Genetic structure and affinities among indigenous populations of northeast Siberia: a literature review

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Abstract

The Asian Eskimo, Chukchi, Koryak and Itel'men are indigenous Siberian ethnic groups that occupy the territory of Northeast Asia. Russian and Western researchers have studied the genetic structure of these groups, and examined the relative role of microevolutionary forces operating on these populations. Genetic affinities between these and other Siberian native groups have been estimated to infer phylogenetic relationships. Overall, it was found that genetic structure was shaped predominantly by migration and random genetic drift. Genetic affinities reflect ethnohistory of Northeast populations and showed that in general, the Chukchi and Asian Eskimo are genetically as similar to Beringian Eskimo as to populations of continental Northeast Siberia. More specifically, Coastal Chukchi are genetically as similar to Reindeer Chukchi as to Asian Eskimo. Koryak were found to be closer to Reindeer Chukchi than to Asian Eskimo. Koryak and Itel'men are generally equidistant to continental Northeast populations and to Far East populations.

Admixture plays an important role in estimating genetic affinities between populations. Undetected admixture can seriously obscure the results of genetic studies and make interpretations incorrect. To realistically estimate the degree of admixture, demographic and genealogical data must be collected for genetic studies. However, a survey of sampling procedures demonstrated relatively simplified approach to this problem. Genealogical studies were rarely employed for estimating admixture in individuals composing a sample. In most cases, individuals were only selected on the basis of "self-assigned" ethnicity with no further verification.

This thesis proposes a new approach for conducting genetic studies that focus on genetic structure and affinities. Social anthropologists should collect and analyze genealogical data, and then samples for genetic study should be drawn from individuals that have been carefully selected by social anthropologists. Furthermore, a number of genetic markers, such as blood groups, HLA, mtDNA and Y chromosome polymorphisms should be analyzed simultaneously. Each of these markers provides unique information about migration and other microevolutionary forces. Using this

approach, a more realistic picture of genetic affinities between populations should be obtained.

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Introduction

Genetic studies of indigenous Siberian populations have been conducted by Soviet and Western researchers for a number of reasons. First, thirty years ago the semi-nomadic and nomadic native groups that occupy vast territories of Northern, Central, and Northeast Asia were virtually unknown to those studying human genetic diversity and evolutionary biology. These populations were extensively studied by archeologists and ethnographers, who described their considerable linguistic and ethnographic diversity. They also reconstructed the history of these populations, and speculated about peopling of Asia. Despite major changes in their social, political, and economic life introduced by the Soviet regime, some of indigenous Siberians still maintained traditional lifestyles as fishermen, reindeer herders, and sea hunters. They also spoke their native languages and retained their distinctive cultures. Their populations typically had low density and non-contracepting reproduction. Taken together, these factors were instrumental in initiating an interest in genetic studies of indigenous Siberian populations. Secondly, circumpolar Siberian populations have lived in harsher ecological conditions of the Arctic and sub-Arctic for generations and they may have undergone adaptation to these climatic conditions. Thus, for scientists who wished to understand how genetic adaptation was reflected in the genetic structure of populations, these were interesting groups. Thirdly, in addition to the study of microevolutionary forces, scientists also consider the problem of migration into the New World. This involves the genetic, archaeological, historical, and anthropological studying of indigenous Siberian populations, since the route to the Americas is thought to go through Northeast Asian part of Russia.

Population genetic studies of native Siberian inhabitants were initiated by Soviet researchers at the end of 1960s. These studies, which were published in Russian, focused on descriptions of blood groups and protein polymorphisms. Over the past 30 years considerable information has accumulated regarding genetic structure, blood group and protein polymorphisms, mitochondrial and nuclear DNA diversity and Y-chromosome polymorphisms. However, comprehensive syntheses remain absent. The purpose of this thesis is to provide an overview of population genetic studies of selected indigenous Siberian populations, to summarize the data available at this time. For this purpose, Russian and English language articles, published over the past 20 years were analyzed. The original scope covered the ethnic groups: Chukchi, Eskimo, Koryak, Itel'men, Nivkh, Yukagir, Sakha (Yakut), Nganasan, Nentsi, Evenk, Even, Buryat, Altai, Tuva, Tof, and, Tofalar, from Central, South-central, North, and Northeast Asian territories of Russia. However, summarizing information on all these peoples and territories proved impossible given time and word limitations. Therefore, native peoples of Northeastern Asia, Chukchi, Asian Eskimo, Koryak, and Itel'men are the focus of this dissertation.

In this thesis, I start with a short outline of the archaeological and ethnographic background of these populations (Chapter I). It will be seen that much of this remains hypothetical and speculative. I then move on to a detailed examination of genetic studies (Chapters 2-4). Finally, in Chapter 5 I shall return to the problem of ethnic categories as they affect sampling procedures and assumptions. Statistical methods are an important part of any genetic analysis. However, a review of all the statistical methods used in the analyzed articles would exceed the scope of this thesis. Therefore, only a brief account of

some statistical methods will be provided. For ease of reference Tables and Figures are gathered together in an Appendix at the end of the thesis.

Chapter I. The Native Peoples of the Northeast Asia: Eskimo, Chukchi, Koryak, and Itel'men.

I. 1 *Archaeological and ethnographic data on their origins and affinities.*

Accumulated archeological, historic, ethnographic and physical anthropological data has allowed researchers to develop theories and hypotheses regarding place and dates of origins and population movements of Northeast Asian populations. However, these theories have not been at all conclusive in their findings and genetic evolutionary studies hope to clarify some of the existing uncertainties. It is crucial to understand the history of the ethnic groups under study, because a correct interpretation of the results of genetic studies is impossible without a thorough understanding of the historic context. Failure to recognize the importance of historic events, ancient and recent, might lead to seriously obscured interpretation of genetic results. This chapter will introduce some of the theories and hypotheses regarding origins of Northeast Asian groups, and outline some of the important recent events in their history for correct interpretation of genetic findings (for an authoritative summary of the ethnography of Siberian native peoples, see Vakhtin, 1992, Vitebsky, 1996; for more extended historical discussions see Armstrong, 1965, Forsyth, 1992, Slezkine, 1994).

In general, archaeological material from the coast of the Sea of Okhotsk, Kamchatka Peninsula, and parts of the Chukotka Peninsula, shows that ancient Northeast Paleoasiatic groups were formed by Neolithic hunters who came from Yakutia and the Amur River region. Perhaps they mixed with the more ancient Mesolithic inhabitants of this area and partially inherited their culture (Gurvich 1980, 1985). According to the

indirect evidence of the archaeological data from the Americas, the appearance of modern humans in Northeast Asia dates to approximately 30,000 years ago (Cavalli-Sforza *et al*, 1994), although the ethnic groups under analysis in thesis appeared there much later. The discoveries of the Ushki site, with typical Mesolithic artifacts on the Kamchatka Peninsula dated by the radiocarbon method to 10-11 thousand years, and of Sumnaginskaya culture in Sakha (Yakutia) dating back 8-5 thousand years, show that separate, sparse groups of hunters were coming to Northeast Asia in the post-glacial period. Supposed traces of combined ancient Mesolithic and early Neolithic cultures of nomadic hunters and fishermen have been identified in the cultures of many modern Siberian and North American populations. During this period, as today, Northeast Asia was already part of an extended cultural area that included the Lake Baikal region, the Amur River region, northern Mongolia, and a considerable part of Northeastern Siberia. Vasil'evskii (1973, 1974) having summed up all the archaeological evidence, produced a hypothesis according to which cultural influence was coming to the north and east from Eastern Mongolia, where the initial formation of Mesolithic cultures took place. This hypothesis is supported by physical anthropological data, where dark skin, black coarse hair, and the characteristic Mongoloid structure of the upper eyelid, point to the probable affinities of the bearers of such characteristics with the inhabitants of what Debets called "eastern Asia", i.e., presumably China and Mongolia (Debets, 1951). Perhaps these waves of nomads also brought the predecessor of what are known as Paleoasiatic languages. The Chukotka-Kamchatkan languages, as shown by Bogoras (1928), have components found neither in Eskimo nor in any American Indian languages. Ancient Koryak tools show clear continental features. Also, Koryak pottery, which has not been

found on the coast of the Sea of Okhotsk prior to the arrival of the continental hunters, has similarities with the pottery of the Neolithic cultures of the Yakutia and Lake Baikal regions. Ancient Koryak culture was also influenced by the cultures of the Priamur'ye (groups living along the Amur River) and Primor'ye (Far East cultures). Archeological data indicate that ancient Koryaks had a design of semi-subterranean dwellings similar to Koryak dwellings belonging to later dates, and to Neolithic dwellings of the lower Amur River cultures that existed 4-5 thousand years ago (Okladnikov, 1953, Vasil'evskii, 1973). Coastal Koryak group was probably formed during this period of Neolithic marine-hunter culture, and perhaps even earlier.

I. 1. 1 **Chukchi, Koryaks, and Itel'men**

In 1989, Chukchi numbered 15,100, with 11,900 living in the Chukotka Autonomous area, another 1,300 living in the Republic of Sakha, and 1,500 inhabiting the Koryak Autonomous area. Koryaks numbered 8,900, with 6,600 living in the Koryak Autonomous territory of the Kamchatka region, and the rest occupying Chukotka and North-Evenk district of the Magadan region. The Chukchi are said to belong to the Chukchi-Kamchatkan language family, together with Koryak and Kamchadal (Itel'men) (Funk and Sillanpaa, 1999). However, the Chukchi and Koryak languages are more similar to each other than to the Itel'men language. This language family has some similarity with the Eskimo-Aleut language family (Ruhlen, 1987). The Chukchi and the Koryaks are similar not only in language, but also in various aspects of their material and spiritual culture. Until very recently, both were divided into the coastal hunters of sea mammals and reindeer herders that inhabit the interior of Chukotka Peninsula. Curiously,

in their economy, life, and culture, there were more similarities between the Reindeer Chukchi and Koryaks than between the Reindeer Chukchi and Coastal Chukchi (Levin, 1956). As for Reindeer and Coastal Koryaks, they spoke different dialects and rarely intermarried (Gurvich, 1980).

Chukchi and Koryak are thought to come from the region that lies to the south of their present territory. From here, the ancestors of the Chukchi spread north, assimilating the Eskimo and in turn being influenced by Eskimo language and culture. Archaeological data indicate that in ancient times the territory between Cape Schmidt to Cape Dezhnev was inhabited by the Eskimo. Today most of this territory is occupied by Chukchi. Apparently, these two ethnic groups underwent a process of fusion, with the Chukchi language replacing Eskimo language. As a result, present coastal Chukchi exhibit traces of Eskimo influence in their culture, economy and life style (Levin, 1956).

Chukchi folk stories reflect clashes between the Chukchi and Asian Eskimo as well as Chukchi and Koryaks. Although even the approximate dating of these folk stories is problematic, Bogoras considers stories about clashes between Chukchi and Eskimo to be older. Besides wars, the intertribal barters with the neighboring native groups, Sakha, Eskimo of Alaska and St. Lawrence Island, Yukagirs, Koryaks, and Evens were also known. The Chukchi had patrilineal ties of kinship, patrilocal families; and they practiced polygamy and "group marriage" (Tishkov, 1994). During the 18th and 19th century expansions of the Reindeer Chukchi, they assimilated Asian Eskimo, Yukagirs, Evens and married captured American Indian women. In the 19th century, East Reindeer Chukchi moved to Kamchatka where they were assimilated by Koryaks, though many of them still spoke the Chukchi language (Gurvich, 1982). Thus, it is clear that the Chukchi

and the Koryaks should exhibit genetic affinities not only due to their common ancestral origin, but also due to admixture documented during at least the last few centuries and probably also earlier. Such admixture also took place between the Chukchi and Yukagirs, Asian Eskimo, and, to some extent, Evens.

The question of the origin of the Itel'men, according to Levin and Potapov (1956), remains open. Later works however, shed some light on this problem. Itel'men (or Kamchadal) in 1994 numbered 2,429 individuals, are thought to have stemmed from the same Mesolithic nomadic hunters as the other two Paleoasiatic groups, though the Itel'men language is quite different from the Chukchi and Koryak languages. Vdovin (1970) attributes this to the earlier formation of the Kamchatka culture compared to the Ancient Koryak culture. Vdovin considers the peculiarities of the Itel'men culture and language as indicative of their origin being different from Koryak and Chukchi, and views common traits as a result of the historic contacts between these three groups. There are observed similarities between the Itel'men culture and cultures of the Kuril Island, Amur River, Far East cultures, and also with the American Eskimo, Athapaskans and Tlingit. These similarities are manifested in the construction of dwellings, dugout boats, the way of weaving and use of nettle to weave nets and floor mats (Gurvich, 1980). This indicates that ancestors of Itel'men have probably come from the regions of the Lena River, Amur River and the Far East, and, having mixed with the remains of older Eskaleut inhabitants, partially inherited elements of their cultures. The arrival of Russians in Kamchatka in the 17th century had a devastating effect on the Itel'men. Their number started to decrease due to epidemics, famines, clashes between them and Cossacks and intertribal wars. Starting with the 18th century, an extensive admixture of Itel'men

women with Cossacks and Russian traders and prospectors led to substantial changes in social structure and culture of the Itel'men, loss of their language and ethnic identity (Tishkov, 1994). This tendency accelerated in the 20th century resulting in the descendants of Itel'men being considerably admixed with Russians, except for a few villages.

Chukchi and especially Coastal Chukchi cultures have many elements in common with Eskimo, such as linguistic similarities, rituals, holidays, and hunting methods, that are indicating that the history of the Asian Eskimo is tightly intertwined with the history of Chukchi.

I. 1. 2 **Asian Eskimo**

Present day Asian Eskimo populations occupy the far Northeast Asiatic Russia. They live in very close proximity to the Coastal Chukchi, sometimes in the same communities, while the Reindeer Chukchi occupy inland of Chukotka Peninsula. According to the 1989 Census, there are about 1,7 thousand Asian Eskimo in Russia.

Their language belongs to the Eskimo-Aleut language family, which spreads eastward across the American Arctic into Greenland. Ruhlen (1987) further subdivides the Siberian branch of the Yupik division of the Eskimo linguistically into three dialects: Navukagmit ("Naukantsy"), Ungazigmit ("Chaplintsy"), and Sirenigmit ("Sireniktsy"). The Sirenigmit dialect has practically disappeared.

There are many theories concerning their origin and culture (Gurvich 1980). Most researchers agree on the roots of Eskimo culture being in Asia. The discrepancies are concerned with the time and exact place of the formation of the Eskimo culture, as well

as which ancient cultures of Northeast Asia and the American Arctic should be considered to be Eskimo, and about the genetic affinities between them. There are two main points of view on the probable region of the final formation of what we now call Eskimo culture. According to the first (Sergeev, 1974), it happened in the Asiatic region of the Bering Strait (Eastern Chukotka), the earliest known Eskimo culture being Ancient Beringian, the earliest stages of which were dated to a little over 2,000 years ago. According to the second point of view, the final shaping of Eskimo culture took place in Southeast Alaska, from where it spread to the north, to the Bering Strait, and then to the east, to Greenland, and finally to the west, to the Chukotka Peninsula. Chard, Oswalt, Bandi, and Laughlin support this second hypothesis. Gurvich's hypothesis based on the analysis of data from Soviet and Western researchers, states that the division of proto-Eskaleuts into proto-Eskimos and proto-Aleuts took place in Northeast Asia. It most likely happened on the Bering land bridge, approximately 8-10 thousand years ago. Alternatively, this might have happened somewhere to the south of Chukotka. The Eskimo then crossed the Bering land bridge and spread first to Southwest Alaska, and then north and east to Greenland. Inupiaq-speaking Eskimo continued their northwards and eastwards migration, leaving Yupik-speaking Eskimo to inhabit Southwest Alaska. The descendants of the latter then moved to the Chukotka Peninsula and, interacting with other indigenous non-Eskimo native groups, especially at Ust'-Bel'skaya, founded the Ancient Beringian culture. The fact that there were no proto-Eskimo artifacts found on the Chukotka Peninsula as old as those found in the American Arctic supports the hypothesis that the Eskimo cultures found in Chukotka were first formed in Southwest Alaska and then brought to Chukotka via the Bering Strait. The opposite view was held

by Bogoras (1934), who thought that Eskimo were relatively recent arrivals to the Chukotka Peninsula, who have wedged themselves in between Paleoasiatic and American Native groups (the "Eskimo wedge" theory). This debate is still not resolved.

In addition to the ancient history of Northeast Asians, events of the 20th century added even more admixture to their gene pool. Firstly, Black and "Caucasian" traders and whalers from America in the beginning of the 20th century were frequent visitors to Chukchi and Eskimo communities and left their mark in the native gene pool. Then from the 1950s onwards, many of the small native communities amalgamated and their inhabitants relocated during a process called *ukrupnenie* (consolidation), putting together Chukchi and Eskimo, or Reindeer Chukchi and Coastal Chukchi, Chukchi and Koryak, etc. It exposed people to a much wider range of potential partners and added more recent interethnic admixture to their gene pools.

The Soviet period also introduced new kinds of mobility. The development of aviation and other modes of transportation made connections easier between European and Asian parts of the USSR, different cities and villages in the Far East easier, thus facilitating migration. Since the 1950s, the exploration of minerals and expansion of mining industries in the Chukotka and Kamchatka Peninsulas brought people of tens of nationalities from the Western part of the Soviet Union [1], many of whom were young single males. This resulted in marriages of native people and with immigrants, both Caucasian and Mongoloids (e.g., Buryat or Sakha). It also led to the birth of out-of-wedlock children who did not necessarily look Caucasoid and whose paternity was sometimes concealed. However, in recording their nationality as native in their passports,

[1] According to Shimkin (1990), in Chukotka between 1959 and 1970 alone, the number of immigrants rose from 35,000 to 80,000, with the native number rising only from 12,000 to 13,000.

their mixed parentage was not reflected in their selection of a single native ethnic identity, thus adding more confusion to the problem of assigned ethnicity. Many of them considered themselves to be "pure" native despite the admixture. Nevertheless, many geneticists choose their samples using these assigned ethnicities. By now it should be clear that the problem of ethnic identity is a complicated one in relation to Northeast Asian individuals, and geneticists should be realistic about their attempt to find ethnically "pure" representatives of any studied ethnic group. I shall pick up this point in Chapter 5.

In conclusion, this overview of the existing hypotheses and theories concerning the dates, origins, and population movements of Northeast Asian ethnic groups shows a diverse history. The first humans came to Northeast Asia in the Paleolithic, but scholars hypothesize that ethnic groups as we know them now started to form in late Mesolithic and Neolithic times. Neolithic continental hunters and fishermen from the regions of Lake Baikal, the Amur River, Yakutia and the Far East mixed with the earlier Mesolithic inhabitants of Northeastern Asia. Historic events, such as wars, epidemics of smallpox and measles that affected many villages, marriages between individuals from the tribes of different ethnic groups and with Russians, expansions of some ethnic groups into the territories of others and subsequent partial assimilation, and considerable recent admixture, all resulted in the intertribal and Caucasian admixture of different degrees. Such events also indicate a complicated demographic history of the populations and imply that the assigned ethnicity of individuals cannot be taken at face value.

Chapter II A review of genetic systems studied in Northeast Siberian populations

A number of genetic systems have been examined in the study of population structure[1] and population history[2] of indigenous Siberian ethnic groups. Early research focused upon serological studies of blood group and protein polymorphisms.
The development of molecular genetic methods for studying nuclear DNA, mitochondrial DNA and Y-chromosome polymorphisms allowed construction of gene trees and reconstruction of population phylogeny.

Studies of polymorphic genetic systems have value but also limitations. Genetic polymorphisms are useful for investigating the relationship among subpopulations. When alleles are shared between subpopulations because of gene flow, migration patterns among subpopulations can be inferred. Within subpopulations, alleles may also be shared due to common ancestry. Therefore, the study of DNA markers, sequences of alleles or patterns of polymorphism can reveal evolutionary history and role of microevolutionary forces (Hartl & Clark, 1996). Shared environments, however, can also result in sequence similarity if, for example, a mutation has a selective value in a particular environment. The determinants of gene frequency change are mutation, natural selection, and population structure, which includes such parameters as gene drift, gene flow, and inbreeding, that depend on the size, dispersal, and mating patterns of the population, respectively (Livingston, 1980). Thus, genetic studies of indigenous Siberians involve the

[1] Population structure- the study of the effects of internal migration, group composition, mating practices, and other factors on the amount and pattern of the genetic drift within an area.
[2] Population history- the degree of similarity among populations, where similarity may reflect either common ancestry or mate exchanges. These two aspects of genetic similarity are ordinarily inseparable (Harpending and Jenkins, 1973).

analysis of the relative role of microevolutionary factors on observed differences in allele frequencies between populations. This chapter gives an outline of genetic systems and techniques to prepare the ground for a detailed survey of how they have been applied to selected populations.

II. 1 *Polymorphisms detected by serological methods*

Blood groups and protein polymorphisms

Since the discovery of ABO blood groups by immunological techniques (Hirszfeld and Hirszfeld, 1919), an array of hereditary markers have been identified on erythrocytes, leukocytes, plasma proteins, and erythrocyte and leukocyte enzyme. The commonly studied polymorphic genetic markers in the Northeastern Siberian populations are summarized in Table I. The presence, distribution and combination of marker genes and allele frequencies in populations, as well as patterns of within-group and between-group allele frequency variation can be used to group populations according to their similarities and differences. However, blood group and protein polymorphisms are considered to be "adaptive"[3] traits (Giblett, 1969). Thus, they may be influenced by environmental conditions. In the case of Siberian ethnic groups, observed similarities between populations may be a result of shared environment, rather than of common ancestry.

[3] Adaptive traits are traits subject to the action of natural selection (Hooten, 1931).

Human leukocyte antigen (HLA) system

The HLA system is located on the short arm of chromosome 6 and encodes two distinct classes of highly polymorphic cell surface molecules that play a central role in cell-mediated immune response to infections. HLA class I molecules -A, -B, and -C are expressed on almost every cell type in the body. HLA class II molecules, -D, are found on cells of the immune system, such as B-lymphocytes and activated T-lymphocytes (Browning & McMichael, 1996). HLA molecules are among the most polymorphic known. However, alleles are not equally distributed among individuals in a given population, or ethnic group. Analyses of allele frequencies and haplotype[4] frequencies and distribution allows to group populations according to their similarities, hence the use the HLA system in anthropological genetics (Schanfield, 1980). The HLA system was discovered serologically through observations of agglutination of leukocytes with antisera taken from unrelated transfused individuals. Recently, molecular genetic techniques for identifying HLA alleles have been developed, allowing for detection of DNA sequence polymorphisms. DNA-based methods include restriction fragment length polymorphism (RFLP) analysis and polymerase chain reaction (PCR).

Molecular genetic methods

Restriction Fragment Length Polymorphism (RFLP)

RFLPs result from the presence or absence of a particular restriction site in DNA. Restriction site is a particular nucleotide sequence, at which a restriction enzyme cleaves DNA. A mutation at this site will prevent cutting. DNA fragments resulting from

[4]Haplotype is a series of alleles found at linked loci on a single chromosome.
Allele is one of the several alternative forms of a gene or DNA sequence at a specific chromosomal location, locus (plural-loci).

restriction of the DNA are subjected to gel electrophoresis and thus separated according to size. It is then transferred to a membrane and hybridized with radioactively labelled probes, or human DNA segments which contain the nucleotide sequence of interest (Fig. 1a). (Hartl and Clark,1996).

