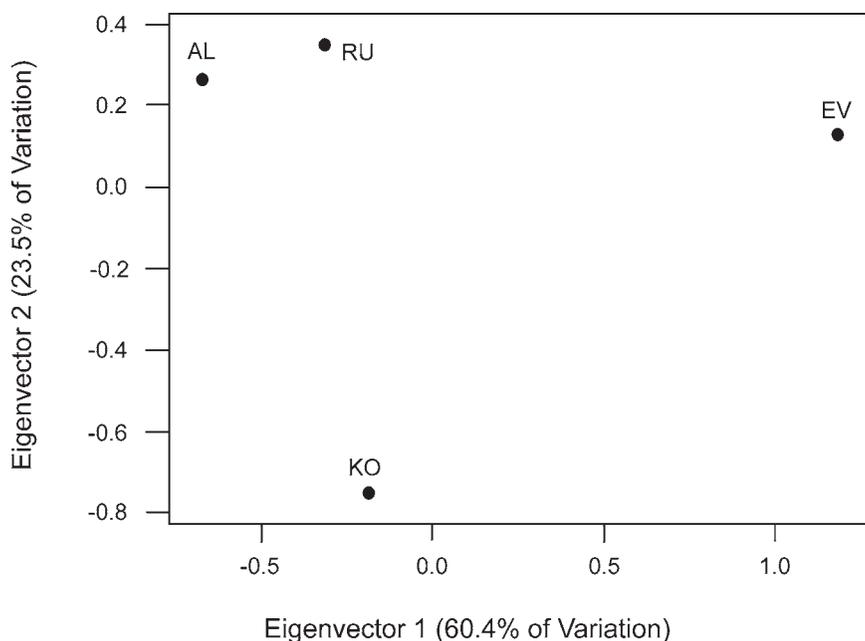


Figure 4. *R* matrix graph of populations (Y-chromosome STRs). AL, Aleut; EV, Even; KO, Koryak; RU, Russian.

these additional haplogroups were not found in a sample of the contemporary populations from the western and central Aleutians (Rubicz et al. 2003), for which genealogical information was carefully collected, nor in the ancient Aleut samples analyzed by Hayes and O'Rourke (2000).

An *R* matrix analysis of the autosomal STR data is presented in Figure 5. The first eigenvector, representing 58% of the total variation, separates the Aleuts, mixed Aleuts, and Russians from the Kamchatkan groups (Koryaks and Evens). The mixed Aleuts, as expected based on the admixture estimate, are located between the Aleuts and Russians, positioned closer to the Aleuts. In this analysis the Koryak and Even populations cluster together. According to Figure 6, the mixed Aleut population has experienced the greatest amount of gene flow, whereas the Koryaks are the most isolated of the populations, likely experiencing genetic drift.

Classic Genetic Markers. The *R* matrix analysis of classic genetic markers (Figures 7 and 8) consists of nine alleles representing five blood group systems for both the Bering and Medni (Commander Islands) populations [these data were collected by Rychkov and Sheremetyeva (1972)], the historically established Pribilof Islands Aleut communities of St. Paul and St. George [published by Majumder et al. (1988)], and comparative Eskimo and Chukchi populations.

