

# Who Were the First Americans?

*Proceedings of the 58th Annual Biology  
Colloquium, Oregon State University  
Robson Bonnichsen, Editor*



*A Peopling of the Americas Publication  
Ruth Gruhn, Series Editor*

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Design and typesetting by C & C Wordsmiths, Blue Hill, Maine

Printed by Downeast Graphics & Printing, Inc, Ellsworth, Maine

ISBN: 0-912933-20-8

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DOI 03939

### **3. mtDNA Variation in Native Americans and Siberians and Its Implications for the Peopling of the New World**

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We have investigated questions about the origins, affinities, and antiquity of the First Americans through the analysis of mitochondrial DNA (mtDNA) variation in contemporary Native American and Siberian populations, using both high-resolution restriction (RFLP) analysis and control region (CR) sequencing to characterize the genetic variation in these groups. Our results indicate that nearly all Native American mtDNAs belong to one of four haplotype groups (haplogroups), designated A–D, which originated in Asia, while recent work has also detected a fifth founding haplogroup which is genetically linked to the rare Eurasian haplogroup X. In addition, we observed the presence of “other” (non-haplogroup A–D) mtDNAs that appear to be autochthonous

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haplotypes which derived from haplogroup C and D mtDNAs after the initial peopling of the Americas, as well as both limited European and African genetic influences on certain Native American populations. By contrast, native Siberian populations had mtDNAs from haplogroups A, C, and D but lacked those from haplogroup B. Furthermore, northeast Siberian populations exhibited a number of other mtDNA lineages, including haplogroups G, Y, and Z, which do not appear in Native American populations, implying their emergence in Asia after the peopling of the New World. Sequence divergence estimates for haplogroups A–D in Asia and the New World suggested that ancestral Native Americans first came to the New World between 30,000 and 40,000 yr B.P., with the Na-Dene Indians and Eskimo-Aleuts originating from genetic stocks occupying Beringia after the last period of glaciation.

## Introduction

For decades, researchers have attempted to provide models which would adequately explain the origins of the considerable cultural diversity, linguistic complexity, and biological variation of Native Americans. At times, the observed diversity has been attributed to multiple migrations of Asiatic peoples to the Americas, a few migrations of ethnically distinct peoples, or in situ differentiation of ancestral Native Americans occurring after the initial colonization of the New World (discussed in Laughlin and Harper 1988; Szathmary 1984). A hypothesis that has guided much of the recent genetic studies of New World populations (Greenberg et al. 1986) proposes that three waves of migration corresponding to the tripartite division of Native American languages [Amerind, Na-Dene, and Eskaleut] (Greenberg 1987) gave rise to all Native American peoples. However, the common origin of the numerous Amerind languages and the time depth required to develop this plurality of tongues have been vigorously disputed (Campbell 1986; Nichols 1990), as have the interpretations of the dental and nuclear genetic data on which the tripartite model was based (Szathmary 1993).

Moreover, estimates of the time of arrival of the first migrants have also varied widely. Archaeological studies of Clovis (Bonnichsen and Turnmire 1991; Haynes 1992) and non-Clovis (Adovasio et al. 1990) sites in North America have dated these materials at 11,000–14,000 yr B.P., while South American sites with different lithic traditions (Bryan et al. 1978; Dillehay 1997; Roosevelt et al. 1996) appear to be at least contemporaneous with, if not older than, the Clovis sites. In addition, linguistic data have been used to approximate the entry time of humans into the Americas, with these dates ranging between c. 12,000 yr B.P. (Greenberg et al. 1986) and 35,000 yr B.P. (Nichols 1990, 1994). Thus, many of the fundamental questions concerning the origins of the First Americans and the timing of their entry into the New World remain incompletely resolved.

To clarify these questions further, a number of investigators began analyzing the

mtDNA variation of Native American and Siberian populations. Because of its haploid, maternal transmission (Giles et al. 1980), high mutation rate (Brown et al. 1979; Stoneking et al. 1986; Wallace et al. 1987), and lack of recombination (Merriwether et al. 1991), the patterns of accumulated sequence changes in mtDNAs along branching female lineages can be reconstructed with minimum ambiguity. These mutational differences also permit the discrimination between closely related or geographically proximate populations on the basis of genotypic variation. Furthermore, based on the distribution of specific mutations in different populations, it is possible to reconstruct ancient human migration patterns, as has been demonstrated by many studies (Ballinger et al. 1992; Cann et al. 1987; Lum et al. 1994; Stoneking et al. 1990). In the following pages, we describe analogous molecular anthropological studies of Native American and Siberian mtDNA variation, and the insights into the peopling of the New World that they have provided.

## mtDNA Variation in the New World

In our studies of mtDNA variation in Native American populations (Figure 1), we have shown that Native American mtDNAs belonged to primarily four haplogroups, or mtDNA lineages, of related genotypes (Schurr et al. 1990; Torroni et al. 1992; Torroni, Schurr, et al. 1993). These haplogroups were designated A, B, C and D, with haplogroup A mtDNAs being defined by a HaeIII site gain at np 663, haplogroup B mtDNAs by the COII/tRNA<sup>Lys</sup> intergenic (Region V) 9-bp deletion, haplogroup C mtDNAs by the linked HincII site loss at np 13259 and AluI site gain at np 13262 (HincII/AluI polymorphism), and haplogroup D mtDNAs by the AluI site loss at np 5176, with both haplogroup C and D mtDNAs also having the DdeI site gain at np 10394, and an AluI site gain at np 10397 (DdeI/AluI sites) (Schurr et al. 1990; Torroni et al. 1992). The mtDNAs which did not fall into these four haplogroups were designated "other" haplotypes (Torroni et al. 1992; Torroni, Schurr, et al. 1993), and attributed to non-native admixture due to their apparent affinities with European haplogroups (Brown et al. 1992; Torroni, Lott, et al. 1994). The exception was Makiritare haplotype AM83, which possessed the DdeI/AluI sites that appear in all haplogroup C and D mtDNAs, but lacked the other RFLP markers of these two mtDNA lineages (Torroni, Schurr, et al. 1994).

We interpreted these data as showing several major trends in the mtDNA variation of Native American groups. First, ancestral Amerindian populations brought at least haplogroup A, C, and D mtDNAs during the initial colonization(s) of the New World. Since each haplogroup appeared to have derived from a single haplotype, we proposed that ancestral Native Americans originated from a limited number of founders, perhaps through a bottleneck effect. We further suggested that the ancestors of the Na-Dene Indians migrated from Asia independently and considerably more recently than the progenitors of the Paleoindians, since the extent of diversity of haplogroup A in the Na-Dene Indians was half of that in Amerindians. Similarly, because of its low diversity relative to the other mtDNA lineages, as well as its



**Figure 1.** A map showing the three major linguistic divisions of Native American languages according to Greenberg (1987), along with the geographic locations of tribal populations analyzed in our mtDNA studies (Schurr et al. 1990; Torroni et al. 1992; Torroni, Schurr, et al. 1993; Torroni, Sukernik, et al. 1993; Torroni, Chen, et al. 1994; Torroni, Neel, et al. 1994; Huoponen et al. 1997).

nonuniform distribution in Asia and the Americas, we suggested that haplogroup B might represent a second migration independent of that which contributed haplogroup A, C, and D mtDNAs to the genetic stock of Paleoindians. We concluded that the extent of Paleoindian mtDNA diversity was compatible with a pre-Clovis colonization of the New World.

However, other studies of mtDNA variation in Asian and Native American populations disputed some of these interpretations. Ward et al. (1991) argued against extensive bottlenecks causing limited mtDNA variation among Native American groups, as they observed CR sequence diversity within a single Native American tribe which was similar to that found in Asian populations, and also apparently detected more than four haplogroups (CR lineage clusters) in Native American groups. Horai et al. (1993) found similar levels of sequence diversity in their Amerindian sample, although claiming that each haplogroup represented a separate migration to the New World. In addition, Shields et al. (1993) also proposed a "late" entry time (12,000–14,000 yr B.P.) of ancestral Amerindians to the New World rather than an "early" entry time, as suggested by Torroni et al. (1992), Torroni, Schurr, et al. (1993), and Torroni, Sukernik, et al. (1993), along with the late expansion of northern populations in the circumarctic region (5,000–7,000 yr B.P.). Thus, there were considerable differences in the interpretation of mtDNA data from Native American populations, indicating that further work was needed to clarify these contrasting perspectives of north Asian prehistory and the colonization of the New World.

Stimulated by these initial findings, a number of researchers began analyzing the nature and distribution of mtDNA variants in New World groups (Bailliet et al. 1994; Batista et al. 1995; Easton et al. 1996; Fox 1996; Ginther et al. 1993; Kolman et al. 1995; Lorenz and Smith 1994, 1996; Merriwether et al. 1994, 1995; Monsalve et al. 1996; Santos et al. 1994; Stone and Stoneking 1994; Torroni, Chen, et al. 1994; Torroni, Neel, et al. 1994; Ribeiro-Dos-Santos et al. 1996; Ward et al. 1996). Without exception, these studies of both ancient and modern population samples have shown that haplogroups A–D constitute the vast majority of all mtDNAs present in Native American tribes. Thus, they can unequivocally be considered founding mtDNA lineages in New World populations.