Polymerase Chain Reaction (PCR)

PCR allows specific amplification of stretches of DNA sequence, through repeated cycles of DNA denaturation, annealing of specific primer to the DNA single strand and nucleotide extension from primer pairs using a thermally stable enzyme, to generate substantial quantities of specifically amplified regions of DNA which can then be used in further analysis (Fig. 1b) (Bell, 1989).

II. 2 ***Polymorphisms detected by DNA-based methods***

Mitochondrial DNA variations

The analysis of variation in the small circular genome of the mitochondria (mtDNA) has been used extensively in human evolutionary research. Characteristics of mtDNA such as maternal inheritance, apparent lack of recombination (Giles *et al*, 1980) and high mutation rate (Miyata *et al*, 1982) make mtDNA anthropologically useful. Direct nucleotide sequencing of mtDNA is used to obtain information about variability in DNA sequences in the non-coding region (Control Region, or CR), expressed in sequence length mutations (insertions and deletions) (Cann and Wilson, 1983). One of the most studied regions is a non-coding segment V. In most cases region V contains two copies of tandem repeat CCCCCTCTA. However, in some human populations this region contains

polymorphisms expressed as deletion of one copy (9bp deletion). In others an insertion polymorphism is found, in the form of insertion of a third copy of the (CCCCCTCTA) tandem, or insertion of four cytosines -CCCC- (Hertzberg *et al*, 1989, Wrischnik *et al*, 1987). Another region of the CR, used extensively in evolutionary studies, is the hypervariable segment I located between 16000 and 16400 base pairs (bp). Polymorphisms in this segment are mostly due to the point mutations.

Although mtDNA polymorphisms are useful in evolutionary studies, a number of problems have been identified. Howell *et al* (1996) and recently Saville *et al* (1998), Hagelberg *et al*, (1999), and Eyre-Walker *et al* (1999) reported a case of mtDNA recombination, implying the possibility of paternal inheritance of mtDNA. The high mutation rate (~ one mutation per 25 generations) detected in human mtDNA D-loop, along with its usefulness for identifying a large number of maternal lineages in human populations, has been a source for ambiguity in phylogenetic analyses (Hammer and Zegura, 1996). Thus, an independent single nuclear locus should be used to test mtDNA hypotheses for the origins of studied ethnic groups. The non-recombining region of Y chromosome (NRPY), a paternally inherited nuclear component, has served as a valuable independent single locus.

Y chromosome polymorphisms

The Y chromosome is also uniparentally inherited. Thus, karyotypically normal males possess a single lineage from father, grandfather, and paternal great-grandfather. Every Y chromosome contains a record of the mutational events that occurred on all previous ancestral Y chromosomes. A number of polymorphic markers have been found

on NRPY. Minisatellites and microsatellites, the two classes of variable number of tandem repeats (VNTRs) with short repeat units, are the most informative. They can vary in the number and in the sequence of individual repeat units. Microsatellites mutate via a stepwise mutation mechanism, replication slippage, which forms small changes in array length (Weber and Wong, 1993). The most extensively studied Y-linked microsatellite has been the DYS19 locus, on the short arm of Y chromosome, with seven alleles. Polymorphism at this locus is the result of differences in the number of GATA tandem repeats. The frequencies of the alleles vary in human populations, and some geographical trends are also evident. Also, single-base substitutions and small insertion/deletion (indel) are known as polymorphisms on the Y chromosome. One of the most useful is the Y Alu polymorphism (YAP). This indel, discovered as an RFLP, results from either the presence (YAP+), or absence (YAP-) of an ~300bp Alu element[5] at a specific site on the long arm of Y chromosome (Hammer, 1994). Asia exhibits an irregular distribution of YAP+ (Hammer and Zegura, 1996).

This brief overview helps to illustrate the reasons for the study of polymorphisms in genetics studies. It also reminds us that inferences about phylogenetic relationships are best made on the basis of a few independent loci, since a single locus provides such limited information. However, it should be recognized that the results of genetic studies should also be interpreted in the light of a historic and demographic context.

[5] The *Alu* family of short interspersed repetitive elements is found in all primate species, with about 500,000 copies per genome occurring in humans. *Alu* elements at specific loci that are not fixed in human populations are likely to have resulted from unique and recent insertion events. Due to their stability, this type of marker promises to be quite valuable for human population studies (Hammer and Zegura, 1996).

Chapter III. Genetic studies of the Northeast populations.

III.1 **Asian Eskimo and Chukchi**

III.1.1 Serological studies

This section of the thesis reviews the results of the studies that focused on the revealing population genetic structure and population history of Asian Eskimo, and the Coastal and Reindeer Chukchi populations using genetic systems determined by serological methods.

Early studies in the 1960s and 1970s were based on blood group and protein polymorphisms with occasional consideration of demographic and genealogical data (Rychkov *et al*, 1972, Gurvich, 1985b). Unfortunately, a comparative analysis of genetic data generated by different researchers is difficult, due to the apparent incoherence in studies. Indeed, studies show little overlapping in the choice of genetic systems; sample sizes were different ranging from 46 to 402 individuals; the number of villages varied; methods of reporting the data different. For example, Asian Eskimo were studied by four groups of researchers. (1) Rychkov *et al* studied some blood groups and some protein polymorphisms in three populations and reported gene frequencies in his tables. (2) Sukernik *et al* conducted two studies, one of four Eskimo populations and second of just one Eskimo and 10 Reindeer Chukchi populations, focusing mostly on blood group and protein polymorphisms, with little overlap between his and Rychkov's genetic systems. Instead of gene frequencies, allele and haplotype frequencies were calculated. (3) Solovenchuk *et al* focused mostly on protein polymorphisms in three pooled Eskimo populations. Interpopulation variability was not possible to identify and compare with the

previous studies because populations were pooled into one sample. (4) Nazarova pooled two populations and studied protein polymorphisms. However, she reported gene frequencies, as opposed to phenotype and allelic frequencies published by Solovenchuk. Thus, the overall picture is patchy. However, when the results are compared, there is some agreement. For example, the high frequency of O allele of the ABO blood group in Eskimo populations was common in all studies. High frequency of AcP and PGM1 loci in Asian Eskimo and Chukchi populations was also observed. Discrepancies between populations were sometimes explained by unique historical events in these populations. In some cases, the reason for discrepancy is difficult to identify, due to the differences in samples[6]. In general, however, there was no major disagreement between genetic and historic data.

Some authors focused on studying population structure of circumpolar Siberian groups trying to establish that these populations, due to living in the harsh arctic environment, experience a significant natural selection pressure, which could alter the effect of random genetic drift (Rychkov, Sheremet'eva, 1984).

Rychkov and Sheremet'eva (1972) studied 12 loci of blood groups and protein polymorphisms in 220 Asian Eskimo and Chukchi and 18 Asian Eskimo-Chukchi

[6] The frequencies of Fy^a allele at Duffy locus were significantly different in Rychkov (1972), Sukernik (1981), and Sukernik (1986) studies, 0,193, 0,913 and 0,916, respectively. Although Rychkov made a note of such unusually low allele frequency, he does not try to explain it. Sukernik attributes the differences in allele frequencies at Duffy and other loci (K, Rh, Di) between his and Rychkov's results by exclusion from their sample of individuals with non-native admixture. Unfortunately, identification of the reason for the discrepancy is not possible, since Rychkov's sampling procedure is not described. However, indirect evidence from other studies (Crawford, 1981) that reports disproportionately high frequency of the Fy^a allele (70-100%) in Alaskan populations implies that Sukernik's results are more accurate.

hybrids (Table 2 and 3). According to the genetic distance estimates (Table 4) Asian Eskimo and Chukchi populations are "intermediate" between Western Hemisphere Eskimo (Alaskan and Greenland) and "Siberian" populations, but "closer" to "Siberian" populations. Unfortunately, it is not clear which "Siberian" ethnic groups were used for the genetic distance calculation. The Chukchi are genetically more distant from the Western Hemisphere Eskimo. Asian Eskimo had genetic distances that were equal with Alaskan Eskimo and Siberian populations. Solovenchuk (1984) concurs with this conclusion. The genetic structure of the Chukchi was explained by the migration and marriage patterns with ethnic groups from continental Northern Asia. However, genetic structure of the Chukchi corresponds to their geographic location among the Asian Eskimo only partially. This supports the assumption that the Chukchi migrated to this region relatively recently. Unfortunately, the manuscript is vaguely worded and it is difficult to determine the exact Siberian populations and populations of continental Northeastern Asia: are they Reindeer Chukchi, Even, or Evenk? On what grounds were Alaskan and Greenland Eskimo referred to as Western Hemisphere Eskimo?

Sukernik *et al* (1981) studied phenotype and allelic frequencies for 13 blood groups and protein polymorphisms (Table 5) in approximately 1,000 Reindeer Chukchi from 10 adjacent populations and approximately 100 Asian Eskimo (Fig. 2). Significant variation, or heterogeneity of allelic frequencies, was reported (1) among Chukchi populations, the MNS, P, Rh, Duffy, and PGM genetic systems contributing to this heterogeneity; and (2) between Reindeer Chukchi and Siberian Eskimo, where the greatest divergence was observed for the MNSs, P, Rh, and AcP loci. Since the MNSs, P, and Rh genetic systems contributed primarily to the heterogeneity observed both among

Chukchi populations and between Chukchi and Eskimos, it was assumed that these systems, more than others, were affected by genetic drift. This finding was supported by Vibe *et al* (1990), who reported high linkage disequilibrium for pairs of loci MN and Ss (MNSs system), Cc and Ee (Rh system)[7].

Two related Chukchi populations, Achaivayam and Middle Pakhachi, differ from the other 8 Reindeer Chukchi populations in this study by having the high frequencies of the B allele of the ABO system[8]. The authors explained this difference by using historical data which indicates that approximately 100 years ago the Reindeer Chukchi diffusing south from the lower Anadyr River crossed the Koryak Range, invaded and assimilated local tribes of Reindeer Koryaks who, apparently, had a higher frequency of the B allele. No reference to historical or ethnographic sources was given to substantiate this statement. Additionally, there was no account of genealogical study. When the frequencies of the B allele in contemporary Koryak were reviewed, they were only slightly higher than in Reindeer Chukchi (weighted mean 0,179, Solovenchuk *et al*, 1985). Since it is impossible that the allelic frequencies were identified a 100 years ago, without proof from the historic records this statement becomes unjustified.

Asian Eskimos differ significantly from Chukchi in frequency of the p^a allele at the AcP locus[9]. Different p^a frequencies between both groups might result from diverse selective forces acting in different environments. Stabilizing selection was assumed to be the principle agent maintaining homogenous allelic frequencies (p^a and p^b) at the AcP

[7] For MN and Ss loci, D (linkage disequilibrium) for Chukchi = -0.0284+0.0034; Eskimo D= -0.0515+0.0091 ($t_d = 2.38$; $p < 0.05$). Linkage disequilibrium for Rh system was even greater: Chukchi D= 0.1545+0.0058 and for Eskimo D= 0.2319+0.0138 ($t_d = 5.17$, $p < 0.001$). Greater D for Eskimos might be explained by the lower effective size of Eskimo population as compared to Chukchi.
[8] This comprised 0,201 and 0.225, respectively, compared to a weighted mean 0.149.
[9] The frequency of the allele was 0.696, compared to the weighted mean of 0.570 in Chukchi, with local frequencies ranging from 0.534 to 0.614.

locus among Reindeer Chukchi subdivisions. The authors imply the selective advantage of the p^a allele in the environment of the Asian Eskimo. However, they do not explain the difference between the environments of Asian Eskimo and Reindeer Chukchi, and what "diverse selective forces" may act in these different environments. Do they refer to the differences in climates, diet, or diseases? For the AcP locus Solovenchuk (1982) assumes different selective advantage of some phenotypes at different stages of ontogenesis, which results in the heterogeneity of generations in a population. He found that AA phenotype frequencies of the AcP genetic system decrease in the age group 20 to 49 years in relation to the age group 5 to 19 years, and then significantly increases in the third group, 50 and older.

Observed and expected phenotypes are in agreement with the Hardy-Weinberg equilibrium for each locus, with the exception of $PGM_{1.}$ This locus showed a deficit of heterozygotes and an excess of homozygotes. The authors suggest that these deviations may result from failure to detect heterozygotes due to the presence of unknown silent alleles. Alternatively, negative selection against heterozygotes or assortative matings could also explain homozygote excess. Vibe *et al* (1990) supported the finding by determining linkage disequilibrium for three pairs of unlinked loci: PGM1 and Hp (for Chukchi and Eskimo), PGM1 and AcP (Chukchi and Eskimo), and AcP and Hp (Chukchi). The values of linkage disequilibrium for the PGM1 and AcP pair of loci did not differ statistically for Chukchi and Eskimo populations. This points to the possibility of the existence of single directional factor for all populations that provides non-random allele association. The PGM_1^1 allele was found to be selectively advantageous in the circumpolar zone. Spitsin (1985) found a positive correlation between frequency of this

allele and the mean January temperature. For the AcP locus, the distribution pattern of this allele may result from its selective advantage in low temperatures, since the frequency of this allele increases from the equator toward the poles and also correlates with the mean annual temperature. Selective advantage of these alleles in circumpolar populations suggests that selection should directly favor their combination. However, the hypothesis that there is a selective advantage of such combination of PGM1 and AcP loci needs further study.

Solovenchuk *et al* (1982a) conducted another study of gene and phenotype frequencies for 11 polymorphic loci of serum and red blood cell proteins in 1245 individuals of Coastal Chukchi from three subpopulations (Fig.3). Variability of gene and phenotype frequencies between subpopulations was insignificant (Table 6). The distribution of phenotypes was the same for all polymorphic systems in all subpopulations studied. A deviation from Hardy-Weinberg equilibrium was found for Gc, where the deficit of heterozygotes was observed. This finding, however, remains unsupported by other studies and should be treated as an isolated case. Also, a deviation was found for GPT, when more than a 50% limit of heterozygosity was recorded in all three populations. For the GPT locus, heterozygosity increased with increasing age of individuals (Solovenchuk, 1982b). Solovenchuk explains this deviation from the Hardy - Weinberg equilibrium as a consequence of natural selection pressure, leading to the differential fecundity and mortality in relation to genotypes of selected polymorphic systems. However, Solovenchuk did not provide references to demographic or genealogical studies that would substantiate his interpretation of deviation from the equilibrium.

The level of heterozygosity in populations of Asian Eskimo, Coastal and Reindeer Chukchi was studied by Solovenchuk (1984, 1989) and Nazarova (1989). They arrived at different conclusions. Solovenchuk, using a sample from 3 subpopulations of Asiatic Eskimos (n=402), 5 subpopulations of Coastal Chukchi (n=1793), and 3 subpopulations of Reindeer Chukchi (n=559) found the level of heterozygosity (H) for all 33 loci for the Asian Eskimo, Coastal and Reindeer Chukchi not to differ substantially from the more southern groups of Mongoloid populations [10]. The level of heterozygosity in Eskimo and Reindeer Chukchi is approximately equal, but both groups are different from Coastal Chukchi. In his later work, however, Solovenchuk found the greatest degree of heterozygosity in blood protein genes in the northernmost groups, with decreasing heterozygosity in the more southerly groups (Solovenchuk, 1989)[11]. Nazarova had a different conclusion. She used a considerably smaller sample of 37 Coastal Chukchi, and 46 Asian Eskimo. She reported that the average heterozygosity of Asian Eskimo and Chukchi populations, calculated from 6, then 10, and then 8 loci is the smallest in Asian Eskimos, but considerably greater in Coastal Chukchi from Novoye Chaplino, and greatest in Lorino Chukchi. Average heterozygosity in Eskimo increases in the following order: Asian Eskimo, St.Lawrence Island, Alaskan, Greenland, and Canadian Eskimos. Thus, the results of the studies of the level of heterozygosity are contradictory and inconsistent. If the discrepancy between the Nazarova and Solovenchuk studies could be

[10] 0,118+ 0,005, 0,130+ 0,002, 0,120+0,004 and 0,129+ 0,003, respectively

[11] Thus, of 35 loci examined in Asiatic Eskimo, Chukchi, and Koryak individuals, 12 were found to be polymorphic: AcP*1, PGM*1, PGD, GPT, GLO*1, EsD, Hp, Pp, E*2Gc, ABO, and PTC. The average heterozygosity (Nei, 1977) of these groups, from northernmost to southmost, is as follows: Eskimo (n=402), 0.333+0.003; Coastal Chukchi (n=1,776), 0.329+0.004; Reindeer Chukchi (n=559), 0.306+0.007; and Kamchatkan Koryak (n=675), 0.259+0.006. These differences in heterozygosity value are significant ($P < 0.01$), except for the comparison of the Eskimo and Coastal Chukchi data ($P < 0.1$).

explained by the different sample sizes used in their research, the reason for the difference in the results of two studies conducted by Solovenchuk is unclear.

Nazarova also calculated genetic distances between different Eskimo populations. Genetic distance between Asian Eskimo and St.Lawrence Eskimo populations appears to be greater than the genetic distances between Asian Eskimo and Alaskan Eskimos (Table 7). This conclusion contradicts previous results by Sukernik *et al* (1986), Ferrel *et al* (1981), and Crawford (1981). All found closer genetic affinities between Asian Eskimo and St. Lawrence Eskimo. It is difficult to assess whether or not a small sample size and unexplained sampling procedure played an important role in the assessment of the genetic distances between populations in Nazarova's study.

Genetic distances calculated by Solovenchuk, more accurately reflect the ethnohistory of these populations (historic contacts and migration pattern) (Table 8). Reindeer and Coastal Chukchi are genetically closer to each other, while the Reindeer Chukchi and Alaskan Eskimo are genetically most distant. Asian Eskimo are genetically intermediate between Coastal Chukchi and Alaskan Eskimo. However, Solovenchuk explains, that the genetic structure of Reindeer Chukchi could be accounted for by migration and admixture with Asian Eskimo. Such explanation does not explain Alaskan Eskimo results. Interpopulation heterogeneity in three Chukchi and Alaskan Eskimo populations did not differ significantly. Consequently, he argues, the isolation of the Asian Eskimo and Alaskan Eskimo did not result in the significant differentiation of their genetic structures. The author concludes that the relative homogeneity of all studied populations located on both shores of the Bering Strait results from the selective forces operating on these populations. In other words, the observed genetic structure is caused

by "ecological factors". Unfortunately, the author often attributes differences or similarities between populations to "ecological conditions" that are not defined or described. In this study, he attributes the similarities between Chukchi and Alaskan Eskimo to some "ecological conditions". Yet excludes the fact that contact between Chukchi and Eskimo populations from both sides of the Bering Strait existed for centuries. This fact would easily explain the small differentiation between of Alaskan Eskimo, Asian Eskimo and Chukchi populations.

Sukernik *et al* (1986a) studied G1m (z,a,x,f)- and G3m (g, b0, b1, b3, b5, s t)-immunoglobulin allotypes in the sample of 1079 Chukchi from 10 populations from interior Chukotka and adjacent Kamchatka (Fig. 2). The observed heterogeneity of Reindeer Chukchi was less then expected from the scattered populations. However, the high mobility of these people enabled them to travel considerable distances between communities and gene flow may have occurred between two populations. Genetic structure of Reindeer Chukchi is demonstrated in Fig. 4. Minimal genetic distance is found between two neighboring populations of Chukchi from Achaivayam and Srednie Pakhachi communities in Kamchatka as a result of intensive gene flow in both directions. The isolation of the Chukchi from Kamchatka on the genetic map is probably due to geographic isolation from the more northern populations of Chukchi and the presence of the Koryak Mountain Range. Alleles of the Gm, ABO, Rh, MNSs, and PGM1 loci contributed the most to the process of the intrapopulation differentiation. The results of the regression analysis show that Kanchalan, Meinypil’gino, and Rytkuchi, comprised of the heterogeneous groups of Chukchi, are positioned above the regression line (Fig.5). This is possibly due to the influence of migration on the distribution of allele frequencies

in these populations. The position of Rytkuchi above the regression line on the genetic map indicates possible assimilation into the Chukchi population of local Yukagir and Even. Kanchalan had the highest Caucasoid admixture (4.3%). This finding explains Kanchalan's position on the map above the regression line indicating gene flow from Caucasoid. Level of heterozygosity in Amguema is considerably lower than expected and is explained by high endogamy of the Amguema Chukchi and reproductive and geographic isolation. Haplotype Gm (f;b035st) was detected in 9 out of 10 populations, which indicated Caucasoid admixture (1.3%).

Sukernik *et al* (1986b) continued studies of polymorphic blood systems by concentrating on Asian Eskimo. There were 332 (3/4 of total Asian Eskimo population) “pure” Asiatic Eskimos from Novoe Chaplino, Sireniki, Uelen, Uel’kal’, Lavrentia, Lorino participating in this study. Samples were taken from teenagers, adults and elderly whose parents were Eskimo. The authors do not provide an explanation of how the "purity" of the sample has been ensured. As we will see later, the sampling problem is an important issue, especially when it comes to estimating evolutionary relationships between ethnic groups.

Minimal genetic distance was found for Sireniki and Novoe Chaplino (Fig. 6). These populations frequently intermarried. The genetic map also showed a relatively close affinity between populations of Sireniki, Novoe Chaplino and St. Lawrence Island (Gambell and Savoonga). These groups are thought to be the descendants of one tribe, and speak the same Yupik dialect of the language of the Siberian Eskimos. They had marriage contacts until the 1948. Alleles that contribute the most into differentiation of 8 Beringian populations are A1, B, Ms, Ns, CDe, and PGM_1^2. The extreme values of allele

frequencies for some loci ($A_{1,}$ B, $PGM_1^{2,}$ and P^b) in Naukan determined the genetic structure of Naukan that is intermediate between Yupik-speaking and Inupiaq-speaking populations of Wales and King Island.

The results of regression analysis show that populations from Naukan, Gambell, Sireniki, and Novoe Chaplino are farthest from the theoretical regression line (Fig 7). Naukan and Gambell are located above the line, indicating the influence of migration on the distribution of allele frequencies. This conclusion is in good concordance with the historic data and reflects gene flow into the Naukan community two generations ago. The high level of heterogeneity in this community is explained by gene flow from Alaska through Imaklik Island (O. Ratmanova) to Naukan, and marriages with neighboring Chukchi. The low level of heterozigosity of Sireniki, located below the theoretical regression line, is explained by bottleneck effect resulting from severe epidemics of the 19th and 20th centuries. The same is true for Novoe Chaplino, which also has low level of heterozygosity. This population, numbering 1400 individuals at the end of 17th century, was reduced to only 300 at the end of 19th century, and were growing slowly in the 20th century. This study is one of the rare examples when interpretation of genetic results was supported by the use of ethnographic and historic data available not just for "Chukchi" or "Eskimo" populations in general, but for the inhabitants of specific villages. The Asian Eskimo were found to be more heterogeneous (Rst=0,031), than the Chukchi (Rst=0,012). Loci responsible for the differentiation of Chukchi and Asian Eskimo are P, MNSs, Rh, PGM1, and Gm. The relative genetic distance between Chukchi and Eskimo was 4 times greater than average genetic distance between Chukchi populations but only 1,6 times greater than the average genetic distance between Asian Eskimo. The authors

conclude that the high level of regional heterogeneity in Eskimo populations could have played an important role in survival and subsequent expansion of some Eskimo tribes during the unfavorable ecological and historic events in the last quarter of 19th and beginning of 20th century.