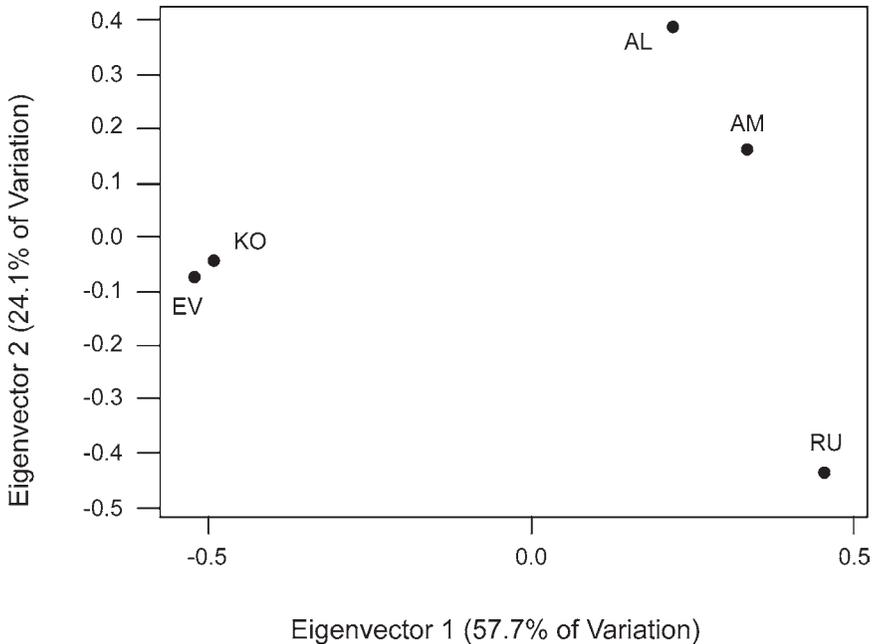


Figure 5. *R* matrix graph of populations (autosomal STRs). AL, Aleut; AM, mixed Aleut; EV, Even; KO, Koryak; RU, Russian.

The Bering, Medni, and St. George Aleuts all cluster together, and along the first eigenvector (accounting for 37% of the total variation) they are closest to the Eskimo populations of Savoonga, King Island, Wales, and Gambell. The St. Paul Aleuts appear to be distinct from the other Aleuts and genetically are most similar to the Siryeniki Chukchi and Kodiak Eskimos from Larson Bay. The Siryeniki Eskimos and Noonyamo Eskimos are genetically most divergent from the other populations, when both eigenvectors are taken into account (for a total of 71% of the variation present).

Discussion

Analyses of molecular and classic genetic markers among Bering Aleuts demonstrate the genetic consequences of the founding of this small, geographically isolated island community. On the maternal side genetic variability is dramatically reduced, as seen by the presence of only two mtDNA haplotypes in the Bering Aleuts, compared to the 26 mtDNA lineages that have been identified in Aleut communities from Alaska (Rubicz et al. 2003; Zlojutro et al. 2006, 2009). According to Derbeneva et al. (2002), this fixation of the mtDNA D2 lineages and absence of haplogroup A mtDNA lineages among Bering Aleuts is due to the

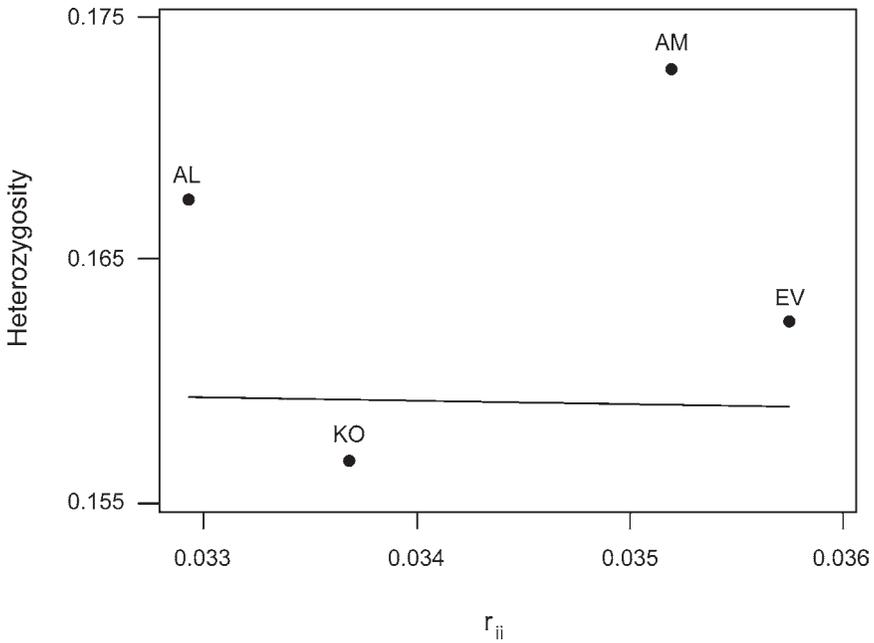


Figure 6. Heterozygosity versus distance from centroid (autosomal STRs). AL, Aleut; AM, mixed Aleut; EV, Even; KO, Koryak.