In addition, some of these studies detected the presence of "other" haplotypes in different Native American groups (Bailliet et al. 1994; Ginther et al. 1993; Lorenz and Smith 1996; Merriwether et al. 1994, 1995). The reports of "other" haplotypes in Native American populations were significant for two primary reasons. If confirmed, their presence would indicate that previously unidentified founding mtDNA lineages from Asia were brought to the New World during its initial phase of colonization, and that the diversity of founding populations was greater than previously hypothesized. Alternatively, their presence could reveal non-native admixture in Native American populations which had occurred in a post-Columbian context. Thus, the molecular characterization of "other" haplotypes was crucial for distinguishing between these possibilities and interpreting the diversity of ancestral Native Americans.

Several different types of putative "other" haplotypes have been identified in New World groups. Those which we first detected in the Ojibwa and Navajo (AM29, AM74-AM76; Torroni et al. 1992; Torroni, Schurr, et al. 1993) appeared to be very similar to haplogroup X mtDNAs seen in French Canadians and other European groups (Brown et al. 1992; Torroni, Lott, et al. 1994; Torroni et al. 1996). Haplogroup X mtDNAs are characterized by the DdeI np 1715 site loss and the np 16223 C->T (16223T) and np 16278 C->T (16278T) transitions in the CR (Forster et al. 1996; Torroni et al. 1996), but would exhibit only the HaeIII np 16517 site when screened for the defining RFLPs of haplogroups A-D (Table 1). Since the Ojibwa had experienced considerable contact with Europeans (Szathmary and Auger 1983), these "other" mtDNAs were thought to be of non-native origin. The presence of 6.6 percent "other" mtDNAs in 25-30 North American tribes (Lorenz and Smith 1996) were also initially attributed to non-native admixture, due to most of these haplotypes occurring in Eastern and Midwestern tribes which had been genetically influenced by European populations (Schell et al. 1978).

However, recent studies have shown the presence of "other" mtDNAs in both contemporary and ancient populations of North America (Hauswirth et al. 1994; Stone and Stoneking 1994, 1998; Torroni et al. 1992; Torroni, Schurr et al. 1993; Ward et al. 1991, 1993) and South (Bailliet et al. 1994; Ribeiro-Dos-Santos et al. 1996; Ward et al. 1996), a number of which had the 16223T and 16278T transitions. These findings suggested that haplogroup X mtDNAs could be more widespread in the New World than previously believed, and were not the consequence of non-native gene flow. In addition, recent phylogenetic analyses have shown that

Table 1. Proposed founding Native American mtDNA haplotypes.

haplogroup	haplotype	polymorphic restriction sites
A	A1	+663e
	A2	+663e, +16517e
B	B1	COII/tRNA <sup>Pro</sup> 9-bp Deletion, +16517e
	C1	+10394c, +10397a, -13259o/ +13262a,
C	C2	+10394c, +10397a, -13259o/ +13262a, +16517e
	D1	-5176a, +10394c, +10397a
D	D2	-5176a, +10394c, +10397a, +16517e
	E	-1715c, +16517e (np 16223T, np 16278T)
X	X6	+10394c, +10397a
	X7	+10394c, +10397a, +16517e

Bailliet et al. (1994) and Merriwether et al. (1995) used a partial haplotype method similar to that of Torroni et al. (1993a) to analyze their South American Indian samples. In this analysis, the samples were screened for only the RFLP markers for haplogroups (A-D) [see above], and then assigned a haplogroup designation based on the presence or absence of these polymorphisms. Haplogroup E was identified by both partial haplotype analysis and CR sequencing. The equivalent nomenclature for RFLP haplotypes proposed to be possible founding mtDNAs in Torroni et al. (1992, 1993a) are as follows: A1 = AM01; A2 = AM09; B1 = AM13; C1 = AM32; C2 = AM43; D1 = AM88; and D2 = AM44. There were no comparable haplotypes for E, which might be considered synonymous with haplogroup X (-DdeI 1715, +16517e). In addition, while AM83 had the DdeI np 10394 and AluI np 10397 sites, making it similar to X6 and X7 mtDNAs, it was never proposed to be an additional founding haplotype (see text).

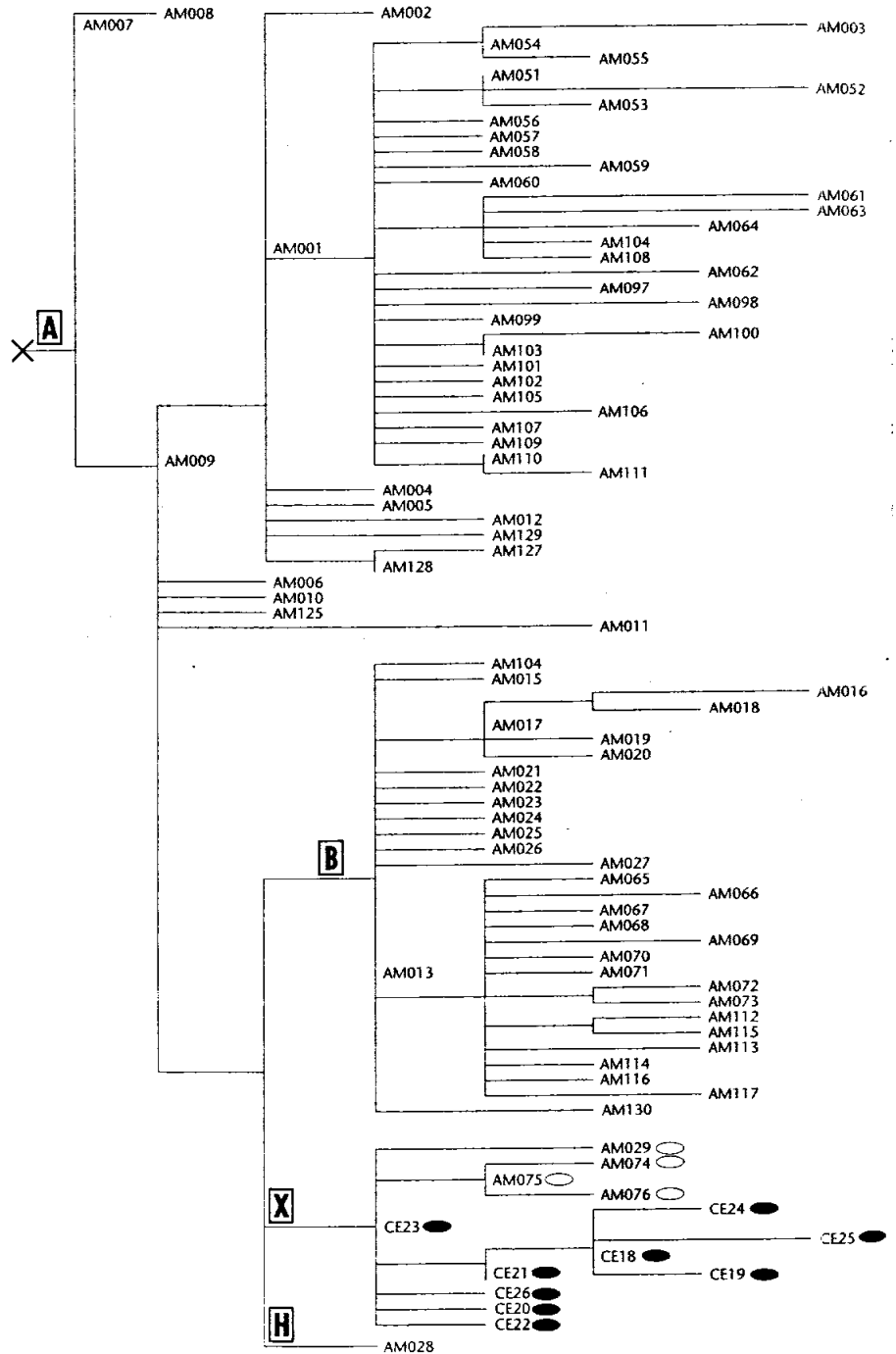


haplogroup X mtDNAs from Native Americans are distinctive from comparable European mtDNAs (Figure 2), that haplogroup X mtDNAs appear at much higher frequencies in North American Indian populations than in those in South America, and that mtDNAs bearing the 16278T polymorphism in North American Indians may differ from similar types in South American tribes (Brown et al. 1998; Schurr, Brown, et al. in prep). Collectively, these data supported the interpretation that haplogroup X was a fifth founding haplogroup in Native Americans (Brown et al. 1998; Forster et al. 1996; Scozzari et al. 1997), and that it could possibly have undergone considerable diversification since arriving in the New World (Brown et al. 1998).

On the other hand, a minority of haplotypes in North American Indians appeared to belong to haplogroup H, the most common mtDNA lineage in modern European populations (Torroni, Lott, et al. 1994; Torroni et al. 1996) (Figure 2). These haplotypes included AM28 in the Maya (Torroni et al. 1992) and a small number of mtDNAs in the Ojibwa (Scozzari et al. 1997). The rarity of haplogroup H mtDNAs in Native Americans and their predominance in Europeans (over 40 percent in most populations) suggested that, in contrast to haplogroup X mtDNAs, these haplotypes were acquired through female European gene flow in the last several hundred years, rather than representing an additional founding mtDNA lineage.