Crawford *et al* (1981) studied the genetic structure of four Alaskan Eskimo communities. Data on Siberian communities were taken from published literature. Population structure of 19 populations is given in Fig 8. The conclusions from the regression analysis do not contradict the ethnohistory of the Eskimo and Siberian peoples. Nunyamo and Mednii Island have high heterozygosity. This pattern suggests migration, or possibly directional selection in these groups. Savoonga is closer genetically to Chaplino than Gambell, because of the marriages. However, the author mentions that recent European admixture, and various resettlement programs, has added a certain amount of "noise" to the interpretation of the reduced space genetic maps. Gene flow between Chukchi, Eskimo, and Russian settlers in Chukotka communities, such as Nunyamo and Sireniki, distorts the precontact relationships of the native groups and vastly inflates the average per locus genetic heterozygosities observed. Nevertheless, population genetic structure of the large circumpolar samples clearly reflects geographical separation of the groups analyzed.

This review of serological studies showed that despite the incoherence in genetic research of the Northeast Asian populations, some conclusions could nevertheless be made. (1). Genetic structure of studied populations is considerably influenced by migration and genetic drift due to the small effective sizes of populations[12]. (2) Action of

[12] Effective size largely determines genetic differentiation, because it relates directly to the reproduction of population in generations, that is, if such genetic differentiation is not counteracted by gene migration.

natural selection is not always evident, although the allele frequencies and distribution of some loci, such as AcP and PGM1 might indicate their selective advantage in high latitudes. "Ecological conditions" remain a very elusive term. No one clearly stated a set of characteristics that describe these conditions. Sukernik (1981) considers the conditions of 10 populations of Reindeer Chukchi to be nearly identical. Solovenchuk (1984) considers ecological conditions of 6 populations of Coastal Chukchi to be different. Neither researcher explained the differences or similarities in these conditions. This matter needs clarification and classification, where information about climatic conditions of each group would be described. Similarly, precise information about lifestyles, diet, disease status and occupation should be provided. (3) Sampling and admixture between populations also continues to be a problem, despite one genealogical and a few demographic studies. (4) Genetic affinities of Reindeer and Coastal Chukchi, Asian Eskimo and Alaskan Eskimo reflect ethnohistory known from historic and ethnographic data.

The past decade revived the interest in the genetic studies of Northeast Asian groups with the availability of more informative molecular genetic methods. Using these methods, researchers were able to obtain information that helped to clarify some of the uncertainties and provide support for some theories regarding the dates and places of the origins of these native groups.

III. 1. 2 Molecular genetic studies

In the beginning of 1990s, attention seemed to shift from describing genetic structure of Siberian populations to attempting to assesess relationships between Siberian

and Native American populations. Although some studies focused on the problem of populating the New World, the scope of Siberian studies became broader. Unfortunately, less and less attention is paid to demography, genealogy, history and proper sampling procedure in these studies. Some data (e.g., estimation of Caucasian admixture) were taken from outdated sources and travel from one article to another, without an attempt to verify the accuracy of this data for a particular population.

Shields *et al* (1993) studied mtDNA sequence in Beringian and Northern American populations by analyzing sequence and nucleotide diversity in 33 mt lineages from 90 individuals belonging to five Circumpolar populations of Beringia, North America, and Greenland. Two major problems in the methodology of this study undermine the results of this otherwise interesting research. Firstly, they used a very small sample size consisting of 7 Chukchi from villages Ust-Belaya (4), Kanchalan (2), Uelen (1) of the former Chukchi Autonomous Territory, Chukotka, and 6 Siberian Eskimos from villages Novoye Chaplino (5), and Lorino (1). Secondly, the sampling procedure was poorly designed. Researchers interviewed donors about their knowledge of their maternal ancestry. All subjects stated that their grandmothers and great-grandmothers belong to their respective native groups. No verification was attempted. The authors accepted the estimates of Caucasian admixture for Beringian populations of approximately 2-5% (Alekseev 1979, Ferrel, 1981).

The analysis of within-population sequence differences for Circumarctic (Chukchi, Eskimos, Athapaskan and Haida) populations showed that mean values of sequence difference for all groups were low, suggesting a recent origin for these populations (Table 9). The estimated mutation rate for mtDNA is approximately 1%

sequence divergence/ 8,950 years (Lundstrom *et al*, 1992). At this rate, approximately 5,100-7,100 years would be required to generate the sequence diversity observed within these Circumarctic populations. Although this time period refers to the ancestry of the molecular lineage, rather than the absolute age of the populations, it suggests an upper limit for the origin of these populations. Also, under the assumption of comparable population sizes and similar levels of population subdivision and migration, the similarity in average molecular divergence found within these populations suggests that their evolutionary ages are also similar. However, this assumption is not necessarily correct because population sizes of Asian Eskimo and Chukchi are not equivalent, as the Chukchi numbered 15,100 in 1989; the Eskimo 1,700. Moreover, the similarity in the level of migration and population subdivision should be substantiated detailed studies rather than simply assumed.

The average between –group pairwise mtDNA sequence difference values can be used as an estimate of the upper limit of population divergence (Nei, 1987). The four Circumarctic populations have the smallest values, suggesting that they are the most closely related populations. The between -group sequence divergence values are very similar for these four populations, they are actually equivalent to their within-group difference. By contrast, three Amerind groups have between-group mtDNA sequence difference values that are twice as large as the Circumarctic populations, suggesting that Amerinds diverged before the separation of the Circumarctic groups.

Phylogenetic analysis (Fig. 9 and 10) has shown that the lack of well-defined lineage clusters, plus the intermingling of lineages among different populations, does not support long-term separation among these Circumarctic groups. Figure 10 presents an

inferred population tree derived from average sequence divergence values for all sequences of Circumarctic populations in Shields' study. The relatively short branch lengths leading to each population is also consistent with the hypothesis of a recent origin. Longer branch lengths between Amerinds suggest a greater age for Amerind populations and perhaps evolution in relative isolation.

Furthermore, Shield's analysis of the distribution of the 9bp length mutation in mtDNA of the Circumarctic populations found that all groups located in northern regions, above 55^0 N, posses two copies of the tandem mutation (Fig. 11). Populations located south of 55^0 N possess one copy of the tandem mutation, irrespective of whether they were in Asia or America.

Shields *et al* state that the most plausible explanation for these trends is that the evolutionary radiation of the Circumarctic groups, in concert with the evolution of their mtDNA lineages, occurred within a shallow time depth. It also appears that despite their geographic separation, Circumarctic populations form a cohesive biological unit that includes Na-Dene-speaking Haida and Athapaskan, West Greenland Eskimo, Inupiaqs, and Chukchi. Mitochondrial data also suggests that all Circumarctic populations had genetic origins about the same time and cultural differences, that subsequently occurred, were accompanied by relatively little biological differentiation. This hypothesis suggests similar dates of origins for contemporary Chukchi and Eskimo native groups, commonly accepted as 8-10 thousand years ago. However, the aforementioned problems with sampling and small sample size could have influenced the results and obscured the interpretations. In addition, unsubstantiated assumptions regarding population sizes and

levels of migration make the conclusion of equal evolutionary age of these populations unjustified.

A larger sample of 36 Asian Eskimo, 75 Chukchi and 14 Yukagir in which no one indicated knowledge of admixture in the interview, was conducted by Shields *et al* (1994). In this study, the time depth of radiation of Amerind-Eskaleut was calculated as 12,100-13,200 years. The authors conclude that it is likely that lineages found in Amerind speakers are descendants from a set of lineages that occurred in a genetically diverse set of early migrants. Fig. 12 represents a proposed phylogeny for the 11 groups studied. It suggests a recent arrival for Eskaleut and Na-Dene in the New World (node F). The similarity of mitochondrial sequences found among the widely scattered Circumarctic populations, the small number of substitutions separating their DNA sequences, their intermingling within the molecular phylogeny and the lack of major clades within phylogenies all suggest that these Circumarctic populations arose by a recent, and rapid, evolutionary radiation (approximately 5,100-7,100 years ago as shown in the previous study). This hypothesis supports Sergeev and Gurvich hypotheses (see Chapter I) of the time depth of Escaleut cultures.

Findings of Torroni *et al* (1993) indicate that little, if any, of the mtDNA diversity that currently exists in Native Americans arose in Siberia prior to Amerind migration. Their study of the mitochondrial DNA variation in 411 individuals from 10 aboriginal Siberian populations from northern Siberia and the Russian Far East: 20 Sel'kups, 49 Nganasans, 43 Evens, 51 Evenks, 46 Udeges, 24 Chukchi, 46 Koryaks, 27 Yukagirs, 57 Nivkhs, and 50 Asiatic Eskimo, showed that these populations exhibit mtDNA haplogroups from three (A, C, and D) of the four haplogroups observed in Native

Americans. Unrelated adults of both sexes were included in the sample. Specific information regarding admixture was given only for two groups: Udege (unhybridized, unrelated individuals, most of whom were older-age people) and Nivkhs (unhybridized, unrelated). In their estimation of admixture for Asian Eskimo, the authors referred to Sukernik *et al* (1986) who stated, that elder generations (of Asiatic Eskimo) show little admixture with the Chukchi or Caucasian populations. However, in this same article Sukernik *et al*, stated that "some groups of Chukchi… assimilated their neighbors, Asian Eskimos, that populated the northern part of Beringia and the northeast shore of the Arctic Ocean" (p.2361). Additionally, Caucasian admixture was found in 9 out of 10 Chukchi villages. Thus, it is likely that Asian Eskimo mixed with non-Eskimo and Caucasoid. As mentioned earlier, generalized information about ethnic groups seems be incorporated in some articles without critical evaluation. In reality, researchers have only a very vague idea of the degree of intertribal and Caucasian admixture in indigenous Siberian populations.

High-resolution mapping revealed 34 distinct haplotypes of mtDNA from only three (A, C, D) of the four haplogroups (A, B, C and D) observed in Native Americans in the 10 populations of aboriginal Siberians. Asiatic Eskimos showed only haplogroup A and D mtDNAs (for the details about haplogroup distribution please see Table 10). Siberians also contained a significant proportion of "other", mtDNAs of probable Asian origin. Torroni *et al* specify that "other" haplogroups are those that do not belong to the haplogroups A, B, C, and D. The Siberians of the Amur region exhibit a close genetic affinity with Japanese, Koreans, and Han, suggesting a progression of population movements from central East Asia, to Siberia, and to the Americas, indicating that

Siberia was colonized by people related to modern Han and Koreans. However, the frequency of group A, C and D haplotypes in modern Han, Chinese and Koreans is low, relative to Siberians. Thus, while Siberian mtDNAs were clearly derived from Asian mtDNAs, the marked change (i.e., an increase in groups A, C, and D) in mtDNA frequencies in Siberia implies that there was a bottleneck during the formation of Siberian populations. The bottleneck was probably not as complete as that which gave rise to Native Americans, since 36.1% of Siberian mtDNA do not belong to haplogroups A, C, and D. These "other" mtDNAs show clear Asian affinities but are absent in Asiatic Eskimos, Na-Dene and Amerinds. While they could be derived from a recent admixture of Siberian and Asian populations, they are more likely to have been carried to Siberia by the Asian migration and subsequently to have been lost by the Native American migrations. Unfortunately, Torroni *et al* did not substantiate this conclusion. Contrary to their conclusion, it seems more likely that these "other" mtDNAs are a result of admixture since many of the studied Siberian populations border Asian populations. Also, it is unlikely that these "other" mtDNAs were altogether lost in Native American groups.

One of the most interesting Torroni *et al* findings is that the Siberian populations apparently lack haplogroup B (the same was found by Starikovskaya *et al*, 1998), which is widely distributed in East Asians, Melanesians, and Polynesian populations, and dispersed throughout the Amerinds. Surprisingly, the geographic location connecting Asian and Amerinds populations is devoid of this variant. Torroni *et al* argue, that this haplogroup might have been lost by genetic drift, although it is unlikely that all 10 populations would loose the same founding mtDNA lineage. Alternatively, the absence

of haplogroup B could be explained as the product of two different migrations, the first carrying the haplogroups A, C, and D, and the second carrying B. The most likely route for such an alternative migration of group B mtDNAs would be an expansion along the coast of Siberia. By this coastal route, the group B migration could have avoided contacts with Asiatic peoples inhabiting the tundra of eastern Siberia. Such a two-migration model could account for the relative paucity of genetic variation within Amerind group B haplotypes relative to those from group A, C, and D and for the prevalence of the founding haplotypes of haplogroup B in both Asians and Americans.

The fact that the Siberian and Native American mtDNA groups C and D share only the nodal and founding haplotypes (S26/AM43 for group C and S13/AM88 for group D) indicates that most of the mtDNA variation of the populations from these two regions arose independently on the two continents. This important finding means that little, if any, of the mtDNA diversity that currently exists in Native Americans arose in Siberia prior to the Amerind migration. Therefore, the genetic diversity that exists on each continent can be considered proportional to the time that these populations have been separated. The time required for the observed mtDNA differentiation was calculated by Torroni *et al* 17,000-34,000 YBP (Table 11).

Starikovskaya *et al* (1998) also conducted a similar study of mtDNA variation of 145 individuals from Chukotka: 66 Chukchi and 79 Asian Eskimos from Novoye Chaplino, Sireniki, Providenie, and Anadyr. Genealogical information was used to select unrelated individuals as potential blood donors.

Neither the Chukchi, nor the Asian Eskimo had haplogroup Y mtDNA (from "other" haplogroups), which is common in the Nivkhs and are also present in the Koryak

and Itel'men. Thus, the high frequency of novel haplotypes from haplogroups A and D in the Chukchi and Asian Eskimo, along with both the low frequency of haplogroup G (also "other" haplogroup) mtDNAs and the absence of haplogroup Y mtDNAs, clearly differentiates Chukotkan populations from the rest of the northern-Asian Mongoloid groups. But the Chukchi, not the Asian Eskimo had mtDNAs from Asian haplogroup G, indicating their genetic affinities with the Koryak and Itel'men of Chukotka.

The occurrence of distinct control region (CR) lineages and sublineages of haplogroup A in contemporary populations of the northern Pacific Rim may reflect the genetic history of human populations that existed in the Beringian and southern Alaskan refugia at the end of the last glacial maximum. Therefore, the Chukchi, Eskimo, and Na-Dene populations are likely to be remnants of the progenitors of the first Americans, who brought haplotypes A, C, and D to the New World, even though they currently retain much lower haplogroup A diversity than seen in Amerindians. The clear affinity between the mtDNAs from aboriginal Chukotkan populations and those of Native Americans, as compared with those of modern Mongoloid populations of northern Asia, is also supported by parallel studies of Y-chromosome polymorphisms (Lell *et al*, 1997) and analysis of variation at the GM locus (Sukernik, 1992). Thus, both the mtDNA and nuclear DNA (nDNA) data reveal the genetic differentiation between Chukotkan populations and adjacent Siberian groups, and also close genetic linkage of the Chukotkan populations to Native Americans (Table 12).

The authors support the two-wave model of the peopling of Americas, with the first bringing the A, C, and D haplogroups around 34,000 years ago and the second, bringing haplogroup B presumably from southeastern Siberia approximately 16,000-

13,000 YBP. The ancestors of the Chukchi and Eskimo, Na-Dene Indians and Northwest Coast Amerindians are the products of substantial genetic differentiation of populations that were occupying different glacial refugia during the end of the last glacial maximum, which is apparent from the diversity of haplogroup A CR sequences (Table 13).

Bonato and Salzano (1997), on the contrary, argued for an early and single (as opposed to a two-wave hypothesis) migration into the New World. They found that sequence diversity and population trees demonstrated that the Amerind, Na-Dene, and Eskimo were significantly closer among themselves than any of these to Asian (e.g. Siberian non-Chukchi, Mongolian or East Asian) populations, with the exception of the Siberian Chukchi, that in some analyses are closer to Na-Dene and Eskimo (Fig. 13). Thus, the Chukchi are as similar to Native Americans as to Siberians. This conclusion supports Rychkov *et al* (1972) and Solovenchuk (1982). Sequence diversity analyses based on haplogroup A sequences suggest that Native Americans and Chukchi originated from a single migration to Beringia, probably from east Central Siberia. This migration may have taken place approximately 30,000 or 43,000 years ago. These results support the model of the peopling of Americas in which Beringia played a central role, where the population that originated the Native Americans settled and expanded. Here Bonato *et al* conclusions parallel that of Starikovskaya *et al* (1998), but not Shields (1994), with respect to considerable genetic differentiation of putative ancestral populations. However, Bonato *et al* give a greater time depth for the peopling of both Northeast Siberia and the Americas, than is generally accepted.

Their argument for the single migration is supported by the findings of Neel *et al* (1994). These researchers studied human T-cell lymphotrophic retrovirus (HTVL-II) and

its distribution in 10 ethnic groups from northeastern Siberia: Chukchi, Eskimo, Nganasans, Yukagir, Even, Sel'kup, Koryak, Nivkh and Udege. It was reported that, as opposed to the widespread seropositivity to HTVL-II in Amerindians, the 10 Siberian populations exhibited a lack of virus. Specifically, of the 459 samples tested, only 4 were positive. However, the retrovirus was detected in Mongolia. Additionally, Northeast Siberian populations lack haplogroup B mtDNA. This haplogroup has been found in both the New World and Mongolia/southeast Siberia. Furthermore, the age of the haplogroups A, C, and D appears to be "younger" in Siberians when compared to Amerindian A, C and D haplogroups, because Siberian mtDNA diversity in these haplogroups is considerably less. This allowed Neel *et al* postulate that the most proximal Asian ancestors of all the Amerinds share a common origin with the indigenous people of the general region designated Mongolia/Manchuria/extreme southeastern Siberia. The most probable route to Beringia was the area adjacent to the Siberian Pacific Coast, where they left the archaeological sites dated (not without controversy) to some 30,000 YBP. The entry into Siberia was generally later, and those entrants were drawn from groups lacking both HTVL-II and the mtDNA haplogroup B. Neel *et al* hypothesis challenges the currently favored hypothesis that the direct ancestors of the Amerindians were primarily drawn from northern and central Siberia. The authors argue further that although Torroni *et al* (1993) have found that the mtDNA haplogroups C and D of Siberians and Amerinds can be traced back to a common ancestral haplotype present in both Siberia and the Americas, this does not necessarily mean that Amerindian haplogroups were derivatives of Siberian haplogroups. It is equally possible that the ancestral haplotypes were derived from lineages living further south, in populations ancestral to Amerindians and some

Siberian ethnic groups. The authors also note that the proportion of "other" mtDNA in the studied Siberian groups (with the exception of Eskimo) or mtDNA not falling into haplogroups A, C, and D (and B is absent) ranges from 2% in Evenks to 82.6% in Udege. Additional study of mtDNA among these populations, in conjunction with further knowledge of the mtDNA types of southeastern Asians, could substantially clarify the provenance of the Siberian native groups. An adequate test of this hypothesis, the authors conclude, depends on the accumulation of more data with respect to the ethnic distributions of both the HTVL-II virus and the mtDNA characteristics of these same ethnic groups, especially in Siberia and the areas to the south.

Further understanding of the origins of Asian Eskimo and Chukchi was facilitated by the study of male migration from Siberia to the Americas on the basis of Y chromosome analysis. Lell *et al* (1997) studied the origins of a polymorphism that involved C-T transition at nucleotide position 181 of the DYS199 locus, previously reported only in Native Americans. Y chromosomes from Siberian, Asian, and Native American populations were screened for DYS199 and other markers that included the DYS287 Y *Alu* polymorphic (YAP) element insertion and DYS287 (YAP+), and a YAP-associated A-G transition at DYS271 locus. The Siberian sample was comprised of Asian Eskimo, Chukchi, Evenk, Nivkh, Udege, Northern Altayan, Ket, Koryak, and Itel'men.

This survey detected the T allele (DYS199"T") in all five Native American populations with an average frequency of 61%, and in two of nine native Siberian populations: the Siberian Eskimo (21%), and the Chukchi (17%). The rest of the Siberian populations, along with Tibetans and Koreans, contained only the C allele (Table 14). Since Southeastern Siberia and Mongolia have been proposed as the ancestor Native

American populations, the absence of the T allele in eastern, western, and southern Siberian ethnic groups suggests that the original C-T mutation occurred in a putative ancestral Native American population in Beringia and was then spread throughout the New World by the founding populations of the major subgroups of modern Native Americans. The T allele would have been maintained in the ancestral Beringian population(s), which subsequently separated from the populations living farther south in the Americas due to glacial barriers. This Beringian population, became ancestral to modern day Eskimo-Aleuts and Chukchi, who could be of Beringian origin based on their genetic affiliations with other circumpolar populations (Szathmary, 1984, Starikovskaya, 1998). The presence of the DYS199 T allele in the Chukchi may be attributed to its presence in the ancestral population of the Paleoasiatic-speaking groups of Northeastern Siberia. This scenario is consistent with the age estimate of 30,000 years before present (YBP) for the original C-T mutation based on linkage with the DYS19 tetranucleotide repeat locus, and a microsatellite mutation rate of $1.5 \ \ 10^{-4}$. In addition, an early human entry into the Americas from Asia has been favored by mtDNA RFLP data (25,000-38,000 YBP, Torroni *et al*, 1993, Starikovskaya *et al*, 1997), mtDNA control region sequence data (20,000-25,000 YBP, Forster *et al*, 1996) and nuclear genetic variation studies (31,000 YBP, Cavalli-Sforza *et al*, 1994). Thus, paternal and maternal and nuclear genetic systems provide support for the early entry into the New World around 30,000 YBP, and for the maintenance and further divergence of lineages in isolated Beringian populations.

Lell *et al* hypothesis, although supported by other studies made on different genetic systems, was constructed on the basis of studying only one Y chromosome

polymorphism, additional Y chromosome markers must be studied in these and other populations to better order and define these events.

Two interesting studies regarding deletion-insertion polymorphism (indel) in the V-region of mtDNA, a so-called "Asian" deletion, implied the possibility of a Siberian-Polynesian relationship. The "Asian" deletion is a deletion of one copy of the two tandem copies of 9bp sequence CCCCCTCTA in the non-coding region of the mtDNA. It is found only in individuals from Asia and Oceania. Insertion of 4 CCCC is more rare than deletion. Petrischev *et al* (1993) studied indel polymorphism in 10 Mongoloid Siberian populations: Chukchi, Asian Eskimo, Yakut (Sakha), Buryat, Altai, Evenks, Mansi, and Nanai. No indel was found in Chukchi, Eskimo, Yakut, western Evenks, and Mansi. Other populations showed frequencies of the 9-bp deletion and 4-bp insertion as follows: 7.7% and 1.5% in Buryats, 10.2% and 0.8% in northern Altayans, 1.4% and 0.0% in Nanays, 1.5% and 0.0% in eastern Evenks. Correlation analysis of the geographic location of studied populations from Siberia, Asia and Oceania, and frequencies of polymorphisms was conducted. It showed a geographic gradient of the deletion frequency increasing towards the South where it was found in higher frequencies. In some cases it reached 100% in island isolates in Oceania (e.g. Samoa) and, to a lesser extent, towards the East. The frequency of the "Asian" deletion in Asian populations decreases in the direction from south-south-east to north-north-west, with the rate of 6% per 1,000 km. The authors challenge the conventional hypothesis that this deletion arose in the ancient Southeast Asian populations and spread by migration. They think that the observed cline may indicate that the deletion arose in the western part of the Pacific Ocean region, where its frequency reaches the world's maximum. The expansion of Southeast Asian

populations made the contacts possible between Polynesian and Asian populations. This facilitated the spread of the deletion into the northern direction. The authors note that many Asian and Siberian native groups, such as Japanese, Ainu, Nivkhs, Koryak, Aleut, and Eskimo, have Polynesian elements in their cultures. This implied if not direct contacts between Polynesian and Siberian groups, then at least indirect cultural contacts through the chain of coastal Asian populations. These contacts, however, were later, and were preceded by the initial peopling of the New World in the Upper Paleolithic by the populations that lacked this deletion. These would be the contemporary descendants who presently live in the circumpolar region of Siberia. The 9-bp deletion was perhaps brought to the New World much later. Other waves of migrants may have brought this deletion to Central and South America. This scenario explains the observed distribution of the deletion in Native American populations that is inverted in relation to the Asian-Pacific region. Petrischev, however, does not provide any references for the evidence of the Polynesian elements in the cultures of the Far East and Northeast Asian groups. Most of the ethnographic studies of Northeast Asians do not mention such connection.