“genocide” of the natives. However, haplogroup A is present in approximately 28% of the Aleut population residing in the Aleutian chain and Pribilof Islands and in more than 70% among some eastern Aleut groups (Zlojutro et al. 2009). Thus it is unlikely that the decimation of the Aleut people by the Russians during the 18th and 19th centuries would have in any way selectively removed haplogroup A from the Commander Islands. Nor is it likely that intergenerational drift would cause the complete loss of haplogroup A lineages over the relatively short time frame of 175 years, or approximately eight generations, especially given the Bering Aleut effective reproductive size (N_e) of 81 (Rychkov and Sheremetyeva 1972). This number is intermediate to the N_e values estimated for other Aleut communities, which range from 127 for Unalaska Aleuts to 31 for Pribilof Aleuts (where both haplogroup A and D mtDNA lineages persist). A far more likely explanation is that the absence of Bering haplogroup A lineages is the result of a founder effect; in other words, the original Aleut families brought from Atka and Attu islands had a disproportionately high frequency of haplogroup D2 mtDNA haplotypes.

In contrast to the lack of diversity observed for the maternal markers, Bering Aleut paternal and autosomal markers show an increase in variation. Based on the Y-chromosome haplogroup data, non-Aleut male gene flow into the Bering Island community was substantial. Both the self-reported “Aleut” and “mixed

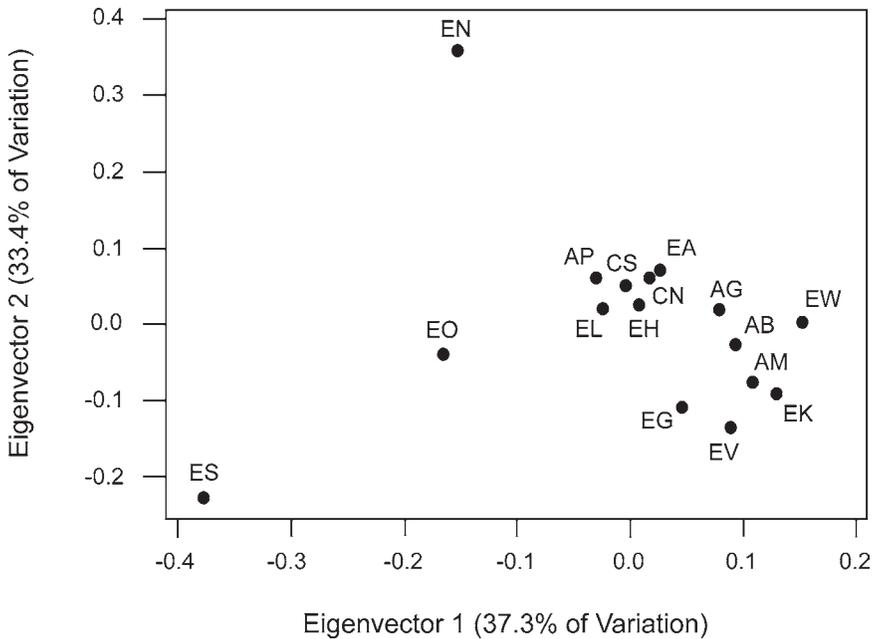


Figure 7. *R* matrix for populations based on classic genetic markers. AB, Bering Aleut; AG, St. George Aleut; AM, Medni Aleut; AP, St. Paul Aleut; CN, Noonyamo Chukchi; CS, Siryeniki Chukchi; EA, Kodiak Eskimo (Akhlok); EG, Gambell Eskimo; EL, Kodiak Eskimo (Larson Bay); EN, Noonyamo Eskimo; EO, Kodiak Eskimo (Ouzinikie); ES, Siryeniki Eskimo; EV, Savoonga Eskimo; EW, Wales Eskimo; EH, Kodiak Eskimo (Old Harbor); EK, Eskimo (King Island).

Aleut" groups are characterized by a large non-Native male component of approximately 73% and 100%, respectively. The Bering Aleut admixed population is mainly the result of Russian males marrying Aleut females, a marriage pattern that was at one time encouraged by the governor of Bering officially as a way of increasing fertility. Although there were concerns about the potential for inbreeding and decreased fitness levels, this practice also served to control the population. The Russian component of the Bering mixed Aleut population is approximately 40%, based on admixture analysis, and the diversity measurement of their autosomal markers is slightly elevated compared to the Bering Aleuts.