Furthermore, another possible founding mtDNA lineage was observed among South American Indians. In contrast to European haplogroups, these X6/X7 haplotypes in the Yanomami had the DdeI/AluI sites while differing by the presence or absence of the HaeIII np 16517 site (Easton et al. 1996). The DdeI/AluI sites define macrohaplogroup M, which encompasses 55–70 percent of all Asian mtDNAs, and includes numerous well-defined haplogroups (Ballinger et al. 1992; Schurr et al. 1999; Torroni, Miller, et al. 1994). For this reason, and because an apparently similar haplotype, AM83, had also been detected in the Makiritare (Torroni, Schurr, et al. 1993), Easton et al. (1996) proposed that X6/X7 mtDNAs belonged to another founding Asian mtDNA lineage with these polymorphisms. However, when the CR sequences of Yanomami X6/X7 mtDNAs and Makiritare AM83 were analyzed with those from other Native American populations, they were shown to cluster among CR sequences from haplogroups C and D (Schurr, Brown, et al. in prep). These results suggested that X6 and X7 mtDNAs, as well as AM83, were autochthonous haplotypes that derived from haplogroup C and D mtDNAs after the peopling of the Americas, rather than representing an additional founding mtDNA lineage in Native American populations.

A further potential complication for determining the origins of "other" haplotypes was the occurrence of recent African gene flow into Amerindian populations through the incorporation of African Americans into native groups over the past several hundred years. Evidence of this sort of African–Native American admixture was shown by the presence of mtDNAs belonging to African haplogroup L (Chen et al. 1995) in the Seminoles of Florida (AM132; Huoponen et al. 1997) (Figure 2), as well as both African mtDNA and Y-chromosome haplotypes in the Mixtec and



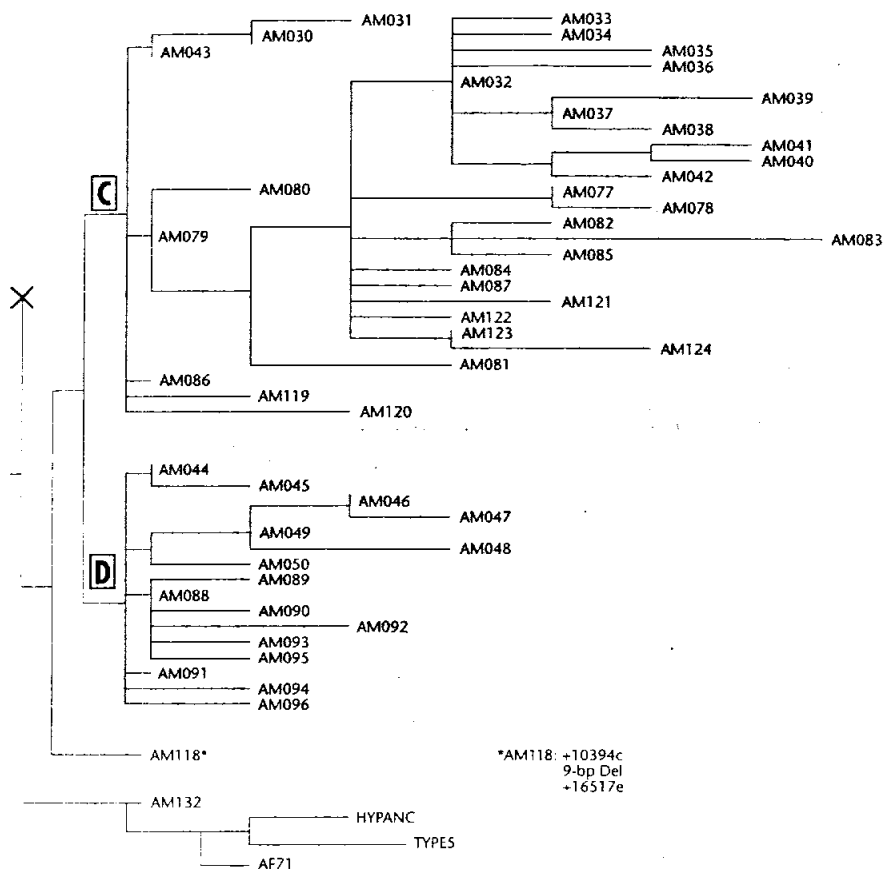


Figure 2. (this page and opposite) A maximum parsimony (MP) tree of Native American P haplotypes generated with PAUP 3.1.1 (Swofford 1994). The haplogroups to which the haplotypes belong are indicated by the boxed capital letters, with the corresponding haplotypes being located in the adjacent branches. All haplotypes are positioned at the terminal ends of the branches, and are numbered according to the publications in which they appear (Torroni et al. 1992; Torroni, Schurr, et al. 1993; Torroni, Sukernik, et al. 1993; Torroni, Chen, et al. 1994; Torroni, Neel, et al. 1994; Huoponen et al. 1997). The white and black oblongs in haplogroup X denote the Native American and European haplotypes, respectively, which belong to this mtDNA lineage. Note that haplogroup L encompasses both AM132 in the Seminoles (Huoponen et al. 1997) and African "Outgroup" haplotypes AF71 (Chen et al. 1995) and HYPANC [Hypothetical Ancestor] and TYPE5 (Torroni et al. 1987).

of the P haplogroup of the Tlaxtec Indians of southern Mexico (Lell et al. 1997; Torroni, Chen, et al. 1994). The P haplogroup L lacks all RFLP markers from haplogroups A–D except the Ddel 10394 site (Chen et al. 1995), and since many African CR sequences also have 16223T and 16278T mutations (Chen et al. Submitted; Vigilant et al. 1989,

1991), the haplotypes from haplogroup L could potentially be misidentified "other" mtDNAs, and inadvertently assigned to Native American haplogroup X.

Finally, recent studies have proposed that two haplotypes from haplogroups C, and D, which differ by the presence or absence of the HaeIII np 16517 site, were both among the original founding Native American mtDNAs (Bailliet et al. 1994; Easton et al. 1996; Merriwether et al. 1994, 1995) (Table 1). Because mtDNAs with and without the HaeIII np 16517 site are found in many Native American groups, it was argued that they were brought together to the Americas in a single migratory wave (Bailliet et al. 1994; Cann 1994; Merriwether et al. 1994, 1995; Merriwether and Ferrell 1996). However, it has been shown that many Native American haplotypes within one of the primary haplogroups (A–D) are identical at the RFLP level with the exception of this polymorphism (Torrioni et al. 1992; Torrioni, Chen, et al. 1994; Torrioni, Neel, et al. 1994; Torrioni, Schurr, et al. 1993; Torrioni, Sukernik, et al. 1993), and the same trend was observed among haplogroups present in Asia (Ballinger et al. 1992; Torrioni, Miller, et al. 1994), Europeans (Torrioni et al. 1994) and Africans (Chen et al. 1995). These data demonstrated that the HaeIII np 16517 site is the most variable site detected by RFLP analysis in world populations, and that the presence or absence of the HaeIII np 16517 site in different haplotypes of different geographic backgrounds is due to new mutations rather than the presence of numerous ancient founding mtDNA lineages.

Therefore, the current mtDNA data indicate that the total number of founding RFLP haplotypes for haplogroups A–D and X may only be 5–6 rather than the 8–10 recently claimed by other groups (Bailliet et al. 1994; Bianchi and Rothhammer 1995; Cann 1994; Easton et al. 1996; Merriwether et al. 1994, 1995). However, each founding RFLP haplotype may have more than one founding CR sequence associated with it because of the higher mutation rate of the non-coding CR (Horai and Hayasaka 1990; Vigilant et al. 1989, 1991; Ward et al. 1991). In any case, the presence of a small number of founding mtDNAs in Native American populations which inhabit a broad geographic expanse continues to suggest that the Americas were populated by a relatively limited number of founders.

## Native Siberian mtDNA Variation

Studies of mtDNA variation in native Siberian populations (Figure 3) have shown them to have only three of the four Asian mtDNA haplogroups present in Native Americans (A, C, and D) (Schurr et al. 1999; Starikovskaya et al. 1998; Sukernik et al. 1996; Torrioni, Schurr, et al. 1993) (Table 2). With very few exceptions (Table 2), all Siberian groups lacked haplogroup B mtDNAs, and those which did have these haplotypes inhabited the southern margin of Siberia adjacent to Mongolia and northern China (Petrishchev et al. 1993; Shields et al. 1993; Sukernik et al. 1996). Haplogroup C mtDNAs were very widely distributed among eastern Siberian populations, but relatively rare (3–10 percent) in East Asia populations (Ballinger et al. 1992; Horai et al. 1984; Kolman et al. 1996; Merriwether et al. 1996), and alm-