Ivanova *et al* (1994) also studied the "Asian" deletion but using larger sample sizes and adding new native groups (Sel'kup). Ivanova *et al* obtained similar results for the geographic gradient in the frequency of 9-bp deletion and absence of the deletion and Ava II-site in Chukchi and Eskimo. However, the authors referring to Ballinger *et al*, note, that it has been found that the "Asian" deletion might have arisen independently a few times. Therefore, more extensive and wider analysis of the deletion distribution should be undertaken. Unfortunately, neither of these two studies included the Koryak, Nivkh, or Itel'men, which would be logical given their geographic location. Interestingly,

the later study of HLA class II loci polymorphism in Siberian populations clustered central Siberian populations (Evenk and Ket) with the Polynesians on the phylogenetic tree, constructed on the basis of genetic distances between human populations (Grahovac *et al*, 1998). Grahovac *et al* suggested that this clustering is either an artifact, or it reflects an ancient connection between the two population groups. They added that more extensive typing of the Siberian populations and sampling of non-Siberian populations will be necessary to resolve this unexpected affinity. Thus, this study supports the previous finding of a Siberian-Polynesian connection, but on the basis of different genetic markers.

Although the deletion has not been found in the studied Circumpolar Siberian populations, the connection between Polynesian and some Siberian cultures certainly deserves more attention.

Taken together there are three hypotheses resulting from molecular genetic studies. First a recent and rapid radiation of Eskaleut and Chukchi with the time depth 5, 100-7,100, and a radiation of Amerind and Escaleut 12,100-13,200 YBP. Second, a two-wave model of migration into the New World, with the first bringing haplogroups A, C and D of mtDNA 34,000 YBP and second bringing haplogroup B 16,000-13,000 YBP. Third, an early and single migration 30,000-43,000 or 22,000-55,000 YBP depending on the substitution rate. The implication of these dates for our research is that they deduce slightly different times of the populating of Northeast Asia, which on the basis of these studies point to the time around 30,000 YBP.

III. 2 **Koryak and Itel'men**

III.2.1 Serological studies

No serological studies of Itel'men have been conducted, perhaps due to the fact that they were considered to be very admixed. There were only a few serological studies of the Koryak group. Genetic structure of Koryak populations in Kamchatka were studied by Sheremet'eva and Gorshkov (1977, 1981, 1982), using 10 non-linked blood groups loci. Koryak populations of Bystrinski, Karaginski, Olyutorski, Penzhinski districts of the Kamchatka region, and Severo-Evenskii district of the Magadan region participated in this study. Samples were comprised of 906 to 96 individuals making up 15% to 1,5% of total Koryak population, respectively. No sampling procedure was given, and no account of modern or intertribal admixture was recorded.

Koryak populations were genetically "intermediate" between Siberian and Far East population. Unfortunately, in the manuscript Siberian populations were not specified. Their conclusion is supported by the archaeological and anthropological data demonstrating that the Koryak ethnic group was formed by the interaction between Neolithic continental tribes that moved to the Okhotsk Sea region, and Neolithic inhabitants of the Amur River and Primorie regions (Far East) that were moving northward. The time of genetic differentiation was found to be 5, 818years, which matches well the archaeological data. Effective size was found to equal 71 individual, or 29% of the total size. This figure is comparable with the estimates for Chukchi (31%), Eskimo (26%), Aleuts (29%), and Amerinds of South America (31%). Depending on the migration model, their estimates of gene migration between populations was m_e =

0.0218+0.0091 for the island model of migration[13], and m_e =0.0260+0.0014 for the stepping stone model[14]. Coefficients of migrations varied from 0 to 0.1381, which points to the different degree of isolation of subpopulations. Peripheral populations exchange genes with each other via the series of steps, and migration becomes dependent upon the geographic distance.

Solovenchuk *et al* (1985) studied 25 blood group and protein polymorphic systems to reveal the genetic structure of the 675 individuals of both sexes, 7 years of age and older, from 4 subpopulations of Kamchatka: Sedanka, Voyamploka, Tymlat, Karaga. People from admixed marriages were excluded. They chose subpopulations of Middle Kamchatka, because North Kamchatka Koyaks live in mixed Koryak-Chukchi villages, and in the Severo-Even district of Magadan region - in mixed Chukchi-Koryak-Even villages. However, the author notes, that his choice of populations does not exclude hidden admixture, which might seriously impede the interpretation of the results of this study. Since Sheremet'eva and Gorshkov chose their sample from the North Kamchatka populations, their results might have been influenced by the admixture if the sample was not chosen carefully. For the monomorphic loci Koryaks do not differ from the rest of Northeast Mongoloids. For the frequencies of the polymorphic loci please see Table16. Solovenchuk *et al* state that their results were in good concordance with the results reported by other researchers for ABO, Hp, and PTC, but were different from the Sheremet'eva and Gorshkov's (1977) for the MNS locus. Solovenchuk's results do not

[13] Island model of migration - a model where a large population is split into many subpopulations dispersed geographically like islands in an archipelago. Each population is assumed to be so large that random genetic drift can be neglected. In the island migration model the intensity of migration between subpopulations is assumed to be equal in both directions.

[14] Stepping stone model of migration- a discrete model in which each subdivision exchanges migrants only with its nearest neighbor.

show higher frequency of M allele over N allele. In 7 out 15 loci the phenotype frequencies in Koryak subpopulations are different from those of Asian Eskimo, Reindeer and Coastal Chukchi. This indicates that along with similarities between these groups, Koryak have genetic structure that differs them from the rest of the Northeast Asian populations for some loci. Genetically Koryak are closest to the Reindeer Chukchi and most distant from Asian Eskimo and Alaskan Eskimo. Solovenchuk *et al* also considered genetic similarities between the Reindeer Chukchi and Koryak with respect to their geographic location, life style, and diet. They state that they were not able to determine intrasubpopulation or intrapopulation (between Koryak, Reindeer Chukchi, Coastal Chukchi and Asian Eskimo) heterogeneity, attributing this fact to "ecological factors". As in the earlier studies, "ecological factors" were not defined, and no alternative explanation was suggested.

Thus, the Koryak were found to be intermediate between Siberian and Far East populations, and closer to Reindeer Chukchi, with whom they are neighbors, than to Eskimo populations. Since serological studies were so few, these inferences about genetic affinities should be tested using more informative molecular genetic methods.

III. 2. 2 Molecular genetic studies

Derenko and Shields (1997, 1998a, 1998b) studied hypervariable segment I (HVS I) of the control region (CR) of mtDNA. The sample included 122 representatives of the three ethnic groups from the North Asia, Yakuts n=22 (Sakha Republic), Even n=65 (Severo-Evenskii and Ol'skii districts of Magadan region), Koryak n=35 (Severo-Evenskii district). Representatives from other populations were also included (Buryat,

Itel'men, Chukchi). No donors were related and each stated that they belonged to their respective ethnic groups. Data were compared with the information on nucleotide sequences in HVS I mtDNA in 8 Mongoloid populations of Siberia and Eastern Asia: Chukchi, Eskimo, Yukagir, Altai, Mongol, Chinese, Koreans and Ainu. Altogether 59 types of the CR of mtDNA were discovered. 49 types were unique e.g. belonged to the individuals from particular ethnic groups, 7 types were common for two populations, and 1 type was found in three populations. Thus, the genetic structure of each ethnic group is very specific with respect to unique types of mtDNA, namely, 80% of Koryak, 78% of Even, and 59% of Yakut possess unique types of mtDNA. The types of the CR of mtDNA found in three populations belong to 9 monophylogenetic clusters. Out of these nine clusters, 4 could be considered as ethno-specific. Clusters K2 and K9 are Even - specific clusters, and K3 and K5 are Koryak-specific with 54.3% Koryak possessing ethno-specific haplotypes. A comparison of the distribution of these clusters among other Asian populations shows that ethno-specific clusters remain to be ethno-specific for the Koryaks K3 and K5, and for the Even clusters K2. The Even K9 cluster with the identical nucleotide sequences was found to have frequency (19.2%) in Japanese Ainu population, possibly due to the recent gene flow between these populations. Nucleotide variation (in % of divergence) of this cluster in Ainu population is 0.357%, which exceeds the variation in Koryak and Even -0.139%. This points to the direction of migration from Northern Japan to the coast of Northern Asia.

Clusters found in individuals from all three ethnic groups were also found in other populations of Siberia and Eastern Asia. These clusters, K1, K4, K6, and K8, could be considered to be race-specific. Their evolutionary age is approximately 36,700, 27,000,

53,430 and 87,200 years. The Koryak ethno-specific clusters K3 and K5 were estimated to be -22,000 and 20,200 years old respectively.

Thus, in the mitochondrial genome of the Koryak and Even two genetic components are found that differ in age and supposedly correspond to two stages of genetic differentiation on the race level and on the ethnic group level. Derenko and Shields argue that the evolutionary age of mitochondrial lineages is much older than age of Koryak and Even ethnic groups, estimated according to the ethnographic and archaeological data (8-10,000 years ago) (Gurvich, 1982), and corresponds to the age of genetic substratum, on the basis of which the formation of ethnic groups took place. The two Koryak-specific clusters K3 and K5 with similar values of nucleotide sequence divergence might be considered to be two founding haplotypes of Koryak. Derenko and Shields also found that just like Chukchi and Eskimo, Koryak and Even do not possess the "Asian deletion" polymorphism in the region V of mitochondrial DNA.

The most comprehensive study of Koryak and Itel'men so far has been conducted by Schurr *et al* (1999). They analysed mtDNA variation in 202 individuals, 104 Koryaks from the three geographically approximate villages Karaga, Ossora, and Tymlan in the Karaginskiy District of the Koryak Autonomous region and 51 Koryak and 47 Itel'men from the villages of Voyampolka and Kovran (Tigil'skii district of the Koryak Autonomous region). All individuals were interviewed about their family histories, which in turn were verified by senior members of the community for accuracy. Only those people lacking maternal or paternal Russian or non-related ancestry through three generations were selected for the collection of blood samples. Based on the genealogical data, approximately half of the Koryaks and most of the Itel'men were estimated to be of

mixed Russian - Koryak or Russian-Itel'men ancestry, respectively, but considered themselves Koryak or Itel'men by nationality primarily because of their maternal ancestry.

High resolution RFLP analysis of 202 Koryak and Itel'men mtDNA revealed a total 22 distinct haplotypes defined by 48 polymorphic sites. Three of the four haplogroups (A, C, and D) observed in Native Americans occurred in Kamchatkan groups, and these encompassed approximately 43% of all Koryak mtDNA and 21% of Itel'men mtDNA, with the majority of these belonging to haplogroups C. Consistent with previous studies of North-east Asia, Kamchatkan groups also lacked haplogroups B mtDNAs, suggesting these mtDNAs were never present in Paleoasiatic -speaking groups. In addition, none of the Koryak or Itel'men individuals had mtDNAs from haplogroups typically seen in European populations, indicating that they had not experienced non-native gene flow through their maternal lineages. The majority of the Koryak (58%) and Itel'men haplotypes (80%) did not belong to haplogroups A, C and D, and as such they could technically be defined as "Other" (G, Y and Z) mtDNAs. Kamchatka harboured the highest frequencies of haplogroup G mtDNAs, which were widely distributed in eastern Siberian and adjacent East Asian populations. The distribution of haplogroup Y was restricted within a relatively small area and pointed to the lower Amur River-Sakhalin Island region as its place of origin.

The mtDNA distribution in the Koryaks and Itel'men was also quite different and genetic differences were statistically significant. Some haplotypes were shared between the two groups, the majority of these were the founding, or nodal, haplotypes for haplogroups C, G, Y, and Z. The same extent of divergence was observed when the

mtDNA variation in all three Paleoasiatic -speaking populations was assessed. Chi-square analysis of the Chukchi, Koryak, and Itel'men mtDNA distribution revealed statistically significant differences between them. Assuming that their languages are closely related, these results could indicate that Paleoasiatic-speaking groups might have undergone significant genetic differentiation since sharing a common origin in Northeast Siberia. Alternatively, these differences could suggest a separate origin and expansion of these populations in this region, with their linguistic affiliations reflecting the considerable language sharing which has taken place over the past millennia. In the case of Chukchi and the Koryaks, whose linguistic connection is more strongly supported, it appears that the Chukchi have become genetically distinctive from the Koryaks through gene flow with Siberian Eskimos and perhaps other ethnic groups from this region, such as Yukagir, and Evens.

There were no significant differences in haplotypic composition between persons who call themselves Maritime or Reindeer Koryaks by ancestry, irrespective of their village of origin. Based on these results, Reindeer Koryaks appeared to be genetically synonymous with Maritime (Coastal) Koryaks, not a separate subgroup of this population, despite speaking a different dialect and practising a different subsistence strategy. While these nonsignificant differences may reflect the fact that a certain proportion of the Koryak individuals sampled were not completely certain of their maternal ancestry in terms of Coastal or Reindeer Koryak ethnicity, it is more probable that they reveal the degree to which sedentary and nomadic groups have become mixed in the past several centuries. However, their assumption contradicts Gurvich (1980) who

stated that Reindeer and Coastal Koryaks spoke different dialects and rarely intermarried (see Chapter I).

Phylogenetic analysis of Siberian haplotypes, using the neighbour-joining tree method (NJ), shows that the Paleoasiatic-speaking groups were split into two separate branches, one representing Chukotka, and the other Kamchatka. The NJ tree demonstrates a large split between Paleoasiatic-speaking populations with the Koryak and Itel'men showing much closer affinities to the Ainu, and the Chukchi being more closely linked with Asian Eskimos and Northwest Coast Amerindian populations. Several measures of haplotypic diversity showed that the Koryak and Itel'men populations were genetically very similar to each other but quite distinct from the Chukchi who are linguistically related to Koryaks. The Kamchatkan groups were also divergent from those which evolved from the ancient Beringian gene pool, such as Eskimo-Aleut and Na-Dene Indians, suggesting that their ancestral populations replaced the survivors of the Bering Sea land bridge in this region during the Neolithic period. However, the possibility that 2 haplotypes (SIB41 from haplogroup A and SIB40 from haplogroup D), that link these earlier populations and Koryaks, were acquired through recent gene flow with the neighbouring Chukchi in whom these haplotypes are common, cannot be excluded.

Koryaks and Itel'men, although being closer to one another than to other Siberian populations, also showed significant differences between them. On the one hand, the Koryak and Itel'men shared the putative founding mtDNAs of haplogroups C, G, Y, and Z, suggesting that they might have originated from a common ancestral population in the Okhotsk Sea region. On the other hand, they exhibit significant differences in haplogroup frequencies and distribution, with nearly all unique CR sequences occurring in one

population or the other. These results support other linguistic and cultural evidence that the Itel'men and Koryak populations arose from temporally distinct expansions into the Kamchatka Peninsula, with the ancestral Itel'men being first to enter this region during the Siberian Neolithic. There are differences between Koryak subgroups. Although sharing several putative founding mtDNAs, the Aluytor, Karagin, and Palan Koryaks exhibited significant differences in haplogroup frequencies and haplotypes distribution. Aluytor and Karagin Koryaks appeared to be more genetically similar to each other than either was to Palan Koryaks, as expected from their closer linguistic association. Although difference in the distribution of the two major CR sublineages from the haplogroup C separated Aluytor Koryaks from the other two subgroups, this could be attributed to considerable admixture between Karagin and Palan Koryaks and Itel'men due to marital exchanges. In general, despite considerable amalgamation of Koryak population as result of intermarriages with non-natives and Russians, the fusion of different settlements into larger communities with the subsequent mixture of different Koryak subgroups, and epidemics, the results of this study suggest, that remnants of the former territorial subdivisions of Koryaks have persisted into modern times, despite the enormous demographic impact of Russian colonisation.

Shurr *et al* argue, that mtDNA data is consistent with archaeological and ethnographic data which show, that during the mid-Holocene climatic optimum, at approximately 6,000-4,000 YBP, there was substantial population growth in the littoral area of the Okhotsk Sea region associated with the spread of continental cultures of reindeer hunters from the Lena and Kolyma River basins. The expansion of these continental tribes into the northern Okhotsk Sea region apparently gave rise to the

ancestral Koryak and Itel'men populations, whereas movement from the lower Amur River-Sakhalin region appeared to have played a supplementary role in their origins. Moreover, the expansion of Neolithic "Southern Okhotsk" cultures into northern Japan from the lower Amur River region might have substantially contributed to the origins of the Ainu. This scenario, based primarily on archaeological data, suggests that the genetic profiles of the Koryak and Itel'men should be distinctive from those of the Nivkhs and Ainu. This interpretation is supported by mtDNA data. On a broader scale, the expansion of Paleoasiatic-speaking peoples into Northeast Asia led to near total replacement 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 north-western Pacific. However, the authors note, the mtDNA data indicated that, while absorbing elements of the Eskimo-Aleut culture during their expansion, ancestral Koryak and Itel'men groups did not extensively incorporate members of these maritime tribes. Both 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 recently evolved genotypes and cultural traditions from Northeast Asia overlapping and/or replacing more ancient ones.

The study by Grahovac *et al* (1998) used the HLA system to study the relationship among Siberian populations. Allele frequencies from five HLA class II loci in seven Siberian populations (Chukchi, Eskimo, Koryak, Even, Udege, Ket, and Nivkh), were determined. A greater number of HLA class II haplotypes have been found in Siberian populations than in other populations. The total number of Siberian haplotypes encompassing three of the four typed loci (DRB1, DQA1 and DQB1) is about the same

as the total number of haplotypes reported to 1996 for all the non-Siberian populations worldwide. The haplotypes fall into two categories: (1) those found in both Siberian and non-Siberian populations ("public" haplotypes) and (2) those restricted to Siberian populations ("private" haplotypes). The bulk (83%) of the haplotype diversity in the Siberian populations is constituted by private haplotypes. Most of the new haplotypes were generated in Siberia by recombination, and are part of the haplotype pool that is turning over rapidly (one recombinanat in 15 generations). The authors suggest that a set of "public", or shared haplotypes were brought to Siberia with the colonisers approximately 1600-2000 generations ago (~40,000 years ago).

The allelic frequencies at the DRB1 locus place all studied populations with other Mongoloid populations into one group that is separate from that of Caucasoids. Also, Siberian populations are divided into eastern (bearing all the eastern Siberian populations together with the Alaskan Eskimo and Amerinds) and central Siberian branches (bearing the two central populations, Evens and Kets, together with Polynesian populations).

The latest analysis of the genetic structure of the indigenous populations of Siberia on the basis of blood group and protein markers, and DNA variable number of tandem repeats (VNTR) variation, was undertaken by Crawford *et al* (1997). A "genetic map"[15] based on 15 populations, 7 loci and 8 alleles is illustrated in Fig.15a and 15 b. This plot of the first versus the second scaled eigenvectors explains more than 74% of the

[15] Populations structure in these populations is represented by the method of Harpending and Jenkins (1973). Sample allelic frequencies are converted into a relationship matrix, R, of dimension (L L), where L is the number of sample groups. The diagonal elements of R, r ii, describe the overall deviation of allelic frequencies for each sample population *i* from the mean allelic frequencies of the array. The weighted mean of the diagonal of the relationship matrix is equivalent to Rst, the mean genetic heterogeneity of all populations. Hence the closer the value of r ii is to Rst for any sample population i, the closer the allelic of that populations are to the mean frequencies of the array. Relative genetic relationships between populations can be graphically represented by a least-squares approximation of the R matrix. Reduced space, eigenvectorial representations provide two-dimensional "maps" of allelic frequency distribution (Crawford, 1981).

total variance. Russian sample is separated from the indigenous Siberian and Mongol populations by the first axis. The second axis clusters the Paleoasiatic-speaking populations from the Forest Nentsy and Turkic speakers. The separation of Paleoasiatic groups is primarily due to the relatively high frequencies of ACP1*A allele and low frequency of GC*1. The "genetic map" of 13 populations based on 5 loci and 9 alleles show the total variance subsumed by the first and second axes to be 65%, slightly lower than found in the previous genetic map for 15 populations upon 7 loci (Fig. 16a and 16b). The first axis separates the Paleoasiatic-speaking groups from the Forest Nentsy group and the remaining populations. The Coastal Chukchi are located between the Asian Eskimos and the Reindeer Chukchi, again documenting the gene flow between these groups in coastal settlements. The dispersal of the Paleoasiatic groups is due to the high frequencies of the GM*A G (GM*A, ZG) haplotype and PGM1*1 allele, the latter one being particularly frequent in both Reindeer and Coastal Chukchi.

A plot of the regression[16] (Fig.17) of mean per locus heterozygosity on r_{ii} shows that Eskimo and Chukchi groups differ from other Siberian populations according to their low heterozygosity levels (associated with below average external gene flow) and genetic isolation from the other groups.

[16] The relationship between heterozygosity and genetic distance, developed by Harpending and Ward, may be represented by a two-dimensional figure whose ordinate is genetic distance from the centroid r_{ii} and whose abscissa is mean per locus heterozygosity, H_0. Mean per locus heterozygosity is computed as $H_0 = 1-(\sum p_i^2/I)$, where p_I is a frequency of the ith allele, and I is a number of loci. These values of H_0 and r_{ii} are computed directly from observed allelic frequencies. An expected mean per locus heterozygosity, is then computed by linear regression. Under the assumption of uniform systematic pressure, $H_0 = H_e$. Deviation from this identity for any sample population is the result of differential systematic pressure relative to the rest of the samples. Positive deviation may be interpreted as admixture (gene flow), negative deviation as drift (or founder effect) (Crawford, 1981).

The Mantel tests[16] have indicated a strong correlation of allele frequencies with geography and with linguistics, which are all significant. The joint effects of language and geography explain a total of 30.6% of the variation in the genetics of Siberian populations. (Table 17). Judging from the moderate correlation between genetics and linguistics and the insignificant relationship between genetics and linguistics when geography is kept constant, it appears that most of the genetic differentiation in Siberia is geographically patterned.

[16] Statistical comparisons of matrices permit the examination of the relationship among genetics, geography and linguistics. Given two distance matrices A and B, Mantel tests examine an association between them by using the statistic $Z_{AB} = \quad A_{ij} B_{ij,}$ where A_{ij} and B_{ij} are elements of row i and column j of matrices A and B (Crawford, 1997).

Chapter IV. Conclusions

Serological studies of Northeast indigenous Siberian populations have shown the following results.