Other potential sources of admixture in the Bering community came from individuals relocated to the Commander Islands by the Russians, including several Pacific Eskimos from Kodiak Island and "Creoles" who were either Russian-Tlingit or Russian-Aleut (Jochelson 1933; Rychkov and Sheremetyeva 1972). In 1887, when the Russian-American Company gave up its control of the Kuril Islands to Japan, eight Ainus were taken to Bering and six were taken to Medni, and Kamchatkan natives (three men and six women) were also relocated to the Commander Islands. In addition to potentially introducing other non-Aleut Y

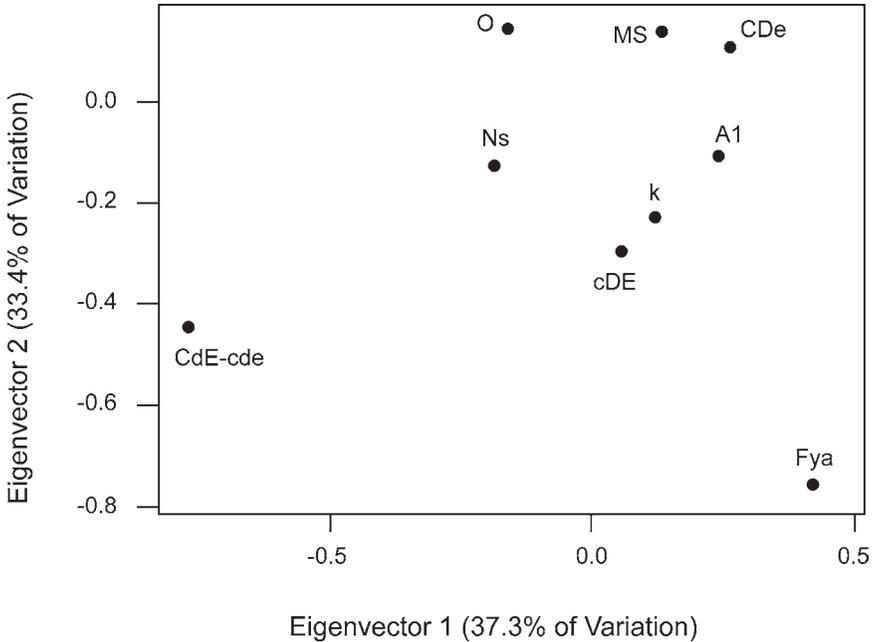


Figure 8. *R* matrix of alleles based on classic genetic markers.

chromosomes to the Bering population, these relocations may also explain the low frequency of non-Aleut mtDNA haplogroups present among the Bering mixed Aleuts, including haplogroup C and G lineages.

Although the classic genetic markers are not highly informative, they do indicate that the Bering Aleuts and the Aleuts of the other historically founded communities of Medni and St. George have genetic similarities. These communities are relatively isolated from surrounding regions because of large interisland distances and frequently adverse weather conditions that make travel difficult. After the initial founding of these communities by Aleuts relocated from the Aleutian archipelago, the genetic contribution from outside sources was somewhat limited and largely male. The other Pribilof Islands community of St. Paul differs genetically, which may be explained by its larger population resulting from an influx of outsiders seeking job opportunities in the Bering Sea crabbing industry.

In summary, it appears that genetic drift has been operating mainly through the maternal side to reduce mtDNA heterozygosity of the Bering Aleut population, whereas the opposing evolutionary effect of male gene flow, predominantly Russian, is responsible for introducing new Y-chromosome and autosomal variants to the Aleut gene pool. This research documents the importance of unique historical events on the distribution of genes in human populations and subsequent

evolutionary changes. Much like Tristan da Cunha, the gene frequencies of the Bering Island Aleut population are unique. The fixation of mtDNA haplogroup D, the result of a founder effect, will have lasting effects on the gene pool of Bering. Likewise, the introduction of non-Aleut Y chromosomes has forever altered the genetic composition of this small community.

Acknowledgments We thank the Aleuts, Russians, Koryaks, and Evens for their participation in this project; we also thank Sergi Makarov and other members of the Russian team for their contribution to the sample collection. This research was supported by the National Science Foundation through grants OPP-9905090 and OPP-0327676.

Received 14 June 2010; accepted for publication 29 June 2010.