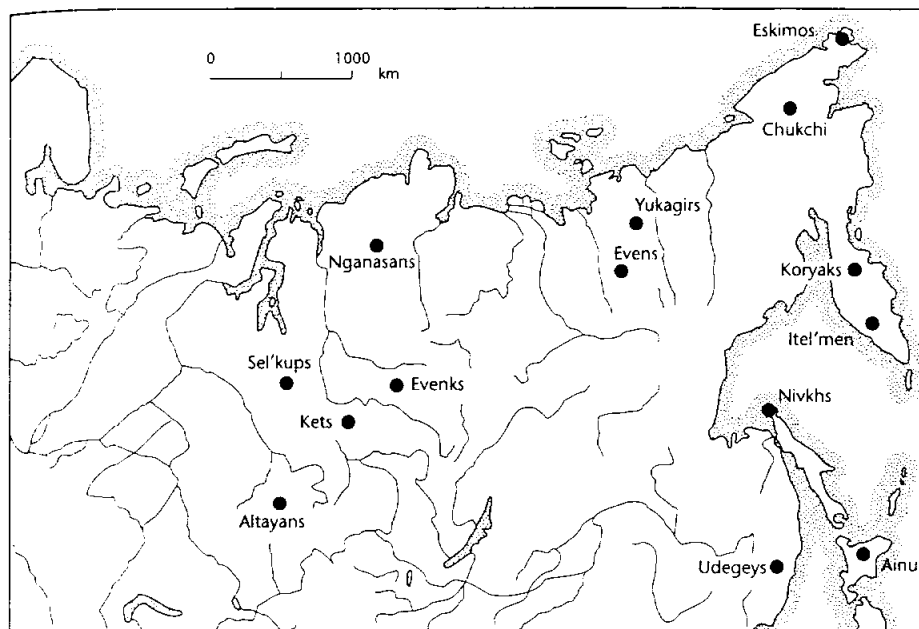


Figure 3. A map of Siberian populations analyzed for mtDNA variation and described in this paper.

completely absent in Southeast Asia (Ballinger et al. 1992). Haplogroup D was also commonly observed in Asia, although its distribution in Siberian groups was much more geographically irregular than that of haplogroup C, and appeared at only moderate frequencies among Tibetans (Torroni, Miller, et al. 1994), Koreans and Han Chinese (Ballinger et al. 1992) and Mongolians (Kolman et al. 1996; Merriwether et al. 1996). Haplogroup A had a marked asymmetric distribution in Siberia, appearing at high frequencies in the Chukchi and Siberian Eskimos of Chukotka, but otherwise being absent or only present at low frequencies in other eastern Siberian populations (Schurr et al. 1999; Starikovskaya et al. 1998; Sukernik et al. 1996; Torroni, Schurr, et al. 1993).

Interestingly, the haplogroup A frequencies in Chukotkan populations were comparable to those in other circumpolar populations in the New World (Table 2), with all circumpolar populations in the New World having predominantly haplogroup A and D mtDNAs (Shields et al. 1993; Torroni, Schurr, et al. 1993; Ward et al. 1991, 1993) (Table 2). Amongst these populations, the Siberian and Alaskan Eskimos appeared genetically very similar to one another by having higher frequencies of haplogroup A, while the Aleuts differed from Eskimo groups by having mostly haplogroup D mtDNAs. In addition, both sets of Eskimo groups had a very low frequency of haplogroup C mtDNAs, suggesting this haplogroup could represent a founding mtDNA lineage in Eskimo-Aleut populations, although those

Table 2. mtDNA haplogroup distribution in Siberian and Native American populations.

population	haplogroup frequencies (%)									other	reference	
	N	A	B	C	D	F	G	Y	Z			
<b>Amerindians</b>												
Bella Coola	32	78.1	6.3	9.4	6.3	—	—	—	—	—	—	1
Bella Coola	25	60.0	8.0	8.0	20.0	—	—	—	—	—	4.0	2
Nuu-Chah-Nulth	63	44.4	3.2	19.0	22.2	—	—	—	—	—	11.1	3
Nuu-Chah-Nulth	15	40.0	6.7	13.3	26.7	—	—	—	—	—	13.3	2
<b>Na-Dene</b>												
Haida	38	92.1	—	7.9	—	—	—	—	—	—	—	1
Haida	25	96.0	—	—	4.0	—	—	—	—	—	—	2
Dogrib	154	90.9	—	2.0	—	—	—	—	—	—	7.1	4
Dogrib	30	100.0	—	—	—	—	—	—	—	—	—	5
<b>Eskimo-Aleut</b>												
St. Paul Aleut	72	25.0	—	1.4	66.7	—	—	—	—	—	6.9	4
Old Harbor Eskimos	115	61.7	3.5	—	34.8	—	—	—	—	—	—	4
Ouzinkie Eskimos	41	73.2	—	4.9	14.6	—	—	—	—	—	7.3	4
St. Lawrence Eskimos	99	76.0	—	7.0	14.0	—	—	—	—	—	3.3	4
Siberian Eskimos	79	77.2	—	2.5	20.3	—	—	—	—	—	—	6
<b>Paleoasiatic</b>												
Koryaks	155	3.9	—	37.4	1.3	—	42.6	9.7	3.9	—	—	7
Itel'men	47	6.4	—	14.9	—	—	68.1	4.3	6.4	—	—	7
Chukchi	66	68.2	—	10.6	12.1	—	9.1	—	—	—	—	6
<b>Isolated Language</b>												
Nivkhs	57	—	—	—	28.1	—	5.3	64.9	—	—	1.8	8
<b>Tungusic</b>												
Udegeys	45	—	—	17.8	—	—	—	8.9	—	—	73.3	8
Evenks	51	3.9	—	84.3	9.8	2.0	—	—	—	—	—	8
Evens	43	—	—	58.1	7.0	—	2.3	—	—	—	32.6	8,9
<b>Uralic</b>												
Yukaghirs	27	—	—	59.3	33.3	—	3.7	—	—	—	3.7	8,9
Nganasans	49	2.0	—	38.8	36.7	—	2.0	—	—	—	20.4	8,9
Sel'kups	20	—	—	35.0	—	—	—	—	—	—	65.0	8
<b>Altaic</b>												
Northern Altayans	28	3.6	3.6	35.7	14.3	—	—	—	—	—	42.8	10
<b>Ketic</b>												
Kets	23	4.3	—	17.4	—	—	—	—	—	—	78.2	10

Distribution of mtDNA haplogroups in eastern Siberian and East Asian populations. The "Other" category specifies haplotypes which do not belong to the well-defined haplogroups identified in the table, but which may have different haplogroup affiliations. The mutational composition of "Other" haplotypes was either (1)-Ddel np 10394, -Alul np 10397, ±HaeIII np 16517; or (2) +Ddel np 10394, +Alul np 10397, ±HaeIII np 16517. For the references, 1 = Ward et al. (1993); 2 = Torroni et al. (1993a); 3 = Ward et al. (1991); 4 = Merriwether et al. (1995); 6 = Starikovskaya et al. (1998); 7 = Schurr et al. (1999); and 8 = Torroni et al. (1993b); 9 = Schurr et al. unpublished; and 10 = Sukernik et al. (1996).

in the Siberian Eskimos may have been obtained through gene flow with the Chukchi (Schurr et al. 1999; Starikovskaya et al. 1998). On the other hand, known gene flow from Europeans (Russians) into the Alaskan Eskimos and Aleuts, and from the Chukchi into the St. Lawrence Eskimos (Crawford et al. 1981; Ferrell et al. 1981; Menovshchikov 1964), as well as tribal interactions with Northwest Coast Amerindian populations (Fitzhugh 1988; Szathmary 1993), make it likely that the very low frequencies of haplogroup B and "other" mtDNAs in Alaskan Eskimos and Aleuts (Merriwether et al. 1994; Merriwether and Ferrell 1996) were acquired through gene flow with non-Eskimo-Aleut populations.

In addition to finding haplogroup A, C, and D mtDNAs in Siberian populations, we also characterized a number of haplogroups which were formerly considered to be "other" mtDNAs in these groups. The primary mtDNA lineages defined by these analyses were haplogroups G, Y, and Z, each of which has a specific set of RFLP and CR sequence polymorphisms that distinguish them from other Asian haplogroups (Schurr et al. 1999; Starikovskaya et al. 1998; Torroni et al. 1993b) (Table 3). mtDNAs

Table 3. mtDNA haplogroups in Siberian populations.

haplogroup	polymorphic restriction sites + CR sequence polymorphisms
A	+663e I: 16111T, 16192T, 16223T, 16290T, 16319A, 16362C II: 16111T, 16223T, 16265G, 16290T, 16319A, 16362C III: 16223T, 16290T, 16319A, 16362C
C	+10394c, +10397a, -13259o/+13262a, +16517e I: 16223T, 16298C, 16327T, 16519C II: 16124C, 16223T, 16298C, 16327T, 16519C III: 16093C, 16189C, 16223C, 16261T, 16288C, 16298C, 16519C
D	-5176a, +10394c, +10397a I: 16223T, 16362C II: 16093C, 16173T, 16223T, 16319A, 16362C III: 16129A, 16223T, 16271T, 16362C
G	+4830n/+4831f, +10394c, +10397a, +16517e I: 16017C, 16093C, 16129A, 16223T, 16519C
Y	+7933j, -8391e, +10394c, +16517e I: 16126C, 16189C, 16231C, 16266T, 16519C
Z	+10394c, +10397a, +11074c, +16517e I: 16129A, 16185T, 16223T, 16224C, 16260T, 16298C, 16519C

Haplogroups defined in eastern Siberians. Polymorphic restriction sites are numbered from the first nucleotide of the recognition sequence according to the published sequence (Anderson et al. 1981). The restriction enzymes used in the analysis are designated by the following singler-letter code: a, *AclI*; b, *Avall*; c, *DdeI*; e, *HaeIII*; f, *HhaI*; g, *HinfI*; h, *HpaI*; i, *HpaII*; j, *MboI*; k, *RsaI*; l, *TaqI*; M, *BamHI*; n, *HaeII*; o, *HincII*. Sites separated by a diagonal line indicate either simultaneous site gains or site losses for two different enzymes, or a site gain for one enzyme and a site loss for another because of a single common nucleotide substitution. These sites are considered to be only one restriction site polymorphism in the statistical analysis. All samples differ from the published sequence (Anderson et al. 1981) by the presence or absence of the following sites: -4769a, +7025a, +8858f, -13702e, -14199o, +14268g, and -14368g.

belonging to haplogroups G and Y occurred solely in northeastern Siberian populations, but were also detected in the Ainu (Horai et al. 1996), Japanese (Horai et al. 1984; Horai et al. 1996), and Koreans (Ballinger et al. 1992; Horai et al. 1996). This distribution suggested that haplogroups G and Y were a major part of the genetic makeup of ancestral Paleoasiatic groups, and that they originated in the area encompassing the Okhotsk Sea and northern Sea of Japan region (Schurr et al. 1999). In addition, the Evens, Yukagirs, and Nganasans were found to have detectable frequencies of haplogroup G mtDNAs (Schurr et al. 1999), implying a considerable degree of genetic contact between eastern Siberian and Paleoasiatic-speaking groups from Kamchatka and Chukotka, an interpretation supported by ethnographic data (Antropova 1964; Bogoras 1910; Jochelson 1908).