Populations structure of Chukchi and Asian Eskimo

Polymorphic loci with significant heterogeneity of allele frequencies has been observed among Chukchi populations and between Chukchi and Asian Eskimo for MNS, P, and Rh, loci (Sukernik *et al*, 1981), PGM_1 and Gm (Sukernik *et al*, 1986). Sukernik *et al* assume that these systems more than others were subjected to the action of random genetic drift. Hardy-Weinberg equilibrium was found for almost all loci, with the exception of: PGM_1 which in populations of Chukchi and Asian Eskimo had a deficit of heterozygotes and excess of homozygotes (Sukernik *et al*, 1981), Pp locus (Nazarova, 1989); GPT in Chukchi populations (excess of heterozygotes), and Gc (a deficit of heterozygotes) (Solovenchuk *et al*, 1982). Linkage disequilibrium was found for pairs of loci MN and Ss (MNSs system), Cc and Ee (Rh system); and three pairs of unlinked loci PGM1 and Hp (for Chukchi and Eskimo), PGM1 and AcP (Chukchi and Eskimo), and AcP and Hp (Chukchi). PGM_1^1 and AcP alleles were found to be selectively advantageous in the circumpolar zone (Vibe *et al*, 1990).

The level of heterozygosity (H) in both ethnic groups was first found not to differ substantially from more southward groups of Mongoloids. Asian Eskimo and Reindeer Chukchi had values of H approximately equal, but both groups were different from Coastal Chukchi (Solovenchuk *et al*, 1984). However, in his later study (Solovenchuk, 1989) the degree of heterozygosity in blood protein genes was found to be greater in the northernmost groups. Asian Eskimo showed lower heterozygosity than Chukchi and the

lowest among other groups of Eskimo (St. Lawrence Island, Alaskan, Greenland, Canadian Eskimo) (Nazarova, 1989). Thus, the results of the studies are different. The regression analyses of average heterozygosity (H) on the distance from the centroid (r_{ii}) reflects specific events in population histories of studied villages. For example, the position of Sireniki and Novoye Chaplino below the regression line indicated bottleneck effect in the histories of these villages due to epidemics. Alternatively, it assumes gene flow into population, e.g., Chukchi individuals from Kanchalan, Meinypil'gino, and Rytkuchi (Sukernik *et al*, 1986a), and Asian Eskimo from Naukan (Sukernik *et al*, 1986b) were positioned above the regression line, indicating migration into these populations.

In general, the genetic structure of studied populations is most probably formed by the migration and random genetic drift.

Genetic affinities between Chukchi and Asian Eskimo

Two serological studies have shown that the genetic structure of Chukchi matches their geographic location among Asian Eskimo only partially, which might indicate that they migrated to this region relatively recently (Rychkov & Sheremet'eva, 1972), and might additionally indicate numerous translocations of the closely related groups to other parts of the territory for the past few tens of years (Sukernik *et al*, 1986b).

Asian Eskimo and Chukchi occupy an intermediate position between Mongoloids of Siberia and Eskimo of the New World, being closer to the Siberian populations (Coastal Chukchi still maintain kinship ties with Reindeer Chukchi) (Rychkov & Sheremet'eva, 1972).

Compared to Alaskan Eskimos, genotypically Coastal and Reindeer Chukchi are closest to each other, while Eskimos and Reindeer Chukchi are the most distant. Asian Eskimo, Coastal Chukchie and Alaskan Eskimo have about the same genetic distance between each other (Solovenchuk *et al*, 1984). Genetically closest to each other were Sireniki and Novoye Chaplino Eskimo populations, and Novoye Chaplino and St. Lawrence Island populations (Sukernik *et al*, 1986b, Crawford *et al*, 1981). Also, Chukchi are found to be close genetically to Koryaks (Solovenchuk, 1985), Evens (Solovenchuk & Glushenko, 1985, Posukh, 1990), which is explained by the intertribal admixture due to expansion of Evens to the North; and Nganasans (Posukh, 1990).

Genetic affinities of the Koryak

Koryak populations belong to Mongoloids of North Asia, and are intermediate between Siberian and Far East populations, revealing their most genetic similarity with Siberian populations (Sheremet'eva & Gorshkov, 1977). Kamchatka Koryaks are rather similar to other ethnic North-East Asiatic groups, being the most approximate to Reindeer Chukchi and the most remote from Alaskan and Asian Eskimos (Solovenchuk *et al*, 1985).

Taken together, most interpretations of the above outlined studies have been suggestive rather than conclusive. A considerable amalgamation of native populations due to various degrees of intertribal and Caucasoid admixture and resettlement programs makes it difficult to infer conclusions in relation to the genetic structure of pre-contact populations. In addition, since blood groups and protein polymorphisms are considered to be "adaptive" traits (Giblett, 1969), it was hoped that native populations might serve as a

unique "nature laboratory" for studying natural selection. However, the conclusions regarding some allele frequencies having selective advantage in high latitudes have not been convincing, despite attempts of such researchers as Rychkov, Solovenchuk, Sukernik, and Vibe. The action of natural selection has proved to be difficult to document because of the statistical noise associated with small groups (Crawford, 1997).

Molecular genetic studies of the Chukchi and the Eskimo groups

Molecular genetic methods have proved to be more informative when compared with serological methods, and added new dimensions to the evolutionary research of indigenous Siberian populations. New information allowed new hypotheses regarding peopling of the New World to be put forward. The first hypothesis argues for the early and single entry into the New World, and the second supports a two-wave migration.

Shields *et al,* (1993) proposed a hypothesis according to which the evolutionary radiation of Circumarctic groups occurred in the shallow time depth, approximately 5,100-7,100 years ago. However, the small sample sizes might have considerably influenced their results. Also, a significant reduction in molecular diversity of mtDNA in Chukchi and Eskimo has been found by Shields *et al* (1994). Malyarchuk (1996) reported that this finding is in marked contrast with the increased degree of heterozygosity in blood protein genes found in the Northeast populations. As Jorde *et al* (1995) noted, there is a discordance between mtDNA and nDNA sequence diversities, and the degree of relatedness between populations differs depending whether mtDNA or nDNA data are used for constructing a phylogenetic tree. The basis of this discordance between mtDNA diversity and blood-protein gene heterozygosity remains unclear. Solovenchuk (1989)

proposed that blood protein gene heterozygosity increases in proportion to climatic severity. Since mtDNA polymorphisms might not be selectively neutral, it is possible that the loss of mtDNA diversity in northern populations reflects a greater degree of selection for optimal mtDNA haplotypes under more extreme conditions.

Nucleotide sequence analyses and population trees showed that the Amerind, Na-Dene, and Eskimo are significantly closer among themselves than any of the above three to Asian populations, with the exception of the Siberian Chukchi, that in some analyses are closer to Na-Dene and Eskimo. Nucleotide and diversity analyses based on haplogroup A sequences suggest that Native Americans and Chukchi originated from a single migration to Beringia, probably from east Central Siberia, which occurred approximately 30,000 or 43,000 years ago (with 95% confidence intervals between approx.22, 000 and 55,000 years). These results support the model of the peopling of the Americas in which Beringia played a central role, where the population that originated the Native Americans settled and expanded. Some time after the colonization of Beringia they crossed the Alberta ice-free corridor and peopled the rest of the American continent. The collapse of this ice-free corridor during a few thousand years approx. 14,000-20,000 years ago isolated the people south of the ice-sheets, which gave rise to the Amerind, from those still in Beringia; the latter originated the Na-Dene, Eskimo, and probably the Siberian Chukchi. Their close affinity with Eskimo and Na-Dene is further corroborated by classical genetic studies, and by language, suggesting that they all may have a common origin. Chukchi diversity and expansion time are smaller than those of the Amerind and, especially, than the Na-Dene and Eskimo, and this is consistent with their having a more recent origin (Bonato & Salzano, 1997).

Y chromosome study (Lell *et al*, 1997) detected the T allele in all five Native American groups and in both Eskimo and Chukchi groups. This allele is thought to be originated in Beringia and then spread throughout New World. The T allele would have been maintained in the ancestral Beringian population(s), which subsequently became separated from the populations living farther south in the Americas due to glacial barriers. This Beringian population, in turn, became ancestral to modern day Eskimo-Aleuts and Chukchi, who are postulated to have had Beringian origin based on their genetic affiliations with other circumpolar populations. The presence of the DYS199 T allele in the Chukchi may be attributed to its presence in the ancestral population of the Paleoasiatic- speaking groups of Northeast Siberia.

Only three of the four haplogroups (A, C, and D) observed in Native Americans were found in Siberians, including Chukchi, Koryak, Nivks and others, in the study of mtDNA variation by Torroni *et al* (1993). Asian Eskimo showed only haplogroup A and D. Siberians also contained a significant amount of "other" mtDNAs of probable Asian origin. This suggested that both Siberian and Native American populations derived from Asian populations, which underwent a series of bottlenecks. The Siberians of the Amur region clearly show a close genetic affinity with Japanese, Koreans, and Han, indicating, that Siberia was colonized by people related to modern Han and Koreans. Most of the mtDNA variation of the populations from these two regions arose independently on the two continents. This important finding means that little, if any, of the mtDNA diversity that currently exists in Native Americans arose in Siberia prior to the Amerind migration. Therefore, the genetic diversity that exists on each continent can be considered

proportional to the time that these populations have been separated. The time required for the observed mtDNA differentiation would be 17,000-34,000 YBP.

Starikovskaya *et al*, (1998) also detected mtDNA from only three haplogroups, A, C, and D, but not B, in the Chukchi and Asian Eskimo. The high frequency of novel haplotypes from haplogroups A and D in the Chukchi and Asian Eskimo, along with both the low frequency of haplogroup G mtDNAs and the absence of haplogroup Y mtDNAs, clearly differentiates Chukotkan populations from the rest of the northern Asian Mongoloid groups (such as Koryak and Itel'men). But the Chukchi, not the Asian Eskimo had mtDNAs from Asian haplogroup G, indicating their genetic affinities with the Koryak and Itel'men of Chukotka. The Chuckhi, Eskimo, and Na-Dene populations are likely to be remnants of the progenitors of the first Americans, who brought haplotypes A, C, and D to the New World, even though they currently retain much lower haplogroup A diversity than seen in Amerindians. Y chromosome, and both the mtDNA and nDNA data reveal the genetic differentiation between Chukotkan populations and adjacent Siberian groups, as well as their close genetic linkage to Native Americans. The authors' scenario of the peopling of the New World is as follows. First, approx. 34,000 YBP, ancient Beringia harbored a population(s) that contained haplogroups A, C, and D and gave rise to the first Americans. Second, approximately 16,000-13,000 YBP, a later migration brought haplogroup B, presumably from southeastern Siberia (this hypothesis is also supported by Torroni *et al*, 1993). Third, the ancestors of the Chukchi and Eskimo, as well as those of the Na-Dene Indians and North-West Coast Amerindians, are the products of substantial genetic differentiation of populations that were occupying different glacial refugia during the end of the last glacial maximum.

Molecular genetic studies of the Koryak and the Itel'men

Derenko and Shields (1997, 1998a, 1998b) in their three sequential studies of Koryak, Even, and Yakut, found that compared with other Northern Mongoloids these groups posses relatively high mtDNA diversity. Out of 9 clusters, 4 were ethnic-specific: K2 and K9 could be considered Even-specific, and K3 and K5- Koryak specific. Cluster K1, K4, K6, and K8 ("public") correspond to the evolutionary age of 36,700; 27,000; 53,430; and 87,200 years, respectively. The evolutionary age of cluster K3 and K5, which include nucleotide sequences of Koryak, is calculated to be 22,000 and 20,200 years old respectively. The time of divergence for the K2 and K9, that includes only Even-specific nucleotide sequences, is 8,800 and 8,100 years old respectively, which is the "youngest" evolutionary age, indicating the time when Evens, or their ancestors, diverged from the ancestral Siberian population. Koryak and Even do not posses 9-bp deletion in the region V mtDNA.

Schurr *et al* (1999) have found that the Koryak and the Itel'men, despite a third of their gene pool consisting of haplogroup A, C, and D mtDNAs, were not closely genetically related to Native American groups. They have stronger genetic affinities with eastern Siberian/east Asian populations. Koryak and Itel'men populations were very similar to one another but quite distinct from the Chukchi, who are linguistically related to the Koryaks. Kamchatkan populations were also quite divergent from the ancient Beringian gene pool, such as Eskimo, suggesting that their ancestral populations replaced the survivors of the Bering land bridge in this region during the Neolithic period. Although sharing some putative founding haplotypes between them, suggesting that the Koryak and Itel'men populations might have originated from a common ancestral

population in the Okhotsk Sea region, they also exhibit significant differences in haplogroup frequencies and haplotype distribution. This supports other linguistic and cultural evidence that the Itel'men and Koryak arose from temporally distinct expansions into the Kamchatka Peninsula, with the ancestral Itel'men being first to enter this region during the Siberian Neolithic. Reindeer Koryaks appeared to be genetically synonymous with Coastal Koryaks, not a separate subgroup of this population, despite speaking a different dialect and practising a different subsistence strategy.

Finally, two studies revealed a possible relationship between Siberian and Polynesian groups (Petrischev *et al*, 1993, Ivanova *et al*, 1994).

Having described the details of genetic studies, it is time to discuss the problem of sampling procedure, which appears not to have been closely scrutinised by most geneticists in the studies analysed in this thesis.

Chapter V. Sampling procedure and its effect on the interpretation of results of genetic studies.

So far most of the studies in this overview considered admixture in the native populations to be negligible and generally it was estimated at approximately 1-5%. An examination of the genealogies of a sample population suggests that this may be much too low. Shurr *et al* (1999) used genealogical data for the selection of individuals for their sample, and these family histories were verified by the community elders for accuracy. Their estimation of admixture is dramatically different: 50% of Koryak and most of the Itel'men were of mixed Russian ancestry, who nevertheless considered themselves Koryak and Itel'men primarily because of their maternal ancestry. It is worth pointing out that peoples' ethnic identity sometimes follows paternal ancestry and may also depend on the viagaries of other, ideological reason or in order to take advantage of shifting state policies. Thus, sampling procedures described in the analysed articles deserve a close scrutiny, due to the importance of this stage in a research project, and possible implication of the sampling error on the conclusion of a study.

It appears that most researchers pay a lot more attention to details of laboratory and statistical analyses, which undoubtedly are very important too, than to the choice of individuals who will represent their ethnic groups in a study. However, if a study of the genetic structure of a population and genetic affinities between populations allows some room for admixture, the results of studies of phylogenetic relationship might be seriously obscured by undetected admixture. A survey of sampling procedures shows that despite the possibility of these complications, many researchers adopted a somewhat simplified approach to this problem, namely, that individuals are selected on the basis of an

interview where they state that they belong to a specific native group, and without further verification are labeled as such, and are treated as a representatives of the named native group.

Thus, a total stranger (a researcher) may come to a village for a short time and ask questions about perhaps some of the most intimate sides of a person's life, such as paternity or admixture, often speaking to older people who do not even know the Russian language well enough. The researcher is often satisfied with just a simple statement. Some of the villages have been visited many times, and yet the results of a study are rarely disclosed to those who participated in the study. Many villagers are quite sceptical about the purposes of these research undertakings. It is worth pointing out that social anthropologists spend much of their fieldwork and their theoretical discussions grappling with exactly this problem. So, to what extent can we rely on ethnic self-perception and self-identification of the study subjects without employing extensive genealogical information before choosing individuals for genetic sampling? A small survey undertaken here while carrying out the literature overview shows some trends.

Some articles do not describe the sampling procedure at all (Nazarova, 1989). Sukernik *et al* (1986b) state in their article that they composed their sample of "pure" Asian Eskimo individuals. Though their assumptions would seem absurd to social anthropologists, they might have gone unchallenged, except for their discovery of an Rh-phenotype cde, which is unusual for the Eskimo, in one Asian Eskimo man, who considered himself to be "pure" Eskimo and who had a typical Eskimo appearance. This prompted further genealogical investigation through the help of a third party who consulted with the village elders, who then confirmed that this individual had a

Caucasoid admixture. It is clear that a person's self-identification as to belonging to a certain ethnic group sometimes does not give a complete picture of his/her genetic ethnicity. In another study, Sukernik *et al* (1986a) detected traces of Caucasoid haplotype Gm (f;b0135) in 9 out of 10 studied Chukchi populations of Chukotka and Kamchatka Peninsula populations, and although they estimated this admixture to be on average 1.3%, some villages had a considerably higher per cent (4.3% in Kanchalan). In an earlier study of blood groups and protein polymorphisms in Reindeer Chukchi and Asian Eskimo, Sukernik *et al* (1981) stated that gene flow from non-Chukchi and non-Eskimo was a rare event until the early 1940s in Eskimo and 1960s in the Reindeer Chukchi. This is surprising, knowing that from recent and ancient history of these populations that they admixed with Evens (Solovenchuk and Glushenko, 1985), and Koryaks (Solovenchuk *et al*, 1985). The authors also underestimate the high mobility of natives in these regions before the Soviet period. The authors also mention that their results significantly differ for Novoye Chaplino from those of Rychkov and Sheremet'eva (1972) with regard to Kell, Rh, Duffy, and Diego. They attribute this partly to their own exclusion from the sample of individuals who had one non-"Eskimo" parent. Such a statement obviously sows a seed of doubt about the correct interpretation of the differences of the results in these two studies: whether they are different because of the influence of factors such as migration, selection, and genetic drift, or because the sample was not as "pure" as it could have been.

Shields *et al* (1993) state that they interviewed donors regarding their knowledge of their maternal ancestry, and all stated that their maternal grandmothers and great-grandmothers belong to their respective self-identified native groups. The sample size

was composed of 7 Chukchi from villages of Ust-Belaya (4), Kanchalan (2), and Uelen (1), and 6 Siberian Eskimos from villages the Novoye Chaplino (5), and Lorino (1). The stated ancestry of individuals was taken at face value, and sample sizes were very small. Yet, the data received from these individuals are being used uncritically elsewhere (Bonato and Salzano, 1997) as representatives of their ethnic groups, and conclusions were drawn as if applicable to the whole "Chukchi" ethnic group.

Another article by Shields *et al* (1994) contains the following description of the sampling procedure from a number of native individuals, including 14 Yukagir: "all donors were interviewed to determine the extent of intertribal admixture; none indicated knowledge [sic] of admixture." However, if before taking genetic samples a proper demographic and genealogical study would have taken place, the authors might have found out that in some Yukagir villages, such as Nelemnoye, no "pure" Yukagirs younger than 20 have been found (Karafet *et al,* 1994), and thus self-identification should be treated cautiously. There is surely a big difference between "knowledge" of admixture admitted to a visiting researcher and the private knowledge which an individual or community may have about family history. Yet, the authors studied correlations between supposed phylogenetic relationships and linguistic classification.

Krylov *et al* (1995) in the study of the HLA allele distribution in Chukchi and Asian Eskimo tested the hypothesis that these populations are closely related to "Orientals" (Chinese and Japanese) and found that the Chukotka native populations are genetically more closely related to Caucasoids and Native Americans than to "Orientals". No account of the sampling procedure was given. Could such conclusions be the result of studying native individuals with heavy Caucasoid admixture?

A good example of the consequences of such an approach is illustrated in the article by Schurr *et al* (1999). They found differences in the haplogroup distribution of the Koryaks of the Kamchatka Peninsula relative to that of the Koryaks from Northeastern Kamchatka, who had haplogroup A and D frequencies comparable to Reindeer Chukchi populations. The reason for this discrepancy was explained by them as being in the source of samples for these populations. The "Koryak" population analysed previously by Sukernik (1981, 1986) consisted of individuals from Middle Pakhachi and Achayvayam villages who were sampled as part of a study of conventional genetic markers in Chukchi populations. Shurr *et al* argues that extensive Chukchi admixture in these villages, if not total replacement of the resident Koryaks resulting from prolonged Chukchi-Koryak wars in the 19th century, probably accounts for the differences in haplogroup composition of Koryak subgroups. Consequently, the Middle Pakhachi - Achayvayam subgroup, originally classified as Reindeer Koryaks by Gurvich, should instead be more properly considered Reindeer Chukchi, as suggested by Bogoras.

Sheremet'eva and Gorshkov (1977) in a study of Koryaks of Kamchatka from different districts, including North Kamchatkan Olyutorski and Penzhinski districts, and Severo-Evenskii district of Magadan region, did not give any account of their sampling procedure or state the possibility of admixture. Yet Solovenchuk *et al* (1985) chose not to study Koryaks from North Kamchatka because in these Northern districts, including Oluytorskii and Penshinskii district, Koryaks live in supposedly mixed Koryak-Chukchi villages, and in Severo-Evenskii district Koryaks live in mixed Chukchi-Koryak-Even villages. However, he notes that his choice of samples still does not exclude hidden admixture, which might seriously impede the interpretation of the results of his study.

Thus, once again, although not undermining the results of both studies, a critical reader would have to question the results and their interpretations.

Novoradovskii *et al* (1991) included only individuals of Buryat nationality in their sample. Nevertheless, there were p^c and ACP1 allele carriers in the sample, which are characteristic of Caucasians. Also, the frequency of the Mongoloid gene ESD^2 was relatively low in all groups. These facts point to the probable Caucasian component in the ethnogenesis of the studied populations, which was not traced genealogically.

There have been, however, a few research undertakings that along with genetics, studied the demography and genealogy of indigenous Siberian populations. Yet they seem to be rather an exception from the rule (Rychkov *et al* 1974a, 1974b, Sheremet'eva and Gorshkov, 1981; Sukernik *et al*, 1977; Osipova, 1994). Posuh *et al* (1990) studied the demographic structure of three contemporary Even populations of Yakutia. They found that these Even populations are growing mainly because of outside migration and assimilation.

This brief overview has shown that demographic, and especially extensive genealogical studies are generally not an integral part of genetic studies, and that this may lead to obscured results, or their incorrect interpretation.

Perhaps one of the most interesting articles that could assist us in illustrating the importance of genealogical data in genetic studies is that of ethnographers Kuznetsov and Missonova (1990) who looked at admixture and ethnic self-consciousness among the indigenous populations of Kamchatka and Chukotka. The authors analyze 76 genealogies of the Chukchi, the Koryak, and the Even recorded in Oklan (Kamchatka) and Keperveem (Chukotka). Their study gives a very detailed account of the number of

individuals of all nationalities living in the village, the number of marriages and nationalities of spouses, the number of children and their paternity, number of children from mixed marriages, per cent of admixture, and migration pattern. The study showed a high (41.4%) European element in the gene pool, very high (69.3%) among children, and forecasts a further increase. Particularly, for combined groups of native people in both villages, out of 198 men only 85 were found to be "pure", 24 were Mongoloid hybrids (i.e. between different native groups), and 89 were Mongolid-Caucasoid hybrids. Out of 228 women 110 were "pure", 30 Mongoloid hybrids, and 88 Mongoloid-Caucasoid hybrids. Out of 205 children, only 42 were "pure", 21 were Mongoloid hybrids, and 142 were Mongoloid-Caucasoid hybrids. However, quite apart from the doubtful concept of "purity", this has little effect on ethnic self-identity, since children tend to adopt their mother's ethnicity. Thus, 1-5% admixture in indigenous populations stated in many articles should be treated with extreme caution in the light of this article and other combined above.

The importance of sampling procedure becomes evident when we consider the purposes of research projects in regards to indigenous Siberian populations, as well as subsequent analytical steps in the analysis of genetic data. Indeed, there are three steps in the course of genetic study: (1) a sample from a target population(s) is identified and biological samples are taken from individuals who are judged to represent our population, (2) laboratory analyses are performed, genetic distances are derived, and the graphic representation of genetic distances is produced in a form of a dendrogram, principal components plot, and finally (3) genetic distances are interpreted. As we see, this edifice

is supported by its foundation, a sample from a studied population. If the sample is not chosen carefully, all subsequent steps may produce inaccurate conclusions.