Literature Cited

- Achilli, A., U. A. Perego, C. M. Bravi et al. 2008. The phylogeny of the four pan-American mtDNA haplogroups: Implications for evolutionary and disease studies. *PLoS One* 3:e1764.
- Bandelt, H. J., C. Hernstadt, Y. G. Yao et al. 2003. Identification of Native American founder mtDNAs through the analysis of complete mtDNA sequences: Some caveats. *Ann. Hum. Genet.* 67:512–524.
- Bandelt, H. J., P. Forster, and A. Rohl. 1999. Median-joining networks for inferring intraspecific phylogenies. *Mol. Biol. Evol.* 16:37–48.
- Bertorelle, G., and L. Excoffier. 1998. Inferring admixture proportions from molecular data. *Mol. Biol. Evol.* 15(10):1298–1311.
- Brown, M., S. Hosseini, A. Torroni et al. 1998. mtDNA haplogroup X: An ancient link between Europe/West Asia and North America? *Am. J. Hum. Genet.* 63:1852–1861.
- Crawford, M. H., and V. B. Enisco. 1982. Population structure of circumpolar groups of Siberia, Alaska, Canada, and Greenland. In *Current Developments in Anthropological Genetics*, M. H. Crawford and J. H. Mielke, eds. New York: Plenum Press, 51–91.
- Crawford, M. H., J. H. Mielke, E. J. Devor et al. 1981. Population structure of Alaska and Siberian indigenous communities. *Am. J. Phys. Anthropol.* 55:167–185.
- Derbeneva, O. A., R. I. Sukernik, N. V. Volodko et al. 2002. Analysis of mitochondrial DNA diversity in the Aleuts of the Commander Islands and its implications for the genetic history of Beringia. *Am. J. Hum. Genet.* 71:415–421.
- Fagundes, N. J., R. Kanitz, R. Eckert et al. 2008. Mitochondrial population genomics supports a single pre-Clovis origin with a coastal route for the peopling of the Americas. *Am. J. Hum. Genet.* 82:583–592.
- Forster, P., R. Harding, A. Torroni et al. 1996. Origin and evolution of Native American mtDNA variation: A reappraisal. *Am. J. Hum. Genet.* 59:935–945.
- Gilbert, M. T. P., T. Kivisild, B. Grønnow et al. 2008. Paleo-Eskimo mtDNA genome reveals matrilineal discontinuity in Greenland. *Science* 320(27):1787–1789.
- Helgason, A., G. Palsson, H. S. Pedersen et al. 2006. mtDNA variation in Inuit populations of Greenland and Canada: Migration history and population structure. *Am. J. Phys. Anthropol.* 130:123–134.
- Harpending, H., and T. Jenkins. 1973. Genetic distance among southern African populations. In *Methods and Theories of Anthropological Genetics*, M. H. Crawford and P. Workman, eds. Albuquerque: University of New Mexico Press, 177–199.
- Harpending, H., and A. Rogers. 1984. *ANTANA: A Package for Multivariate Data Analysis*. Distributed by the authors.