Most of the remaining "other" haplotypes belonged to haplogroup Z, and were seen in the Koryaks and Itel'men. All of these mtDNAs had the DdeI/AluI sites and the HaeIII np 16517 site gain, which define Asian macrohaplogroup M (Ballinger et al. 1992; Torroni, Miller, et al. 1994), as well as the novel DdeI np 11074 site gain. Potentially similar partial haplotypes were present in the Evens, Reindeer Chukchi, Yukagirs, and Nganasans (Torroni, Sukernik, et al. 1993), suggesting that haplogroup Z mtDNAs may have originated in Tungusic-speaking populations and spread to other ethnic groups through population contact. However, CR sequences similar to those for haplogroup Z in Kamchatkan populations have also been identified in Mongolians (Kolman et al. 1996) and Koreans (Horai et al. 1996), implying that this mtDNA lineage could have originated in regions farther west or south of Kamchatka.

By contrast, the "other" haplotypes seen in the Udegeys and Nivkhs were distinct from haplogroup Z mtDNAs, as well as all other Siberian mtDNAs, even those bearing the DdeI/AluI sites. These results suggested that the "other" haplotypes in Amur River groups were more closely related to those present in Chinese, Manchurian, or East Asian populations with whom they have had cultural and genetic contact (Ivanov, Levin, et al. 1964; Ivanov, Smolyak, et al. 1964a, b) than to eastern Siberian groups per se. In addition, eastern Siberian populations have been influenced by populations from more southerly regions, as evidenced by the presence in the Evenks of SIB20 (Table 2), a mtDNA haplotype belonging to a haplogroup F which commonly occurs in Tibetan and Southeast Asian populations (Ballinger et al. 1992; Torroni, Miller, et al. 1994; Torroni, Sukernik, et al. 1993).

Furthermore, although "other" haplotypes have been also detected in Native American populations, the limited RFLP and CR sequence data for these samples indicate that they do not belong to haplogroup G, Y and Z mtDNAs (Bailliet et al. 1994; Ginther et al. 1993; Hauswirth et al. 1994; Lorenz and Smith 1996; Stone and Stoneking 1994, 1998). This finding indicates that haplogroup G, Y and Z mtDNAs were not part of the original genetic composition of ancestral Native American populations. Moreover, haplotypes resembling haplogroup X mtDNAs in Amerindians or Europeans have not yet been detected in Asian or Siberian populations (Ballinger et al. 1992; Brown et al. 1998; Schurr et al. unpublished data;

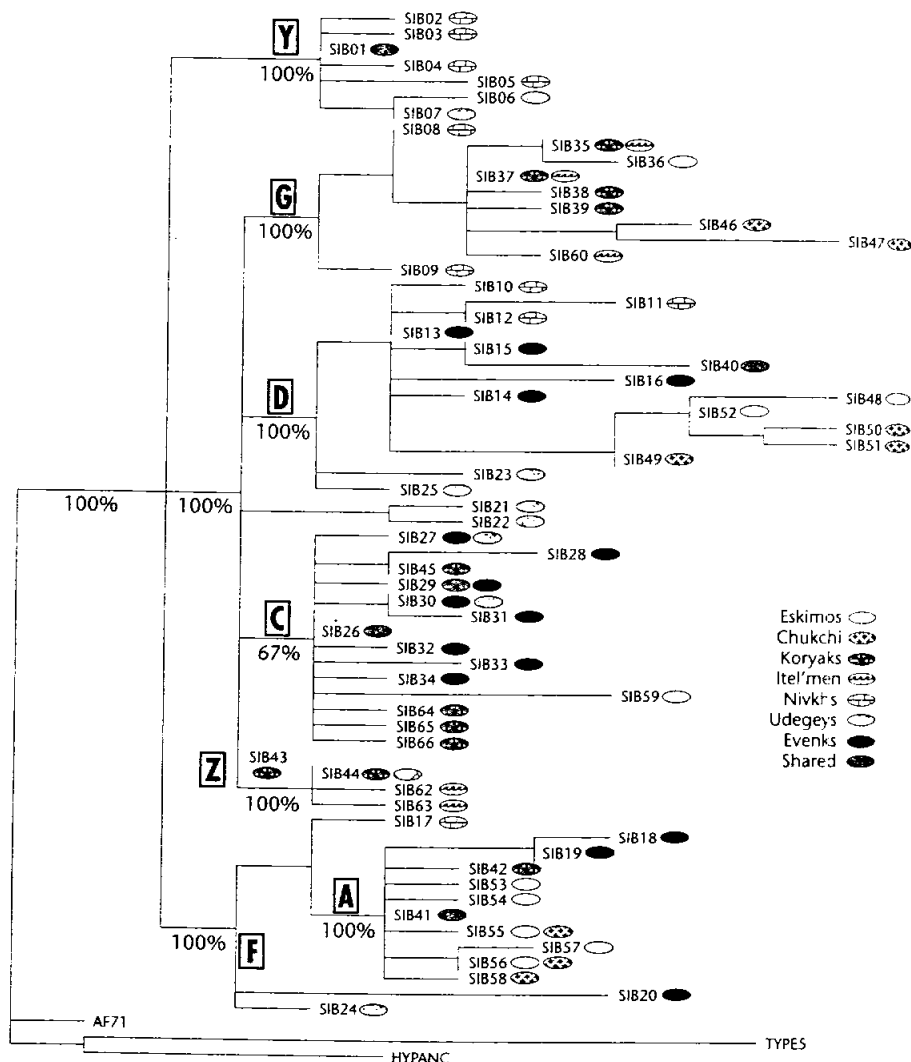


Torrioni, Miller, et al. 1994). Thus, if these mtDNAs evolved in Asia, they must have originated in a region outside of Siberia proper, and then had a transient existence in Siberia itself.

Phylogenetic studies of mtDNA variation in eastern Siberian groups have suggested that most of them derived from a common northern Asian gene pool containing at least the founding haplotypes for haplogroups A, C, and D. Since their common origin, subsequent population dispersal and isolation allowed them to become genetically divergent from one another. The maximum parsimony (MP) analysis (Swofford 1994) of Siberian haplotypes (Figure 4) shows the effects of these factors quite clearly, as the great majority of the haplotypes shared among these populations were positioned at nodal or interior locations in the dendrogram (founding haplotypes). The more recently derived haplotypes from each of these haplogroups were largely population-specific and located at the terminal positions of the branches, while population- or region-specific clusters also occurred within some of these haplogroups (e.g., AM48-53 from Chukchi and Eskimos in haplogroup A). The same trend was observed when Siberian and Native American RFLP haplotypes were analyzed together with the MP method (Starikovskaya et al. 1998).

The phylogenetic analysis of CR sequence variation in Siberian populations provided additional insights into the genealogical relationships between Siberian and Native American populations (Figure 5). All haplogroup A mtDNAs were defined by the np 16223 C->T (16223T), np 16290 C->T (16290T), np 16319 G->A (16319A) and np 16362 T->C (16362C) transitions. In addition, two main lineages were detected within this haplogroup for these populations. The first had the np 16111 C->T (16111T) transition which characterizes nearly all Chukotkan and Native American haplotypes from this mtDNA lineage. Because the 16111T mutation was absent from all Asian and Siberian mtDNAs (Kolman et al. 1996; Schurr et al. 1999; Starikovskaya et al. 1998; Torrioni, Schurr, et al. 1993; Torrioni, Sukernik, et al. 1993), it marks the emergence of ancestral Native American populations in the New World. Within this lineage were two major sublineages, the first (I) having the np 16192 C->T (16192T) mutation which distinguished haplotypes recently evolved in Beringian populations, and the second (II) having the np 16265 A->G (16265G) mutation, which occurs almost exclusively in Eskimoan populations (Shields et al. 1993; Starikovskaya et al. 1998; Torrioni, Schurr, et al. 1993; Ward et al. 1991, 1993). In contrast, the second major lineage (also representing sublineage III) lacked the 16111T mutation. Since these kinds of haplotypes comprise the vast majority of Asian and Siberian haplogroup A mtDNAs (Horai et al. 1996; Kolman et al. 1996; Schurr et al. 1999; Torrioni, Sukernik, et al. 1993), they likely represent the more ancient branch of this mtDNA lineage in Asia, which gave rise to the sublineage which was brought to the New World.