Of these three steps, it seems to me that each one stands at a different level of sophistication. The second step is constantly updated, as could be seen from the array of methods for calculating genetic distances, or development of more and more sophisticated laboratory methods and analysis.

The final stage of the genetic distance analysis, the interpretation, has recently experienced a shift in paradigm, with new theories coming to forth. This has been discussed in the works of Relethford (1994, 1995), Templeton (1993, 1996, 1997), Felsenstein (1982) and others, who argue that contrary to popular belief, genetic data gives us little direct information about phylogeny, because while genetic distances are assumed to reflect primarily a branching process, in reality genetic distances reflect overall dissimilarity resulting from a variety of causes. A history of bifurcational splits in a dendrogram is one, but not the only explanation. The same patterns could be produced by an appropriate matrix of migration rates among populations. Felsenstein (1982) noted a basic problem in interpreting genetic distances, that the same distances can be produced by a branching model or by a migration matrix model [17]. So, two completely different models can produce exactly the same genetic distances (Fig.18). Relethford concludes that rather than phylogenetic relationships, genetic data reflect the demographic history of our species. Thus, new models allow researchers to test a variety of hypotheses by developing comparative matrices. Distance matrix comparison includes migration matrix methods, which provide estimates of genetic distance under given demographic

[17] In a migration model all populations are of equal size and have exchanged migrants according to the migration matrix (Relethford, 1995).

scenarios, as opposed to assuming that all populations have equal size or that long-term migration rates are the same across all populations. These estimates can be compared with observed genetic distance matrices to test hypotheses regarding patterns of gene flow, the effects of population size, and other potential influences (Relethford, 1996).

Developing new models not only helps researchers to interpret results differently, but also stresses the importance of collecting demographic and population history data while conducting a genetic study. The necessity to combine genetic and demographic research has also been expressed in other sources (Wilbert and Layrisse 1980).

However, the first and very important step, the sampling procedure, has not change significantly over the years. It seems reasonable to call for an increased awareness of the problem of ethnic self-identity and its implication for the sampling procedure**.** Here, I would like to propose a new method of sampling where sample for genetic studies, especially those focusing on phylogeny, should be drawn from carefully selected individuals that social anthropologists identify as being unadmixed on the basis of genealogical data with a reasonable degree of certainty for the most recent 3-4 generations. This research assumes a field study where social anthropologists, using the "participant observation" method, study the social background, life and family histories, and concept of ethnic identity, thus collecting information that emerges slowly and is not disclosed readily to a total stranger. Ideally, a wide range of genetic systems should be chosen (nDNA, Y chromosome, mtDNA, protein polymorphisms) for a simultaneous study in target populations, since different markers have different selective and informational value. This combined genetic and social anthropological approach, where the study and understanding of genetic variation and population structure is

complimented by detailed accounts of the genealogy and demography of the community, will yield a more comprehensive and realistic picture of a populations genetic affinities.

APPENDIX

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Table 1 Polymorphic genetic markers in Northeast Siberian populations

In plasma-

Immunoglobulins (IgG, IgM, IgA, IgD IgE) and their allotypes: Gm and Inv, Haptoglobin (Hp), Transferrin (Tf), Gc protein, Beta Lipoprotein allotypes: the Ag and Lp systems, Pseudocholinesterase (E_1 and E_2 loci), Alkaline Phosphatase (p^0, p^+, p^{++}), Albumin, Ceruloplasmin, a_1-Antitripsin, Xm system, $Alpha_1$-Acid Glycoprotein (types I, II, III).

In blood cells-

The red cell antigens: blood groups

loci ABO (alleles A_1, A_2, B, and 0), P (P and p), Duffy (Fy_a and Fy_b), Diego (Di^a and Di^b), Kell (K and k), MNSs (MS, Ms, NS, Ns), Rh (haplotypes R^0 +r, R^1 + R', R^2 + R'', R^z + r^y), Kidd (Jk^a, Jk^b), Lewis (Le and le)

Enzymes Hemoglobin (Hb), Acid Phospatase (AcP), Glucose-6-Phosphate dehydrogenase (G6PD), 6 Phosphogluconate Dehydrogenase (6PGD), Phosphoglucomutase (PGM), Adenylate Kinase (AK)

Enzymes whose electrophoretic variants are either very infrequent or too difficult to differentiate with certainty:

Lactate dehydrogenase (LDH), Malate dehydrogenate (MDH), red cell Esterase (B, P, C, A), Peptidase (A-E), Glutathione reductase (NAD(P) H_2, Methemoglobin reductase (NADH and NADPH Diaphorases), Catalase, Galactose-1-Phosphate Uridyl Transferase (adapted from Giblett, 1969).

Table 2
The phenotype and allele frequencies in the two Coastal Chukchi populations

(from Rychkov and Sheremet'eva, 1972)

Phenotypes \ Populations	Nunyamo		Sireniki	
	N_o	Allele frequencies	N_o	Allele frequencies
O	19	$O = 0,6275$	25	$O = 0,7293$
A_1	15	$A_1 = 0,2083$	14	$A_1 = 0,1716$
A_2	1	$A_2 = 0,0162$	0	$A_2 = 0,0000$
B	10	$B = 0,1480$	8	$B = 0,0990$
A_1B	3		1	
A_2B	0		0	
Σ	48		48	
$MS.MS$	7	$MS = 0,2202$	1	$MS = 0,1515$
$MS.Ms$	2	$Ms = 0,4131$	3	$Ms = 0,2121$
$Ms.Ms$	8	$NS = 0,1688$	2	$NS = 0,1515$
$MS.NS$	3	$Ns = 0,1979$	2	$Ns = 0,4848$
$MS.Ns$	6		1	
$Ms.Ns$	14		9	
$NS.NS$	3		4	
$NS.Ns$	1		2	
$Ns.Ns$	1		9	
Σ	45		33	
$DCCEE$	0	$CDE = 0,0491$	0	$CDE = 0,0859$
$DCCEe$	9	$C^WDE = 0,0595$	6	$C^WDE = 0,0532$
DCC^WEe	0	$CDe = 0,4188$	0	$CDe = 0,3887$
$DCCee$	7	$C^WDe = 0,0917$	6	$C^WDe = 0,0786$
DCC^Wee	1	$cDE = 0,3199$	1	$cDE = 0,1374$
$DCcEE$	2	$cDe = 0,0610$	2	$cDe = 0,2562$
$DCcEe$	7		11	
DC^WcEe	4		4	
$DCcee$	5		14	
$DccEE$	5		0	
$DccEe$	2		1	
$Dccee$	0		2	
Σ	42		47	
Hp 1—1	0	$Hp^1 = 0,2222$	2	$Hp^1 = 0,3749$
Hp 2—1	8	$Hp^2 = 0,7778$	14	$Hp^2 = 0,6249$
Hp 2—2	10		8	
Σ	18		24	
Tf^C	16	$Tf^c = 0,9706$	20	$Tf^c = 1,0000$
$Tf^{CD_{chi}}$	1	$Tf^{1-c} = 0,0294$	0	$Tf^{1-c} = 0,0000$
Σ	17		20	
Gc 1—1	6	$Gc^1 = 0,7500$	9	$Gc^1 = 0,7500$
Gc 2—1	6	$Gc^2 = 0,2500$	6	$Gc^2 = 0,2500$
Gc 2—2	0		1	
Σ	12		16	
Le $(a—b+)$	37	$Le^a = 0,0000$	34	$Le^a = 0,0000$
Le $(a—b—)$	8		5	
Le $(a+b—)$	0		0	
Σ	45		39	
$P+$	21	$P_1 = 0,2562$	13	$P_1 = 0,2072$
$P—$	26	$P_2 = 0,7438$	22	$P_2 = 0,7928$
Σ	47		35	
$K+$	5	$K = 0,0559$	1	$K = 0,0163$
$k—$	41	$k = 0,9441$	30	$k = 0,9837$
Σ	46		31	
Di $(a+)$	9	$Di^a = 0,1031$	3	$Di^a = 0,0781$
Di $(a—)$	37	$Di^b = 0,8969$	17	$Di^b = 0,9219$
Σ	46		20	
Fy $(a+)$	22	$Fy^a = 0,2777$	13	$Fy^a = 0,2572$
Fy $(a—)$	24	$Fy^b = 0,7223$	16	$Fy^b = 0,7428$
Σ	46		29	

Table 3

The distribution of phenotype and allele frequencies in three Asian Eskimo populations

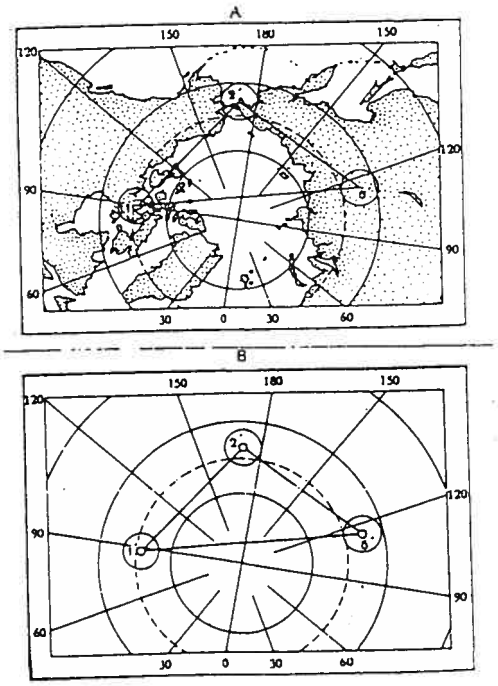
(from Rychkov and Sheremet'eva, 1972)

Populations / Phenotypes	Naukan		Chaplino		Sireniki	
	N_o	allele frequencies	N_o	allele frequencies	N_o	allele frequencies
O	11		24		24	
A_1	7		8		12	
A_2	2	$O=0,6472$	0	$O=0,7296$	2	$O=0,6681$
B	2	$A_1=0,2356$	13	$A_1=0,1036$	13	$A_1=0,1445$
A_1B	1	$A_2=0,0586$	1	$A_2=0,0000$	4	$A_2=0,0271$
A_2B	0	$B=0,0586$	0	$B=0,1667$	0	$B=0,1603$
Σ	23		46		55	
$MS.MS$	3		0		0	
$MS.Ms$	2		4		2	
$Ms.Ms$	3	$MS=0,2334$	13	$MS=0,0776$	4	$MS=0,0637$
$MS.NS$	1	$Ms=0,4096$	0	$Ms=0,5146$	1	$Ms=0,3151$
$MNSs$	1	$NS=0,0285$	4	$NS=0,0672$	4	$NS=0,1787$
$Ms.Ns$	9	$Ns=0,3285$	7	$Ns=0,3406$	8	$Ns=0,4425$
$NS.NS$	0		0		1	
$NS.Ns$	0		3		6	
$Ns.Ns$	2		7		7	
Σ	21		38		33	
$DCCEE$	1		0		0	
$DCCEe$	3		2		6	
DCC^WEe	0		1		0	
$DCCee$	2	$CDE=0,1274$	4	$CDE=0,0630$	1	$CDE=0,1793$
DCC^Wee	1	$C^WDE=0,0476$	0	$C^WDE=0,0532$	1	$C^WDE=0,0100$
$DCcEE$	1	$CDe=0,4370$	5	$CDe=0,2687$	10	$CDe=0,3007$
$DCcEe$	7	$C^WDe=0,0812$	15	$C^WDe=0,0406$	18	$C^WDe=0,0100$
DC^WcEe	1	$cDE=0,1803$	4	$cDE=0,4583$	0	$cDE=0,3506$
$DCcee$	4	$cDe=0,1265$	2	$cDe=0,1162$	6	$cDe=0,1494$
$DccEE$	0		8		4	
$DccEe$	1		6		2	
$Dccee$	0		0		2	
Σ	21		47		50	
Hp 1—1	2		4		3	
Hp 2—1	7	$Hp^1=0,4583$	15	$Hp^1=0,3485$	13	$Hp^1=0,3800$
Hp 2—2	3	$Hp^2=0,5417$	14	$Hp^2=0,6515$	9	$Hp^2=0,6200$
Σ	12		33		25	
TfC	10		33		22	
$TfCD_{chi}$	1	$Tf^c=0,9545$ $Tf^{1-c}=0,0455$	0	$Tf^c=1,0000$ $Tf^{1-c}=0,0000$	1	$Tf^c=0,9783$ $Tf^{1-c}=0,0217$
Σ	11		33		23	
Gc 1—1	3		4		3	
Gc 2—1	6	$Gc^1=0,6667$	11	$Gc^1=0,5588$	9	$Gc^1=0,6250$
Gc 2—2	0	$Gc^2=0,3333$	2	$Gc^2=0,4411$	0	$Gc^2=0,3750$
Σ	9		17		12	
$Le\ (a-b+)$	16		35		35	
$Le\ (a-b-)$	5	$Le^a=0,0000$	5	$Le^a=0,0000$	3	$Le^a=0,0000$
$Le\ (a+b-)$	0		0		0	
Σ	21		40		38	
$P+$	9	$P_1=0,2584$	17	$P_1=0,2122$	9	$P_1=0,1472$
$P-$	11	$P_2=0,7416$	28	$P_2=0,7878$	24	$P_2=0,8528$
Σ	20		45		33	
$K+$	3	$K=0,1190$	1	$K=0,0148$	1	$K=0,0163$
$K-$	18	$k=0,8810$	33	$k=0,9852$	30	$k=0,9837$
Σ	21		34		31	
$Di\ (a+)$	4	$Di^a=0,1056$	3	$Di^a=0,0872$	3	$Di^a=0,0986$
$Di\ (a-)$	16	$Di^b=0,8944$	15	$Di^b=0,9128$	13	$Di^b=0,9014$
Σ	20		18		16	
$Fy\ (a+)$	5	$Fy^a=0,1416$			15	$Fy^a=0,2220$
$Fy\ (a-)$	14	$Fy^b=0,8584$			23	$Fy^b=0,7780$
Σ	19				38	

Table 4

The mean square root $(\overline{\Delta v})^2$ of the differences in the gene frequencies of the studied populations and the genetic distances between them (d ij)

i \ j		1	2	3	4	5	$(\overline{\Delta v})^2$
1	American Eskimo	—	.0122	.0279	.0163	.0275	
2	Asian Eskimo	.1105		—	—	.0117	
3	Coastal Chukchi	.1673	—		—	.0148	
4	Chukotka populations	.1277	—	—		.0097	
5	Siberia (without Chukotka)	.1658	.1095	.1218	.0987	—	
				d_{ij}			



Geographic location (A) and genetic distances (B) of Asian Eskimo (2) in relation to Siberian (5) and American and Greenland Eskimos (1)

(from Rychkov and Sheremet'eva, 1972)

TABLE 5. Allele frequencies for eight blood group systems observed in ten Reindeer Chukchi and one Siberian Eskimo populations

Blood system	Allele	Reindeer Chukchi A	B	C	D	E	F	G	H	J	K	Total Chukchi	Siberian Eskimos
ABO	*O*	0.593	0.573	0.725	0.643	0.736	0.681	0.695	0.664	0.690	0.685	0.656	0.672
	A_1	0.206	0.202	0.129	0.224	0.126	0.210	0.245	0.227	0.215	0.182	0.195	0.143
	B	0.201	0.225	0.146	0.133	0.138	0.109	0.060	0.109	0.095	0.133	0.149	0.184
MNSs	*MS*	0.019	0.036	0.005	0.047	0.085	0.029	0.089	0.177	0.085	0.047	0.051	0.088
	Ms	0.404	0.348	0.389	0.306	0.381	0.361	0.182	0.355	0.431	0.412	0.367	0.456
	NS	0.0	0.0	0.0	0.007	0.009	0.022	0.010	0.0	0.0	0.0	0.003	0.010
	Ns	0.577	0.616	0.606	0.640	0.526	0.588	0.719	0.528	0.484	0.541	0.579	0.446
Kell	*K*	1.000	1.000	1.000	1.000	1.000	1.000	1.000	1.000	1.000	1.000	1.000	1.000
P	P^1	0.389	0.416	0.530	0.382	0.336	0.531	0.488	0.436	0.426	n.t.	0.422	0.204
Rh	R^1	0.703	0.699	0.726	0.755	0.662	0.780	0.738	0.696	0.702	0.688	0.710	0.436
	R^2	0.159	0.214	0.212	0.206	0.319	0.220	0.205	0.201	0.245	0.247	0.216	0.549
	$R^0 + r$	0.138	0.087	0.062	0.039	0.019	0.0	0.057	0.103	0.053	0.065	0.074	0.015
Duffy	Fy^a	0.965	0.953	0.889	0.907	0.990	0.941	0.934	0.949	0.920	n.t.	0.944	0.916
Kidd	Jk^a	0.514	0.536	0.470	n.t.	n.t.	n.t.	0.601	0.685	n.t.	n.t.	0.540	n.t.
Diego	Di^a	0.024	0.048	0.039	0.050	0.025	0.017	0.017	0.048	0.005	0.012	0.030	0.020

. Allele frequencies for transferrins (Tf), haptoglobins (Hp), phosphoglucomutase 1 (PGM_1), adenylate kinase (AK), 6-phosphogluconate dehydrogenase (PGD), and acid phosphatase (AcP) in ten Reindeer Chukchi and one Siberian Eskimo populations

Locus	Allele	Reindeer Chukchi A	B	C	D	E	F	G	H	J	K	Total Chukchi	Siberian Eskimos
Tf	Tf^C	1.0	1.0	1.0	1.0	1.0	1.0	1.0	1.0	1.0	1.0	1.0	1.0
Hp[*]	Hp^1	0.295	0.232	0.243	0.287	0.227	0.307	0.242	0.310	0.263	0.345	0.275	0.242
PGM_1	PGM^1	0.953	0.908	0.921	0.946	0.843	0.881	0.885	0.886	0.953	1.0	0.921	0.941
AK	AK^1	1.0	1.0	1.0	0.985	1.0	1.0	1.0	1.0	1.0	1.0	0.999	1.0
6-PGD	PGD^A	0.962	0.952	0.930	0.917	0.936	0.932	0.959	0.950	0.958	0.913	0.944	0.946
AcP	p^a	0.536	0.585	0.599	0.559	0.534	0.610	0.598	0.614	0.558	0.560	0.570	0.696
	p^b	0.464	0.415	0.401	0.441	0.466	0.390	0.402	0.381	0.442	0.440	0.430	0.304
	p^c	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.005	0.0	0.0	0.001	0.0

[*] Individuals with a haptoglobin 0-0 phenotype were excluded from calculation of allelic frequencies.

(from Sukernik, 1981)

Table 6

Gene frequencies in three populations of Reindeer Chukchi (from Solovenchuk et al, 1982)

Polymorphic systems	Alleles	Subpopulations		
		A	B	C
AcP	A	0,592	0,546	0,610
	B	0,408	0,454	0,390
PGM_1	1	0,867	0,882	0,907
	2	0,133	0,118	0,093
PGD	A	0,962	0,905	0,955
	C	0,038	0,096	0,045
GPT	1	0,627	0,542	0,521
	2	0,373	0,458	0,479
GLO-1	1	0,186	–	0,244
	2	0,814	–	0,756
EsD	1	0,887	–	0,890
	2	0,113	–	0,109
AB0	r(0)	0,674	0,636	0,726
	P(A)	0,175	0,241	0,173
	q(B)	0,151	0,123	0,103
Hp	1	0,314	0,257	0,243
	2	0,686	0,743	0,756
Gc	1	0,749	0,866	0,833
	2	0,251	0,134	0,167
PTC	r(t)	0,488	0,458	0,505
	$P(T_1)$	0,450	0,486	0,490
	$q(T_2)$	0,061	0,066	0,040

Table 7

Genetic distance (d) matrix for Eskimo and other Siberian and circumpolar populations

Populations	Alaskan Eskimos	Canadian Eskimos	Chuckotka Chuckchi	St. Lawrence Island Eskimos	Greenland Eskimos	Evenks of Krasnojarski region	Lapps of Sweden and Finland	American Indians
Chuckotka Eskimos	0.0445315	0.1045941	0.1060560	0.13232121	0.19310328	0.20531	0.2569947	0.4083213

(from Nazarova, 1989)

Table 8

Genotype distances between Eskimo and Chukchi populations
(from Solovenchuk, 1984)

Asian Eskimo-Coastal Chukchi	Coastal Chukchi-Reindeer Chukchi	Asian Eskimo-Reindeer Chukchi	Asian Eskimo-Alaskan Eskimo	Coastal Chukchi-Alaskan Eskimo	Reindeer Chukchi-Alaskan Eskimo
PGM_1 **	E_2 ***	PGM_1 *	PGD ***	PGM_1 **	PGM_1 **
GPT ***	Gc ***	GPT ***	GPT ***	PGD ***	PGD ***
GLO-1 ***		GLO-1 **	Gc **	GLO-1 ***	GLO-1 *
E_2 **		Gc ***	PTC ***	Gc ***	Gc ***
PTC ***		PTC ***		E_2 ***	
		AB0 ***		AB0 *	
ΣI 96,521 ***	68,254 ***	128,911 ***	97,483 ***	109,287 ***	143,152 ***

* $P<0.05$; ** $P<0.01$; *** $P<0.001$.

Table 9

Within-Group Mean Pairwise Sequence Differences for the First 360 Nucleotides of the mtDNA Control Region Observed in Five Circumarctic Populations

Group	No. of Individuals	No. of Lineages	Mean	SD
Chukchi	7	6	2.57	1.29
Athapaskans	21	12	2.47	1.63
West Greenland Eskimos	17	9[a]	2.06	1.61
Inupiaqs	5	4	4.00	1.94
Haida	41	10	2.49	3.18

[a] While 10 lineages were observed, lineage 83 was excluded from this analysis, because it was presumed to be present because of admixture.

(from Shields et al, 1993)

Table 10

Percent Frequencies of mtDNA Haplogroups in Aboriginal Siberians

Population	Haplogroup[a]					
	A	B	C	D	Other	N
Eskimos	80.0	...	...	20.0	...	50
Chukchi	37.5	...	16.7	16.7	29.2	24
Koryaks	23.9	...	21.7	8.7	45.6	46
Yukagirs	...	...	59.3	33.3	7.4	27
Evens	...	...	58.1	7.0	34.9	43
Nivkhs	...	...	...	28.1	71.9	57
Udegeys	...	...	19.6	...	80.4	46
Evenks	3.9	...	84.3	9.8	2.0	51
Nganasans	2.0	...	38.8	36.7	22.4	49
Sel'kups	...	...	35.0	...	65.0	20

[a] The haplotypes grouped into haplogroups A, B, C, D, and Other are expressed as a percentage of the total no. of individuals in the tribe who were analyzed.

Table 11

Sequence Divergence and Radiation Time of Aboriginal Siberian and Amerind mtDNA Haplogroups

Haplogroup	n[a]	N[b]	Sequence Divergence (%)	Radiation Time[c] (years)
C:				
Siberian	9	51	.060	15,000–30,000
Amerind	23	61	.096	24,000–48,000
D:				
Siberian	7	21	.040	10,000–20,000
Amerind	16	60	.053	13,250–26,500
C + D:				
Siberian	16	72	.054	13,500–27,000
Amerind	39	121	.075	18,750–37,500
Combined C + D	53	193	.067	16,750–33,500

[a] No. of haplotypes.
[b] No. of subjects.
[c] Estimated using a mtDNA evolution rate of 2%–4%/MYR.