- Harpending, H., and R. Ward. 1982. Chemical systematics and human populations. In *Biochemical Aspects of Evolutionary Biology*, M. Nitecki, ed. Chicago: University of Chicago Press, 213–256.
- Hayes, M. G., and D. O'Rourke. 2000. Replacement versus continuity in the prehistoric North American Arctic as assessed by ancient mtDNA. *Am. J. Phys. Anthropol.* 30(suppl.):174–175.
- Jeran, N., D. H. Augustin, B. Grahovac et al. 2009. Mitochondrial DNA heritage of Cres Islanders: Example of Croatian genetic outliers. *Colleg. Antropol.* 33(4):1323–1328.
- Jochelson, W. 1933. *History, Ethnology, and Anthropology of the Aleut*. Salt Lake City: University of Utah Press.
- Kivisild, T., H. V. Tolk, J. Parik et al. 2002. The emerging limbs and twigs of the east Asian mtDNA tree. *Mol. Biol. Evol.* 19:1737–1751.
- Lantis, M. 1984. Aleut. In *Handbook of North American Indians*, v. 5, *Arctic*, D. Damas, ed. Washington, DC: Smithsonian Institution, 161–184.
- Laughlin, W. 1980. *Aleuts: Survivors of the Bering Land Bridge*. New York: Holt, Rinehart & Winston.
- Liapunova, R. 1996. *Essays on the Ethnology of the Aleuts*, W. Workman and L. Black, trans. Fairbanks: University of Alaska Press.
- Lorenz, J., and D. Smith. 1996. Distribution of the four founding haplogroups among Native North Americans. *Am. J. Phys. Anthropol.* 101:307–323.
- Majumder, P., W. Laughlin, and R. Ferrell. 1988. Genetic variation in the Aleuts of the Pribilof Islands and the Eskimos of Kodiak Island. *Am. J. Phys. Anthropol.* 76:481–488.
- Merriwether, D., F. Rothhammer, and R. Ferrell. 1995. Distribution of the four founding lineage haplotypes in Native Americans suggests a single wave of migration for the New World. *Am. J. Phys. Anthropol.* 98:411–430.
- Ota, T. 1993. *Manual for DISPAN: Genetic Distance and Phylogenetic Analysis*. University Park: Institute of Molecular Evolutionary Genetics, Pennsylvania State University.
- Rogers, A., and H. Harpending. 1992. Population growth makes waves in the distribution of pairwise genetic differences. *Mol. Biol. Evol.* 9:552–569.
- Rubicz, R., P. E. Melton, V. Spitsyn et al. 2010. Genetic structure of Native circumpolar populations based on autosomal, mitochondrial, and Y-chromosome DNA markers. *Am. J. Phys. Anthropol.* 143:62–74.
- Rubicz, R., T. Schurr, P. Babb et al. 2003. Mitochondrial DNA variation and the origins of the Aleuts. *Hum. Biol.* 75:809–835.
- Rychkov, Y., and V. Sheremetyeva. 1972. Population genetics of the Commander Islands Aleuts related to the problems of history and of adaptation of the ancient Beringian peoples. *Vopr. Antropol.* 40:45–70 (in Russian).
- Saillard, J., P. Forster, N. Lynnerup et al. 2000. mtDNA variation among Greenland Eskimos: The edge of the Beringian expansion. *Am. J. Hum. Genet.* 67:718–726.
- Schneider, S., D. Roessler, and L. Excoffier. 2000. *Arlequin Version 2.00: A Software for Population Genetics Data Analysis*. Geneva: Genetics and Biometry Laboratory, University of Geneva.
- Schurr, T., and D. Wallace. 1999. Mitochondrial DNA variation in Native Americans and Siberians and its implications for the peopling of the New World. In *Who Were the First Americans: Proceedings of the 58th Annual Biological Colloquium*, R. Bonnicksen, ed. Corvallis: Center for the Study of the First Americans, Oregon State University, 41–77.
- Shields, G., A. Schmiechen, B. Frazier et al. 1993. mtDNA sequences suggest a recent evolutionary divergence for Beringian and northern North American populations. *Am. J. Hum. Genet.* 53:549–562.
- Slatkin, M., and R. Hudson. 1991. Pairwise comparisons of mitochondrial DNA sequences in stable and exponentially growing populations. *Genetics* 129:555–562.
- Soodyall, H., T. Jenkins, A. Mukherjee et al. 1997. The founding mitochondrial DNA lineages of Tristan da Cunha islanders. *Am. J. Phys. Anthropol.* 104:157–166.
- Soodyall, H., A. Nebel, B. Morar et al. 2003. Genealogy and genes: Tracing the founding fathers of Tristan da Cunha. *Eur. J. Hum. Genet.* 11(9):705–709.

- Starikovskaya, Y., R. Sukernik, T. Schurr et al. 1998. mtDNA diversity in Chukchi and Siberian Eskimos: Implications for the genetic history of ancient Beringia and the peopling of the New World. *Am. J. Hum. Genet.* 63:1473–1491.
- Tamm, E., T. Kivisild, M. Reidla et al. 2007. Beringian standstill and spread of Native American founders. *PLoS One* 2:829.
- Thangaraj, K., L. Singh, A. Reddy et al. 2003. Genetic affinities of the Andaman Islanders, a vanishing human population. *Curr. Biol.* 13:86–93.
- Torrioni, A., J. V. Neel, R. Barrantes et al. 1994. Mitochondrial DNA “clock” for the Amerinds and its implications for timing their entry into North America. *Proc. Natl. Acad. Sci. USA* 91(3):1158–1162.
- Torrioni, A., T. Schurr, M. Cabell et al. 1993. Asian affinities and continental radiation of the four founding Native American mtDNAs. *Am. J. Hum. Genet.* 53:563–590.
- Vanstone, J. 1984. Exploration and contact history of Western Alaska. In *Handbook of North American Indians*, v. 5, *Arctic*, D. Damas, ed. Washington, DC: Smithsonian Institution, 149–160.
- Zlojutro, M., R. Rubicz, and M. H. Crawford. 2009. Mitochondrial DNA and Y-chromosome variation in five eastern Aleut communities: Evidence for genetic substructure in the Aleut population. *Ann. Hum. Biol.* 36(5):511–526.
- Zlojutro, M., R. Rubicz, E. Devor et al. 2006. Genetic structure of the Aleuts and circumpolar populations based on mitochondrial DNA sequences: A synthesis. *Am. J. Phys. Anthropol.* 129:446–464.