Several sublineages were also present in haplogroup C. The first (I) exhibited all of the CR polymorphisms which define haplogroup C mtDNAs in both Siberian and Native American populations, including the 16223T, np 16298 T->C (16298C), and np 16327 C->T (16327T) transitions. The fact that the nodal haplotype for this haplogroup, SIB26/AM43, had this sequence motif supported its role as a found-



**Figure 4.** MP tree of eastern Siberian RFLP haplotypes. The mtDNA haplogroups observed in native Siberian populations are indicated by the black capital letters in shaded boxes, while haplotypes appearing in each population are identified by filled circles specified in the Symbol Key. "Shared" haplotypes are those observed in more than one population. The African haplotypes used as outgroups to root this tree include AF71 (Chen et al. 1995) and HYPANC and TYPE5 (Cann et al. 1987). The numbers located under the major branches of the MP tree represent the percent support for each branch observed in the 50% Majority Rule consensus tree.

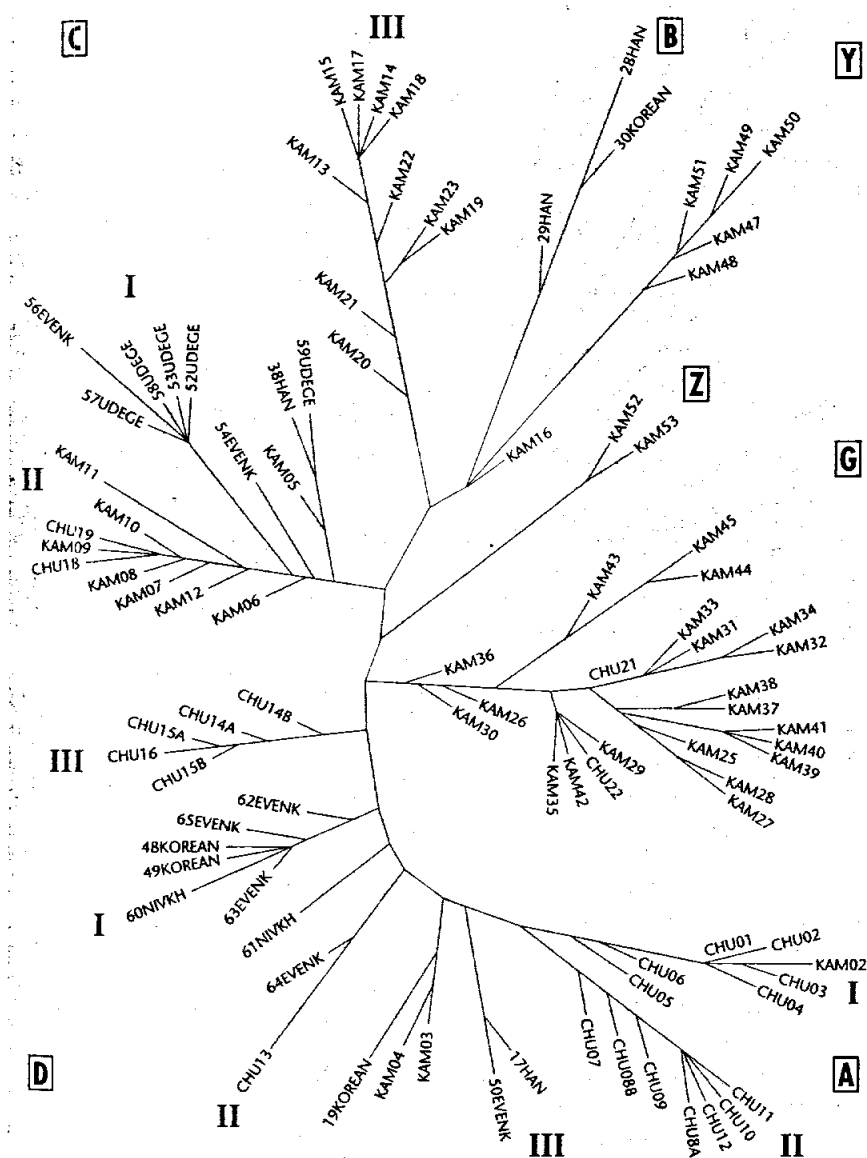


Figure 5. An unrooted NJ tree based on genetic distances estimated from CR sequences from northeast Siberian and East Asian populations with the Kimura two-parameter model (Kimura 1980) in DNADIST (Felsenstein 1994). Samples are indicated as numbered in Torroni et al. (1993b), Starikovskaya et al. (1998), and Schurr et al. (1999). The Roman numerals specify the distinct sublineages which appear in haplogroups A, C, and D (see text), while all haplogroups are indicated by the boxed capital letters.

ing haplotype in both sets of populations (Torroni, Schurr, et al. 1993; Torroni, Sukernik, et al. 1993). However, all other haplotypes with this motif occurred in either Siberian/Asian or Native American tribes (Schurr et al. 1999; Torroni et al. 1992; Torroni, Chen, et al. 1994; Torroni, Neel, et al. 1994; Torroni, Schurr, et al. 1993; Torroni, Sukernik, et al. 1993). A related sublineage (II) defined by the np 16124 T->C (16124C) transition and np 16318 A->T (16318T) transversion occurred only in Paleoasiatic populations of northeastern Siberia (Schurr et al. 1999; Starikovskaya et al. 1998). Interestingly, this subbranch was associated with both putative founding haplotypes SIB26 and SIB45 of this haplogroup. Since both of these haplotypes differed from equivalent haplotypes in Native American populations (AM32 and AM43; Torroni et al. 1992; Torroni, Schurr, et al. 1993; Torroni, Sukernik, et al. 1993) by having the 16124C and np 16318T mutations, they must have arisen in Paleoasiatic groups independent of seemingly identical mtDNAs in New World populations. Thus, these haplotypes cannot be considered founding mtDNAs for Native American populations.

The third sublineage (III) differed from the first by having the np 16093 T->C (16093C), np 16189 T->C (16189C), np 16261 C->T (16261T), and np 16288 T->C (16288C) transitions, and lacking the 16298C and 16327T mutations. As seen in other Asian and Native American mtDNAs, the 16189C transition creates a homopolymeric stretch of C's within a 14-bp hypervariable domain (np 16180-16193), which typically resulted in the insertion of an additional one or more C's (Horai et al. 1993; Horai and Hayasaka 1990). This sublineage also only occurred in Paleoasiatic-speaking populations from Kamchatka and Chukotka (Schurr et al. 1999; Starikovskaya et al. 1998), although related types may be present in other eastern Siberian groups (Torroni, Sukernik, et al. 1993). Moreover, a subset of SIB26 haplotypes also had this CR sequence motif, implying that they, too, must have arisen in Paleoasiatic groups after ancestral Asian populations entered the New World.

Similarly, haplogroup D had several sublineages within it. The first sublineage (I) consisted of mtDNAs from East Asian and eastern Siberian populations having the CR sequence motif most similar to haplogroup D mtDNAs in Native American populations (16223T, 16362C) (Torroni, Schurr, et al. 1993; Torroni, Sukernik, et al. 1993). Because the nodal haplotypes for this haplogroup (SIB13/AM88) had this motif, they probably represent the founding haplotypes in both sets of populations (Torroni, Schurr, et al. 1993; Torroni, Sukernik, et al. 1993). In contrast, the other two lineages occurred almost exclusively among northeastern Siberian populations. The second (II) was present at very low frequencies among Paleoasiatic groups and Eskimo populations (Shields et al. 1993; Starikovskaya et al. 1998) and perhaps East Asians (Ballinger et al. 1992; Horai et al. 1996), while the third (III) was confined to only Chukotkan populations (Starikovskaya et al. 1998). Thus, like haplogroup C, this mtDNA lineage has undergone considerable differentiation in various regions of Asia since the peopling of the New World.

In contrast to the other haplogroups, only Central and East Asian mtDNAs are found within haplogroup B (Ballinger et al. 1992; Horai et al. 1996; Kolman et al. 1996; Lum et al. 1994; Merriwether et al. 1996; Redd et al. 1995). This haplogroup

has a specific CR sequence motif (16189C-16217C-16519C) and a founding haplotype (AS54/AM13), which are seen in both Asian and Native American mtDNAs that belong to this mtDNA lineage, thereby confirming their genealogical relatedness. Of the other Asian mtDNA lineages not present in Native Americans, haplogroup G was composed of only Koryak, Itel'men, and Chukchi mtDNAs (Schurr et al. 1999; Starikovskaya et al. 1998). This haplogroup also showed some degree of substructure, particularly when other Asian mtDNAs were added to the analysis. In contrast, haplogroups Y and Z formed small unbranched clusters of mtDNAs, and were present in only the Koryak and Itel'men (Schurr et al. 1999).