(from Torroni et al, 1993)

Table 12

mtDNA Haplogroups in Siberia and the Northern Pacific Rim

Population (No.)	Frequency in Haplogroup (%)							Reference(s)
	A	B	C	D	G	Y	Other[a]	
Tungusic:								
Evenks (51)	3.9	.0	84.3	9.8	.0	.0	2.0	Torroni et al. (1993*b*)
Udegeys (45)	.0	.0	17.8	.0	8.9	.0	73.3	Torroni et al. (1993*b*)
Linguistic isolate:								
Nivkhs (57)	.0	.0	.0	28.1	5.3	64.9	1.8	Torroni et al. (1993*b*)
Paleoasiatic:								
Itel'men (47)	6.4	.0	14.9	.0	68.1	4.3	6.4	Schurr et al. (in press)
Koryaks (155)	5.2	.0	36.1	1.3	41.9	9.7	5.8	Schurr et al. (in press)
Chukchi (66)	68.2	.0	10.6	12.1	9.1	.0	.0	Present study
Eskimo-Aleut:								
Siberian Eskimos (79)	77.2	.0	2.5	20.3	.0	.0	.0	Present study
St. Lawrence Eskimos (99)	76.0	.0	7.0	14.0	.0	.0	3.3	Merriwether et al. (1995)
Old Harbor Eskimos (115)	61.7	3.5	.0	34.8	.0	.0	.0	Merriwether et al. (1995)
Ouzinkie Eskimos (41)	73.2	.0	4.9	14.6	.0	.0	7.1	Merriwether et al. (1995)
St. Paul Aleuts (72)	25.0	.0	1.4	66.7	.0	.0	6.9	Merriwether et al. (1995)
Na-Dené:								
Haida (38)	92.1	.0	7.9	.0	.0	.0	.0	Ward et al. (1993)
Haida (25)	96.0	.0	.0	4.0	.0	.0	.0	Torroni et al. (1993*a*)
Dogrib (154)	90.9	.0	2.0	.0	.0	.0	7.1	Merriwether et al. (1995)
Dogrib (30)	100.0	.0	.0	.0	.0	.0	.0	Torroni et al. (1992)
Amerind:								
Bella Coola (32)	78.1	6.3	9.4	6.3	.0	.0	.0	Ward et al. (1993)
Bella Coola (25)	60.0	8.0	8.0	20.0	.0	.0	4.0	Torroni et al. (1993*a*)
Nuu-Chah-Nulth (63)	44.4	3.2	19.0	22.2	.0	.0	11.1	Ward et al. (1991)
Nuu-Chah-Nulth (15)	40.0	7.6	13.3	26.7	.0	.0	13.3	Torroni et al. (1993*a*)

[a] Haplotypes that belong to the haplogroups listed but that may have different haplogroup affiliations.

Table 13

Sequence Divergence and Divergence Time of Native Siberian and Native American mtDNA Haplogroups

Haplogroup and Region	No. of		Sequence Divergence (%)	Divergence Time[b] (YBP)
	Haplotypes[a]	Individual mtDNAs/ Haplogroup		
A:				
Siberia	10	119	.028	12,727–9,655
America	46	189	.079	35,909–27,241
B:				
America	30	99	.039	17,727–13,448
C:				
Siberia	14	123	.043	19,545–14,828
America	31	72	.122	55,545–42,069
D:				
Siberia	13	47	.111	50,455–38,276
America	16	62	.057	25,909–19,655
G:				
Siberia	11	106	.024	10,909–8,276
Y:				
Siberia	7	58	.014	6,364–4,828

[a] Data are from Torroni et al. (1992, 1993*a*, 1993*b*, 1994*a*, 1994*b*), Huoponen et al. (1997), Schurr et al. (in press), and present study.

[b] Estimated on the basis of an mtDNA evolutionary rate of 2.2%–2.9%/MYR (Torroni et al. 1994*b*).

(from Starikovskaya et al, 1998)

Table 14 Frequencies of Y chromosome-specific polymorphisms in Native American and Asian populations

Population	*n*	DYS199 "T"	DYS287 (YAP+)	DYS
Native Americans				
Mixe	14	85.7	14.3	–
Mixtecs	10	70.0	–	–
Zapotecs	6	50.0	–	–
Seminoles	25	48.0	–	–
Navajo	9	55.5	–	–
Asians/Siberians				
Siberian Eskimos	34	20.6	–	–
Chukchi	24	16.7	–	–
Koryaks	27	–	–	–
Itelmen	19	–	–	–
Nivkhs	19	–	–	–
Udegeys	20	–	–	–
Evenks	31	–	–	–
Northern Altayans	9	–	–	–
Kets	12	–	–	–
Tibetans	22	–	36.4	–
Koreans	4	–	–	–

(from Lell et a

Table 16

Phenotype and allele frequencies for 15 blood group and protein polymorphic systems in four Koryak populations

(from Solovenchuk et al, 1985)

Locus	Phenotype	Sedanka			Voyampolka			Tymlat			Karaga			Total sample			min — max for Northeast native groups
		Phenotype frequency	Allele	Allele frequency	Phenotype frequency	Allele	Allele frequency	Phenotype frequency	Allele	Allele frequency	Phenotype frequency	Allele	Allele frequency	Phenotype frequency	Allele	Allele frequency	
AcP	AA	0,318	AcPA	0,554	0,350	AcPA	0,0563	0,519	AcPA	0,726	0,336	AcPA	0,584	0,389	AcPA	0,616	0,342–0,397
	BA	0,473			0,425			0,415			0,496	AcPB	0,409	0,454	AcPB	0,383	0,463–0,501
	BB	0,209			0,225			0,066			0,154	AcPC	0,007	0,154	AcPC	0,0015	0,143–0,152
	BC	0,000			0,000			0,000			0,014			0,003			0,000–0,000
	N	239			80			212			143			674			
PGM_1	1	0,926	PGM_1^1	0,961	0,835	PGM_1^1	0,899	0,939	PGM_1^1	0,969	0,888	PGM_1^1	0,944	0,911	PGM_1^1	0,953	0,693–0,783
	2-1	0,069			0,127			0,061			0,112			0,083			0,203–0,267
	2	0,004			0,038			0,000			0,000			0,006			0,007–0,040
	N	231			79			212			143			665			
PGD	AA	0,970	PGDA	0,985	0,961	PGDA	0,980	0,830	PGDA	0,915	0,937	PGDA	0,968	0,917	PGDA	0,959	0,887–0,926
	AC	0,030			0,039			0,170			0,063			0,083			0,074–0,113
	CC	0,000			0,000			0,000			0,000			0,000			0,000–0,000
	N	233			76			212			143			664			
GPT	1	0,338	GPT	0,598	0,437	GPT^1	0,613	0,425	GPT^1	0,656	0,319	GPT^1	0,571	0,371	GPT^1	0,611	0,301–0,456
	2-1	0,521			0,350			0,462			0,503			0,479			0,456–0,538
	2	0,142			0,213			0,113			0,177			0,150			0,088–0,177
	N	240			80			186			141			647			
GLO-1	1	0,053	$GLO\text{-}1^1$	0,253	0,042	$GLO\text{-}1^1$	0,222	0,009	$GLO\text{-}1^1$	0,078	0,007	$GLO\text{-}1^1$	0,087	0,028	$GLO\text{-}1^1$	0,156	0,047–0,113
	2-1	0,400			0,361			0,137			0,161			0,258			0,339–0,451
	2	0,547			0,597			0,850			0,832			0,715			0,435–0,614
	N	225			80			212			143			660			

Table 16 (continued)

Locus	Phenotype	Sedanka			Voyampolka			Tymlat			Karaga			Total sample			min — max for Northeas native populat
		Phenotype frequency	Allele	Allele frequency	Phenotype frequency	Allele	Allele frequency	Phenotype frequency	Allele	Allele frequency	Phenotype frequency	Allele	Allele frequency	Phenotype frequency	Allele	Allele frequency	
EsD	1	0,793	EsD^1	0,884	0,823	EsD^1	0,905	0,877	EsD^1	0,937	0,629	EsD^1	0,783	0,788	EsD^1	0,882	0,764–0,79
	2-1	0,181			0,165			0,118			0,308			0,186			0,190–0,21
	2	0,025			0,013			0,005			0,063			0,025			0,013–0,02
	N	237			79			212			143			671			
MN	M	–	–	–	–	–	–	0,198	N	0,363	0,196	M	0,448	0,197	M	0,397	0,187–0,30
	MN	–			–			0,330			0,503			0,400			0,380–0,49
	N	–			–			0,472			0,301			0,403			0,247–0,36
	N							212			143			355			
P_1	+	–	–	–	–	–	–	0,844	P^1	0,605	0,804	P^1	0,557	0,828	P^1	0,585	0,357–0,81
	–	–			–			0,156			0,196			0,172			0,181–0,64
	N							212			143			355			
Lo	a^-b^-	–	–	–	–	–	–	0,009	Lo	0,095	0,077	Lo	0,277	0,037	Lo	0,192	0,048–0,15
	a^+b^-	–			–			0,000			0,021			0,008			0,000–0,00
	a^-b^+	–			–			0,991			0,902			0,955			0,845–0,95
	N							212			143			355			
ABO	0	0,496	r(0)	0,706	0,325	r(0)	0,597	0,307	r(0)	0,567	0,378	r(0)	0,609	0,391	r(0)	0,620	0,405–0,45
	A	0,300	P(A)	0,186	0,263	P(A)	0,166	0,350	P(A)	0,243	0,217	P(A)	0,162	0,293	P(A)	0,201	0,310–0,40
	B	0,167	q(B)	0,108	0,375	q(B)	0,237	0,269	q(B)	0,190	0,322	q(B)	0,228	0,256	q(B)	0,179	0,123–0,24
	AB	0,038			0,038			0,075			0,083			0,059			0,047–0,07
	N	240			80			212			143			675			

Table 16 (continued)

Locus	Phenotype	Sedanka			Voyampolka			Tymlat			Karaga			Total sample			min -- max for Northeast native groups
		Phenotype frequency	Allele	Allele frequency	Phenotype frequency	Allele	Allele frequency	Phenotype frequency	Allele	Allele frequency	Phenotype frequency	Allele	Allele frequency	Phenotype frequency	Allele	Allele frequency	
Pp	1	0,488			0,500			0,476			0,455			0,479			0,416–0,473
	2	0,504			0,500			0,500			0,545			0,511			0,478–0,528
	3	0,008			0,000			0,023			0,000			0,010			0,045–0,053
		240			80			212			143			675			
E_2	C_3+	0,100	E_2+	0,051	0,075	E_2+	0,038	0,052	E_2+	0,036	0,028	E_2+	0,021	0,067	E_2+	0,039	0,842–0,859
	C_3-	0,900			0,925			0,929			0,958			0,924			0,041–0,158
	var	0,000			0,000			0,019			0,004			0,007			
		240			80			212			143			675			
Hp	1-1	0,013	Hp^1	0,098	0,013	Hp^1	0,188	0,075	Hp^1	0,250	0,056	Hp^1	0,175	0,042	Hp^1	0,173	0,071–0,111
	2-1	0,171			0,350			0,350			0,238			0,262			0,389–0,442
	2-2	0,817			0,637			0,575			0,706			0,696			0,457–0,500
		240			80			212			143						
Gc	1-1	0,743	Gc^1	0,863	0,750	Gc^1	0,863	0,830	Gc^1	0,913	0,818	Gc^1	0,909	0,787	Gc^1	0,888	0,625–0,813
	2-1	0,240			0,225			0,165			0,182			0,202			0,174–0,327
	2-2	0,017			0,025			0,005			0,000			0,010			0,013–0,048
		237			80			212			143			672			
PTC	–	0,130			0,165			–			–			0,138			0,197–0,436
	–	0,870			0,835			–			–			0,862			0,564–0,803
	–	239			79			–			–			318			–

var- rare variant

Table 17 *Correlations of genetic (GENE), geographic (GEOG), and linguistic (LING) distance matrices*

	Correlation (r)	p[1]
(1) Correlations:		
Distances compared		
GENE * GEOG	0.546	0.001
GENE * LING	0.351	0.001
GEOG * LING	0.500	0.001
(2) Partial Correlations[2]:		
Test of relationship		
GENE * GEOG (LING)	0.456	0.001
GENE * LING (GEOG)	0.108	0.152
(3) Multiple Correlations[3]:		
Test of relationship		
GENE * GEOG, LING	0.553	0.001

[1] Mantel test probabilities.
[2] Partial correlations between two matrices influencing the third matrix.
[3] Multiple correlation obtained through multiple regression of genetic distance matrix against both geographic and linguistic distance matrices.

(from Crawford, 1997)

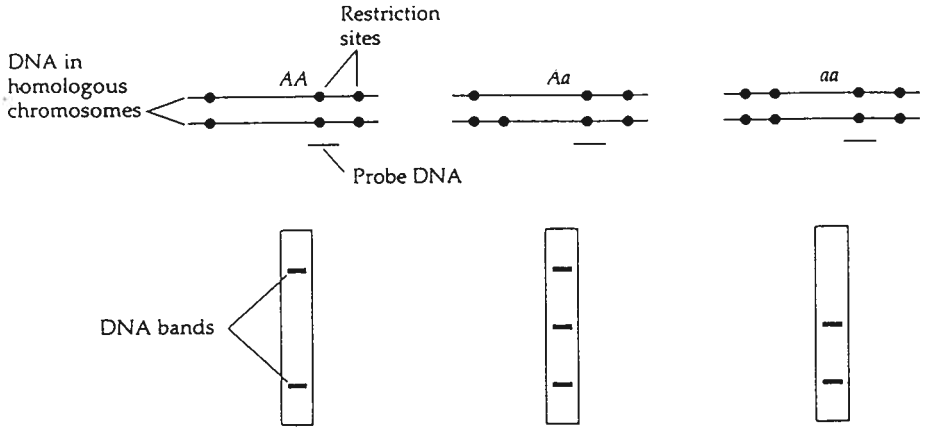


Figure 1. 1 Restriction fragment length polymorphisms (RFLPs) result from the presence or absence of particular restriction sites in DNA. In this example, the DNA molecule designated *A* contains three restriction sites, and the one designated *a* contains four. Genotypes *AA*, *Aa*, and *aa* each yield a different pattern of bands in Southern blot using the indicated probe DNA.

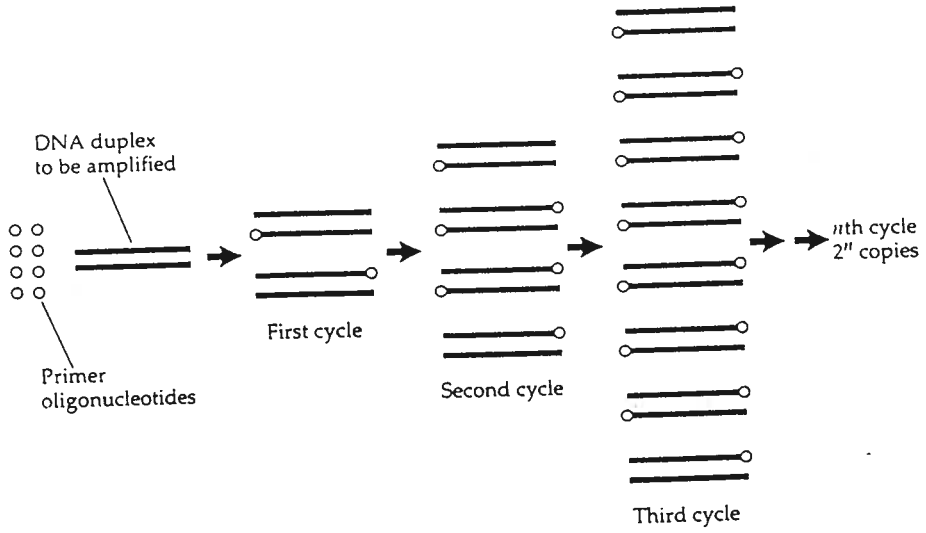


Figure 1. 2 The polymerase chain reaction (PCR). Short primer oligonucleotides are used as primers to initiate DNA replication from opposite ends of a DNA duplex to be amplified. After each round of replication, the DNA is heated to separate the strands and then cooled to allow new primers to anneal. Repeated rounds of replication result in an exponential increase in the number of target molecules. (from Hartl and Clark, 1996)

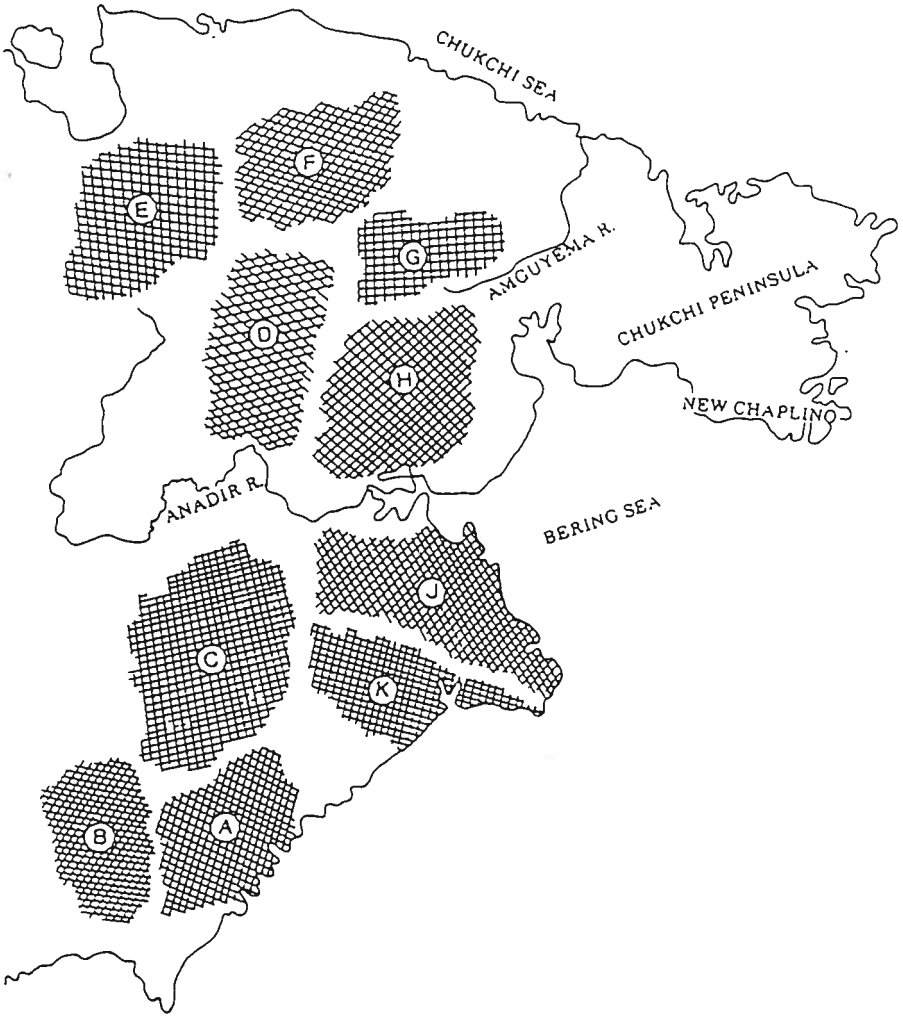


Fig. 2. Schematic designation of territories occupied by roaming subgroups of Reindeer Chukchi (See Table 1 for key to names).

REINDEER CHUKCHI AND SIBERIAN ESKIMOS: GENETIC HETEROGENEITY

Data on 10 Reindeer Chukchi and one Siberian Eskimo populations sampled

Place[1]	Designation of population (Fig. 1)	Type of sample	Approximate size of population	Sample N.	Sample %
Reindeer Chukchi					
Achaivayam	A	Adults and adolescents	370	214	57.8
Middle Pakhachi	B	Adults and adolescents	300	138	46.0
Vayegi	C	Adults and adolescents	240	104	43.3
Ust-Belaya	D	Mostly adolescents	350	102	29.1
Rytkuchi	E	Adults and adolescents	310	102	32.9
Ryrkaipiy	F	Mostly adolescents	250	59	23.6
Amguyema	G	Mostly adolescents	230	61	26.5
Kanchalan	H	Adults and adolescents	320	107	33.4
Alkatwaam	J	Mostly adults	290	94	32.4
Mainypylgino	K	Mostly adults	350	85	24.3
Total			3,010	1,066	35.4
Siberian Eskimos					
New Chaplino		Almost all adults	250	102	40.8

[1] Village or settlement where boarding schools and headquarters of reindeer-breeding state farms are located.