A closer examination of CR sequence diversity in haplogroup A revealed additional subbranches of phylogenetic importance (Figure 6). As previously shown, two main clusters of haplotypes appear in haplogroup A, one having the 16111T mutation that characterizes nearly all Chukotkan and Native American mtDNAs from this mtDNA lineage, and another lacking this mutation, to which most Asian and Siberian mtDNAs belonged. Within the first main cluster, a large sublineage defined by the 16192T mutation delineates recently evolved Beringian haplotypes from those common to all Native American groups. In addition, both of these sublineages had several population- or ethnic-specific clusters, including those occurring in Eskimos, Na-Déne Indians, Haida/Bella Coola, and NW Coast Amerindians. This extensive differentiation of haplogroup A CR sequences was mirrored by the diversity of its RFLP haplotypes, as well as the distribution of CR sequences from haplogroups B-D, in the same populations (Schurr et al. 1999; Shields et al. 1993; Starikovskaya et al. 1998; Torroni et al. 1992; Torroni, Sukernik, et al. 1993; Ward et al. 1991, 1993). These findings indicated that most of the sequence diversity within haplogroup A accumulated in the circumarctic regions of Asia and the Americas, and that ancient Beringia was the geographic area in which much of haplogroup A sequence diversity accumulated before and after this mtDNA lineage was brought to the New World.

## **Ages of mtDNA Haplogroups in Siberia and the Americas**

To estimate the ages of the primary mtDNA lineages present in Siberia and the Americas, we estimated their sequence divergence using the maximum likelihood (ML) method of Nei and Tajima (1983) (Table 4). This analysis showed that haplogroup C was one of the oldest mtDNA lineages in both Siberia and America, with the divergence times from a common founding haplotype (SIB26/AM43) being c. 27,000–36,000 yr B.P. in Siberia, and c. 33,000–43,000 yr B.P. in America. The sequence divergence of haplogroup D from the ancestral haplotype SIB13/AM88 was also comparable to that of haplogroup C, with the highest divergent time being c. 33,000–44,000 yr B.P. in Siberia. However, the divergence time for haplogroup D in Native American populations was found to be only 24,000–18,000



By contrast, the divergence values for haplogroup A in Siberia and the Americas were quite different. The estimated sequence divergence in Siberia was 0.028 percent, a value considerably less than 0.079 percent for the Americas, and these values gave correspondingly different divergence times for Siberia (10,000–13,000 yr B.P.) and the Americas (27,000–36,000 yr B.P.). This apparent discrepancy is likely due to the fact that most of the Siberian haplogroup A mtDNAs were taken from populations inhabiting Chukotka (i.e., the Chukchi and Siberian Eskimos), and that the founding populations of the Chukchi, Eskimo-Aleuts, and Na-Dene Indians underwent one or more genetic bottlenecks, then re-expanded in the Beringian region. This interpretation is supported by the star-shaped phylogeny of its CR sequences with many deep branches (Figure 6). Under such conditions, the resulting small populations would have experienced significant fluctuations in population size, along with reduced genetic diversity, thereby causing an underestimation of the age of haplogroup A (Haeseler et al. 1996). This scenario is also consistent with recent estimations of the antiquity of haplogroup A in the former Beringian region (Bonatto and Salzano 1997a).

In contrast, the divergence estimate for haplogroup B in the New World of c. 13,000–17,000 yr B.P. was considerably smaller than that of haplogroups A, C, and D. This ML value, along with the near absence of deletion haplotypes in Siberian populations, and the predominance of the founding haplotype for this mtDNA lineage, AM13, in Native American populations (approx. 60 percent of all haplotypes analyzed) (Huoponen et al. 1997; Torroni et al. 1992; Torroni, Chen, et al. 1994;

Table 4. Sequence divergence of mtDNA haplogroups in Siberia and the Americas.

haplogroup	geographic region	n	N	sequence divergence (%)	divergence time (yr B.P.)
A	Siberia	11	119	0.028	12,727–9,655
	America	46	189	0.079	35,909–27,241
B	America	30	99	0.039	17,727–13,448
C	Siberia	14	123	0.043	19,545–14,828
	America	31	72	0.122	55,545–42,069
D	Siberia	13	47	0.111	50,455–38,276
	America	16	62	0.057	25,909–19,655
G	Siberia	11	106	0.024	10,909–8,276
Y	Siberia	7	58	0.014	6,364–4,828
Z	Siberia	4	12	0.021	9,545–7,241

Sequence divergence of native Siberian and Native American mtDNA haplogroups. In this table, n = number of haplotypes, and N = number of individual mtDNAs, for each haplogroup. The sequence divergence estimates were weighted by the number of individuals within each haplogroup. Divergence times were calculated by multiplying the haplogroup sequence divergences by the mtDNA evolutionary rate of 2.2%–2.9% per myr (Torroni, Neel, et al. 1994). The divergence estimates for the Siberian haplogroups are based on data presented in Torroni, Sukernik et al. (1993), Starikovskaya et al. (1998), and Schurr et al. (1999), whereas all values for Native American haplogroups are based on data presented in Torroni et al. (1992), Torroni, Schurr et al. (1993), Torroni, Neel et al. (1994), Torroni, Chen et al. (1994), and Huoponen et al. (1997).

Torrioni, Neel, et al. 1994; Torrioni, Schurr, et al. 1993; Torrioni, Sukernik, et al. 1993), continues to imply that haplogroup B mtDNAs were brought to the Americas in a later and separate migration from the earlier one bringing haplogroups A, C, and D. However, recent estimates of CR sequence diversity within haplogroup B in Native American groups suggest that this mtDNA lineage was present in the New World by 25,000–30,000 yr B.P. (Bonatto and Salzano 1997b). This date would mean that haplogroup B could have arrived in the Americas at about the same time as haplogroups A, C, and D. While the source of the discrepancy between these divergence estimates is not entirely clear, it may be attributable to differences in the assumptions of the statistical methods used to make these estimates, or the sampling of Amerindian populations with high frequencies of closely related deletion haplotypes (Torrioni, Chen, et al. 1994; Torrioni, Neel, et al. 1994; Torrioni, Schurr, et al. 1993).

The remaining haplogroups in Northeast Siberian populations (G, Y, and Z) appear to have arisen in eastern Asia during postglacial times, and apparently never reached Beringia at a time when they could have been brought to the Americas. Consistent with this hypothesis, the divergence times of haplogroups G, Y, and Z are considerably shallower than those of haplogroups A–D in Siberia and the Americas. Of these mtDNA lineages, haplogroup G was the oldest and most diverse mtDNA lineage, a result paralleled by its broader distribution within Asia itself (Ballinger et al. 1992; Horai et al. 1984; Schurr et al. 1999; Starikovskaya et al. 1998; Torrioni, Miller, et al. 1994; Torrioni, Sukernik, et al. 1993). These lower divergence time estimates for haplogroup G, Y, and Z further indicate that none of the putative “other” haplotypes recently observed among Native American populations are likely to belong to these three Asian haplogroups.

## **Ancient Peopling of the New World**

Based on these phylogenetic and statistical results, we can attempt a reconstruction of the genetic prehistory of Siberia and the New World. Recent archaeological records indicate that, by 40,000 yr B.P. or even earlier, the Upper Lena–Lake Baikal Region of east Siberia was populated by modern humans (Goebel and Aksenov 1995). Between 30,000 and 35,000 yr B.P., Paleolithic Siberian populations underwent geographical expansions in the climatically favorable southern belt of Siberia and the adjacent tundra-steppe of the Siberian Arctic and Subarctic under conditions of unlimited availability of easily hunted big game. In addition, archaeological data from this region (Dikov 1994; Goebel and Aksenov 1995; King and Slobodin 1996; Wright 1991) make it clear that early eastern Siberian prehistory was more complex and diverse than traditional colonization models have indicated. They further imply that several different archaeological traditions that existed in eastern Siberia between 15,000 and 30,000 yr B.P. could possibly have entered the New World from ancient Beringia.

Biogeographical data suggest that the most propitious time for the first entry of humans into the New World from Beringia occurred during this same period, when



deglaciation was sufficient to form an "ice-free corridor" from western Alaska to the Great Plains. The subsequent entry into the New World during the last glacial maximum (c. 26,000–15,000 yr B.P.) was unlikely, since, during this period, glacial coalescence would have prevented further human dispersal southward out of the Beringian region. With the global climate change that brought the Ice Age to an end, the ice-free corridor opened again around 11,000 yr B.P. (Lemmen et al. 1994), presumably permitting a second wave of human immigrants to spread southward throughout North and South America (Butzer 1991; Wright 1991).

Several primary lines of evidence support this general model. First, the broad geographic distribution of haplogroups A, C, and D, matched with the ethnic and regional histories of Siberian populations in which they occur (Levin and Vasiliev 1964; Simchenko 1976; Tugolukov 1979, 1985; Vasilevich and Smolyuk 1964), indicates the origin of these mtDNA lineages in southeastern Siberia, probably the Altai-Sayan/Baikal region. Although absent in eastern Siberians, its distribution in Central-East Asia suggests that haplogroup B may also have originated in the same general region (Ballinger et al. 1992; Kolman et al. 1996; Redd et al. 1995).

Secondly, when divergence sequence estimates for haplogroups A, C, and D were averaged, they gave a divergence time of approximately 28,000–35,000 yr B.P. for these mtDNA lineages in both Siberia and America, dates which coincided with an open Alberta corridor. Since previous work among Asian populations has shown that haplogroup B haplotypes were present in Central-East Asia by at least 24,000–30,000 yr B.P. (Ballinger et al., 1992; Horai et al. 1996; Lum et al. 1994), and since recent estimates suggest that haplogroup B is as diverse as haplogroups A, C, and D in the New World (Bonatto and Salzano 1997a, b; Lorenz and Smith 1996), this mtDNA lineage may also have entered the New World during this period. Thus, the molecular data suggest that the four primary haplogroups in Native Americans were brought to the New World before the last glacial maximum.

Third, various phylogenetic analyses of Siberian, Asian and Native American mtDNAs have shown that these sets of populations are quite divergent from one another (Schurr et al. 1999; Starikovskaya et al. 1998; Torroni, Schurr, et al. 1993; Torroni, Sukernik, et al. 1993). Aside from having the putative founding haplotypes for haplogroups A (SIB41/AM09), B (AS54/AM13), C (SIB26/AM43) and D (SIB13/AM88) in common, the remaining haplotypes in these populations were largely population- or region-specific. In fact, even within the shared founding haplotypes, a number of CR sequence sublineages unique to either Siberian or Native American populations have evolved. Taken as a whole, these data indicate a considerable antiquity of the primary mtDNA lineages shared between Siberians and Native Americans, and their extensive divergence from each other since being isolated in each continental region.

How these mtDNA data can be reconciled with archaeological data for Paleoindian sites is becoming clearer. Not including the archaeological studies of North American sites described in this volume, recent archaeological data have identified at least two ancient and distinct cultures of Paleoindians in the Americas. Long suggested to represent the first immigrants to the Americas (Haynes 1992; Lynch 1990),

the Clovis lithic tradition apparently consisted of specialized big game hunters who were adapted to open, temperate, terrestrial habitats with the technology of fluted bifaces. However, Bryan (1991) and Stanford (1991) suggest that the Clovis lithic tradition may have instead originated in the New World somewhere south of the North American ice sheet about 11,000–13,000 yr B.P., and then spread throughout much of North and Central America. In addition, a second cultural tradition present in the Amazon basin had a different subsistence pattern based on gathering fruits and nuts, fishing and hunting small animals, and appeared to be at least contemporaneous with North American Clovis sites (Roosevelt et al. 1996). These data, along with findings at the Monte Verde site in southern Chile (Dillehay 1997), suggest that the First Americans may have arrived much earlier than the ancestors of the Clovis lithic tradition.

If this general scenario developed from the archaeological data is correct, it implies that at least two distinct cultural traditions existed in different parts of the New World by c. 15,000 yr B.P. This time depth, in turn, implies that humans had reached the Americas before the last glacial maximum, since travel through the interior of North America would have been impossible between roughly c. 15,000–25,000 yr B.P. Such an earlier entry time would have allowed for the diversification of lithic traditions in the different continents of the Americas where ancestral Amerindian populations had spread, hence, the emergence of the Clovis culture at the same time that different cultures were developing in the Amazon and Chilean areas. On the other hand, if the Clovis culture truly represents a distinct migration from Siberia to the New World, where human populations were apparently already established, this event would mean that the Paleoindian gene pool was at least bipartite in origin. Such a model is congruent with our previous model for the origin of the Paleoindians, in which haplogroups A, C, and D were brought to the Americas in an early migration (c. 30,000 yr B.P.), and haplogroup B mtDNAs in a second and later migration (13,000–17,000 yr B.P.), although recent claims for the antiquity of haplogroup B argue against this scenario.

Alternatively, Asian populations could have entered the New World through both an interior and a coastal route, as proposed by Gruhn (1987, 1988), Fladmark (1979), and Nichols (1990, 1994). Such a model could account for the different subsistence strategies and lithic traditions of archaeological cultures in different parts of the Americas, as well as their linguistic diversity, and might explain the problem of the chronological simultaneity of these traditions. However, Hoeffecker et al. (1993) have questioned the availability of such a coastal route between 26,000–15,000 yr B.P., stating that such a coastal route would have to have been used before or after the last glacial maximum. Gruhn (1994, 1997) has suggested precisely this scenario, that ancestral Paleoindians had utilized the coastal route to enter the New World by 50,000 yr B.P., a time depth which would have allowed for the extensive linguistic diversification of Amerindian populations. Since this date is earlier than the estimated ages of the primary mtDNA lineages that were brought to the Americas during its initial colonization (Table 4), these alternative scenarios will require further testing with archaeological, linguistic and genetic data.

Fitting haplogroup X and the "other" mtDNAs found in Native American groups into this picture is also tricky. The cumulative evidence suggests that, once ancestral Native Americans had reached Beringia, their gene pool consisted of at least haplogroups A, C, and D, and possibly haplogroup B. By contrast, the origins and timing of entry of haplogroup X mtDNAs remain somewhat ambiguous, although it appears to have arrived in the New World prior to the last glacial maximum (Brown et al. 1998). Irrespective of when it was brought to the Americas, the lack of haplogroup X mtDNAs in Siberian groups (Schurr et al. 1999; Schurr et al. unpublished data; Starikovskaya et al. 1998; Torroni, Sukernik, et al. 1993) suggests that these haplotypes originated in a region outside of eastern Siberia, and were taken through Beringia by ancient Eurasian populations, where they became part of the original colonization of the New World.

## Reexpansion of Ancient Beringian Populations

In addition to showing an ancient origin of the First Americans, the mtDNA data have revealed strong evidence for the subsequent re-expansion of ancient Beringian populations into northern North America after the last major period of glaciation. The 16111T polymorphism in haplogroup A mtDNAs marks the emergence of the ancestral Beringian populations which gave rise to all Native American groups. After the initial occupation of the New World, these ancient Beringian populations were isolated and became genetically divergent from the ancestral Amerindian groups to the south for a considerable period of time. Among these last inhabitants of Beringia evolved the large "North Pacific Rim" branch of haplogroup A defined by the 16192T mutation whose mtDNAs are found in only the Koryaks, Chukchi, Siberian and Alaskan Eskimos, and Na-Dene Indians, as well as a number of population- or region-specific clusters within these two major sublineages of haplogroup A (Figure 6). Such results apparently reflect the emergence of remnant populations isolated in biogeographic refugia in Beringia and southern Alaska that existed until the end of the last glacial maximum (Rogers et al. 1991).

Furthermore, the clear affinity of aboriginal Chukotkan populations with Native Americans as revealed by mtDNA variation was also seen in parallel studies of Y-chromosome haplotype polymorphisms. Specifically, it has been shown that a C->T transition at np 181 (181T) of the DYS199 locus is broadly distributed in all Native American populations (Lell et al. 1997; Underhill et al. 1996), but has been found in Siberia only among the Chukchi and Siberian Eskimos (Lell et al. 1997). Hence, like the 16111T polymorphism in haplogroup A mtDNAs, the DYS199 181T mutation apparently occurred in the ancient Beringian populations which gave rise to all Native American populations (Lell et al. 1997). Thus, both the mtDNA and Y-chromosome data reveal the genetic differentiation of northeastern Siberian populations from other northern Asian groups, as well as their genetic links to New World populations.

## Origins of Paleoasiatic-speaking Populations

A remarkable finding in these studies was the genetic discontinuity between Paleoasiatic-speaking populations of Chukotka and Kamchatka. Both the distribution of haplogroups A–D, G, Y, and Z haplotypes in the Koryaks, Itel'men, and Chukchi and several different measures of haplotypic diversity showed Kamchatka populations to be genetically very similar to one another, and quite divergent from the linguistically related Chukchi, who were more similar to the Siberian Eskimo (Schurr et al. 1999; Starikovskaya et al. 1998). In addition, despite a third of the gene pool consisting of haplogroup A, C, and D mtDNAs, Kamchatkan groups were quite distantly related to Native American populations, sharing only founding haplotypes from haplogroups A (SIB41/AM01) and C (SIB26/AM43) (Schurr et al. 1999). Moreover, these analyses clearly show the strong genetic affinities of the Paleoasiatic populations of Kamchatka with the Ainu as well as Amur River-Sakhalin groups such as the Nivkhs and Udegeys, implying that ancestral Paleoasiatic population arose and expanded into northeast Asia from this general region (Schurr et al. 1999).

The genetic history of the Koryaks and Itel'men is, therefore, concordant with the prehistory of northeastern Siberia. Archaeological evidence indicates that, since 14,000 yr B.P., the proto-Eskimo-Aleut cultures of the Asiatic coastline of the Bering Sea were gradually replaced by cultures having different subsistence strategies and lithic technologies. Eventually, the entry of ancestral Koryaks and Itel'men led to the near total absorption of the ancient Bering Sea cultures in Kamchatka, with different varieties of the ancient Koryak culture diffusing extensively along the Okhotsk Sea and coastline of the northwestern Pacific (Arutiunov and Sergeev 1990; Dikov 1994; Vasilievskiy 1964, 1971). Thus, both the genetic and archaeological data indicate that multiple population and/or cultural expansions have taken place in the Okhotsk Sea and Bering Sea region over the last 10,000 years, with more recent evolved genotypes and cultural traditions from East Asia overlapping more ancient ones (Aikens and Rhee 1992; Arutiunov and Fitzhugh 1988; Arutiunov and Sergeev 1990; Dikov 1994; Fitzhugh and Crowell 1988; Vasilievskiy 1964, 1971).

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