(from Sukernik et al, 1981)

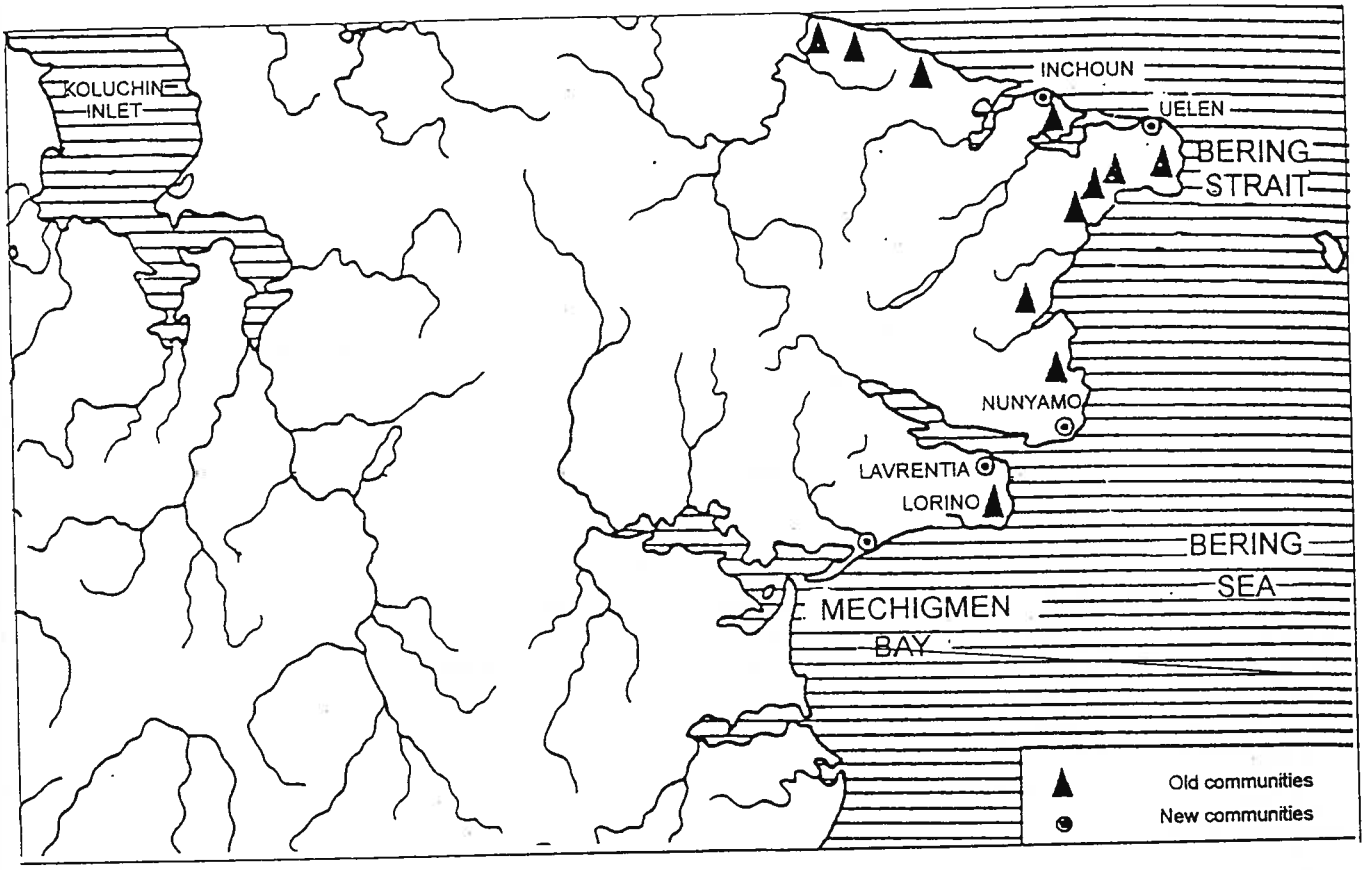


Figure 3 Map showing the location of Coastal Chukchi communities

Subpopulation A: Lorino + Nunyamo Villages
Subpopulation B: Neshkan + Enurmino Villages
Subpopulation C: Uelen + Inchoun Villages

Solovenchuk et al, 1982

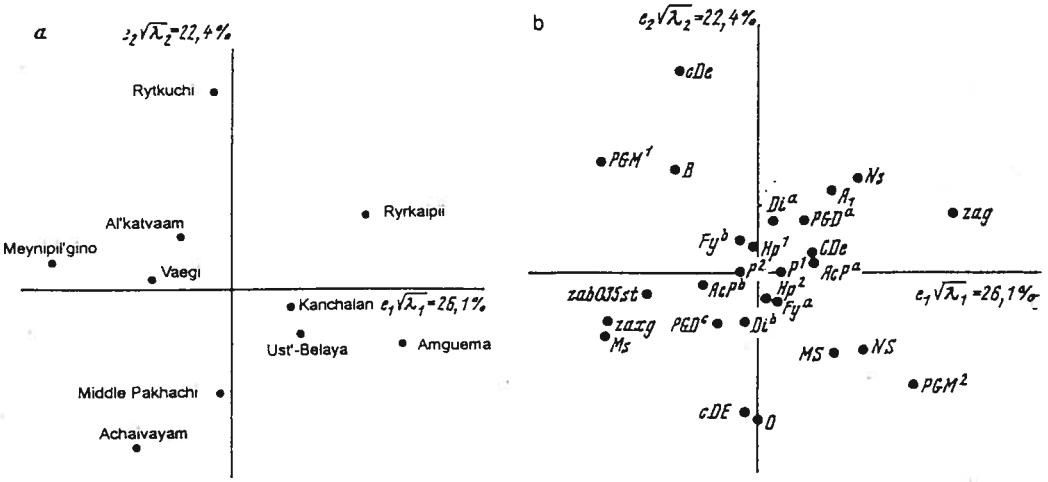


Figure 4 Genetic structure of the 10 Reindeer Chukchi populations
a- location of the populations on the "genetic' map
b- distribution of the alleles plotted along the first two scaled eigenvectors
(from Sukernik, 1986)

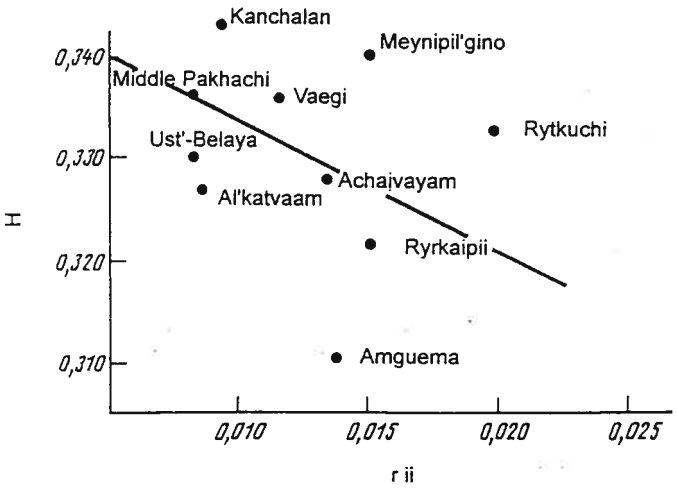


Figure 5 Plot of mean per locus heterozygosity (H) against relative distance from the centroid of the distribution (r ii) for 10 Chukchi populations

(from Sukernik, 1986)

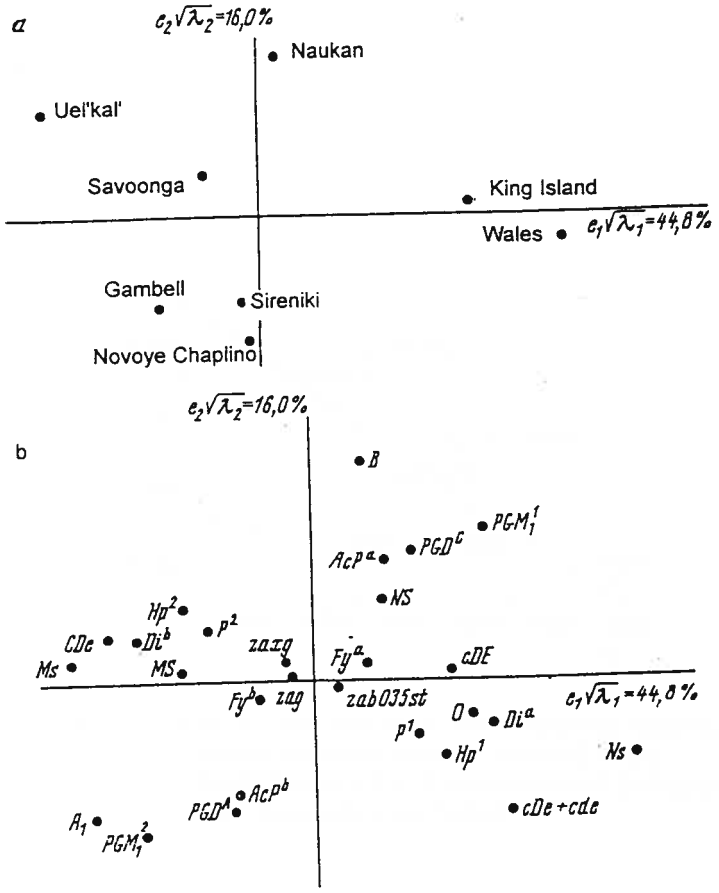


Figure 6 Genetic structure of the Beringian Eskimos

a- location of the populations on the "genetic" map

b- distribution of the alleles plotted along the first two scaled eigenvectors

(from Sukernik et al, 1986b)

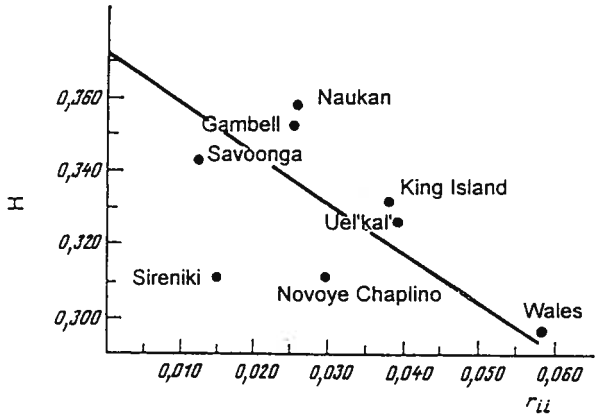


Figure 7 Plot of mean per locus heterozygosity against relative distance from the centroid of the distribution (r ii) for 8 populations of Beringian Eskimo (from Sukernik et al, 1986b)

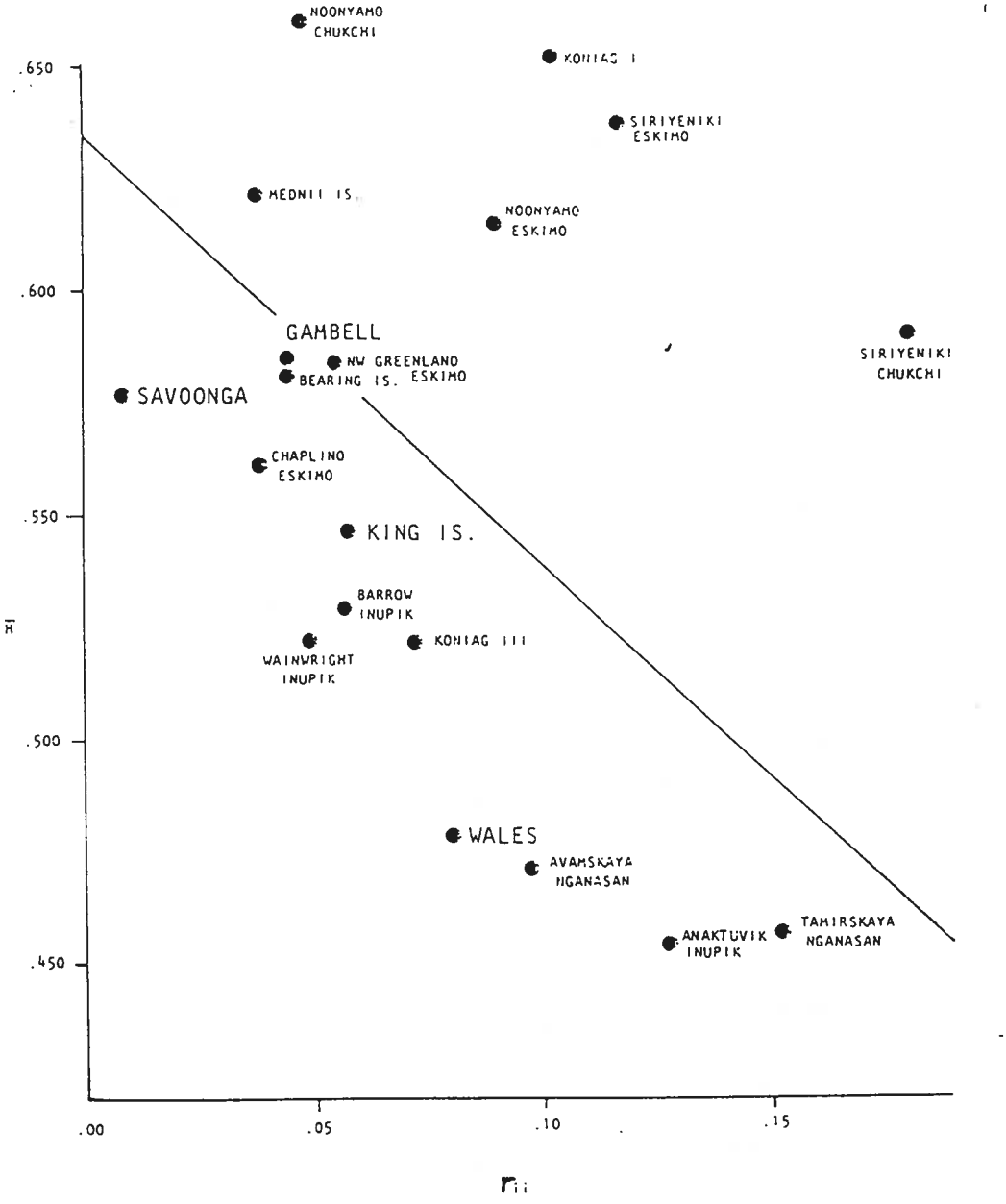


Fig. 8. Plot of mean per locus heterozygosity against relative distance from the centroid of the distribution (r_{ij}) for 19 circumpolar populations. Regression line indicates predicted heterozygosity and is a linear regression on the sample data.

(from Crawford, 1981)

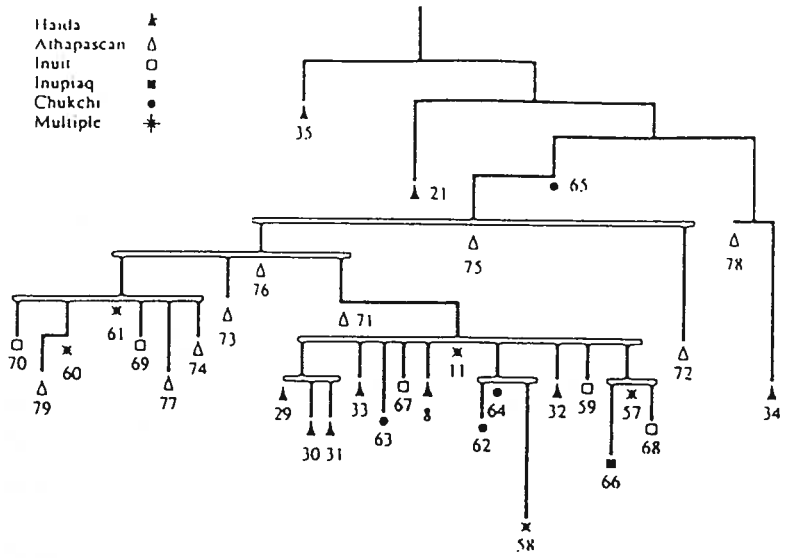


Figure 9. Maximum-likelihood tree of Circumarctic populations. Unshaded bars represent multifurcations in which branching orders cannot be resolved. Lineages identified by an asterisk (*) refer to Circumarctic lineages which are also found in other New World groups (see table 3). Lineage 57 is shared with another Inupiaq, two West Greenland Eskimos, and a Siberian Eskimo. Lineage 58 is shared with another Inupiaq and a West Greenland Eskimo. Lineage 60 is shared with three other Athapaskans, two Inupiaqs, and a West Greenland Eskimo. Lineage 61 is shared with two other Athapaskans, seven West Greenland Eskimos, two Siberian Eskimos, and two Chukchi. Lineage 11 (originally described in a Nuu-Chah-Nulth) is shared with 4 other Nuu-Chah-Nulth, 3 Bella Coola, 1 Yakima, 20 Haida, 6 Athapaskans, and 1 Chukchi.

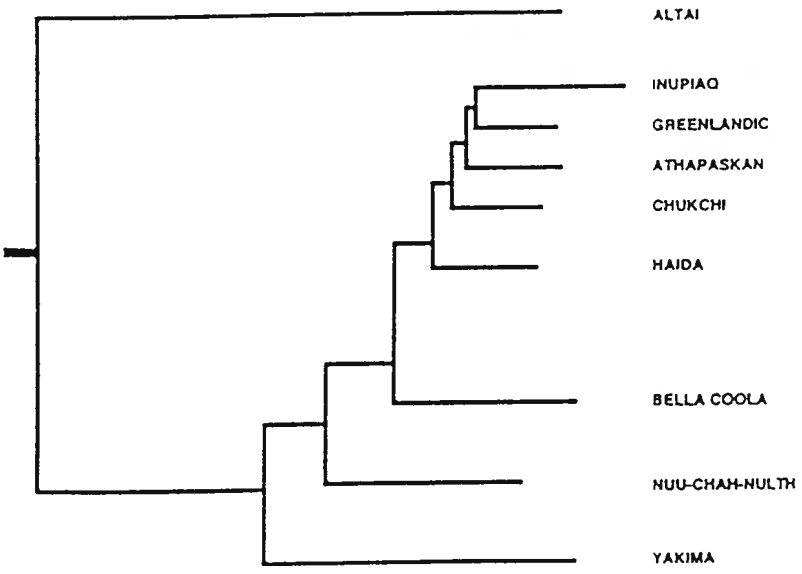


Figure 10. Inferred family tree based on least-squares analysis of the mean pairwise sequence differences between mtDNAs of populations (Felsenstein 1991). "Greenlandic" refers to the West Greenland Eskimo. Siberian Eskimos are not included because of their small sample size

(from Shields et al, 1993)

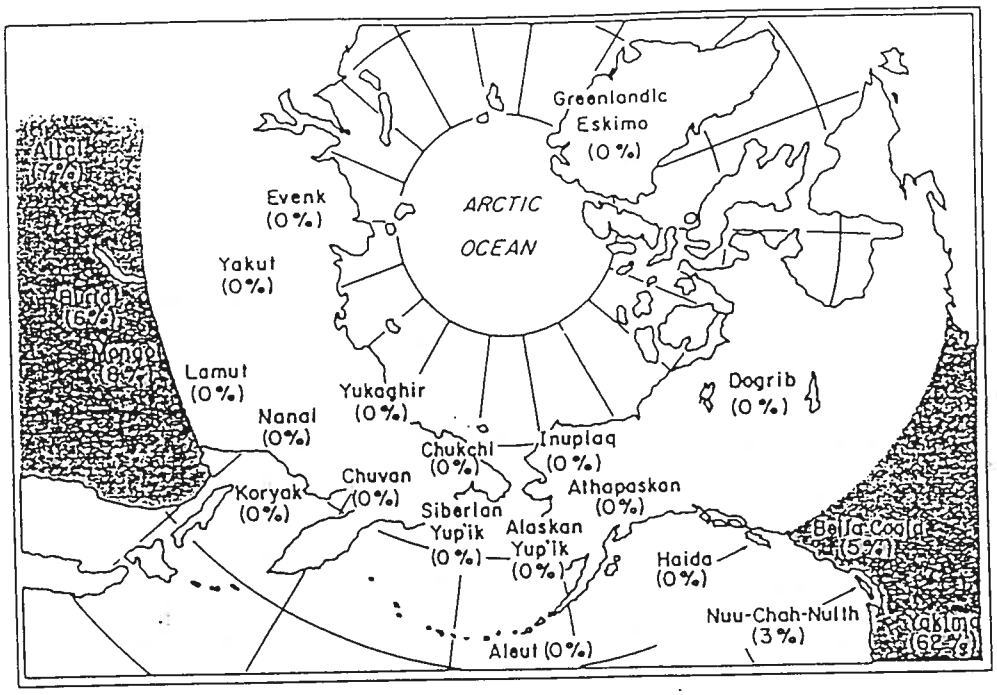


Figure 11 Percentage distribution of the region V 9-bp deletion in Circumarctic populations. Sources for data are as follows: Dogrib (Torroni et al. 1992); Nuu-Chah-Nulth (Ward et al. 1991); Haida and Bella Coola (Ward et al., in press); Mongols (Sambuughin et al. 1991); and Altai, Nanai, Lamut, Koryak, Yukaghir, Chukchi, Chuvan, Siberian Yup'ik, Inupiaqs, Alaskan Yup'ik, Aleut, and Athapaskans (Shields et al. 1992). Shading indicates geographic regions where the region V 9-bp deletion has been observed to occur.

(from Shields et al, 1993)

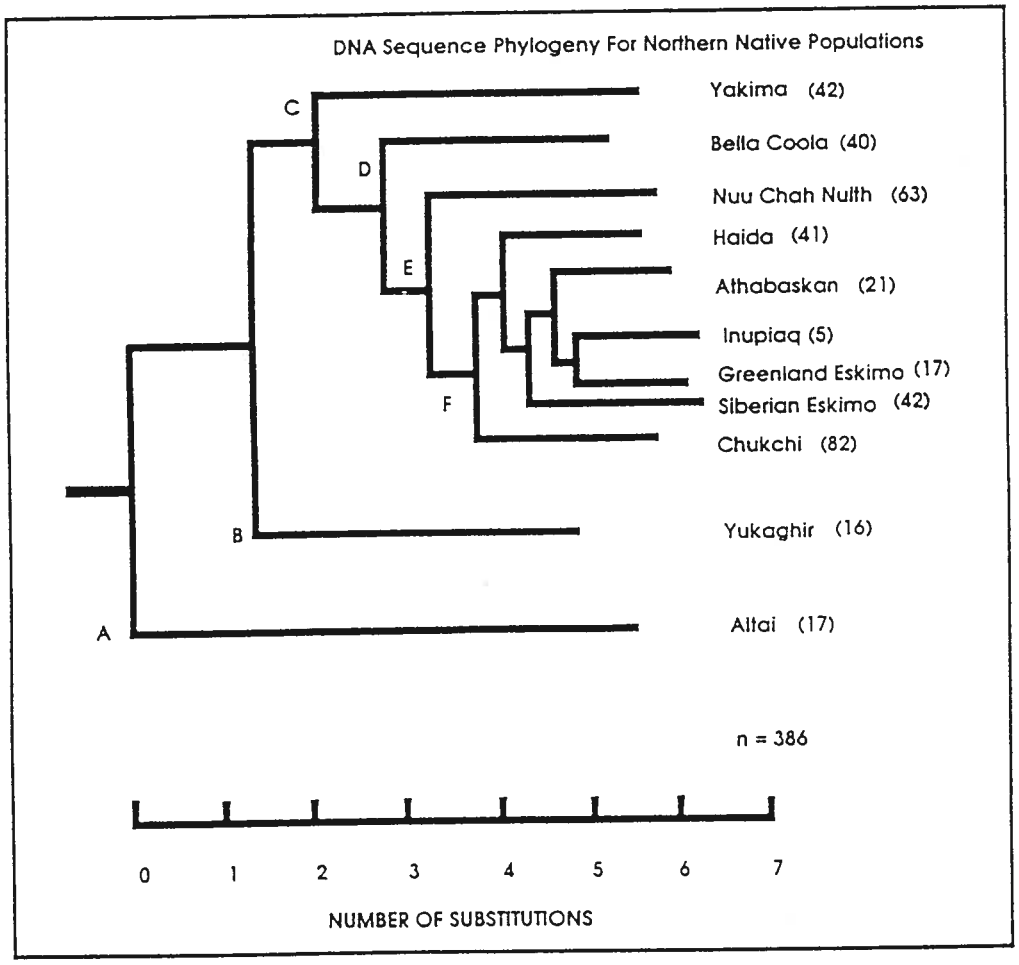


Figure 12 *Inferred family tree based on least-squares analysis of the mean pairwise sequence differences between populations. Numbers in parentheses indicate sample sizes; letters on the tree indicate major cladal nodes.*

(from Shields et al, 1994)

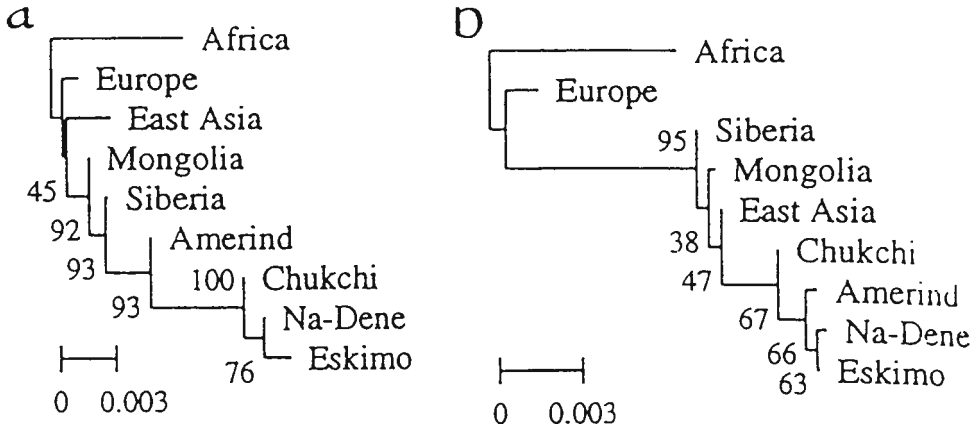


FIG. 13 NJ trees for the populations based on the d_A distance using (*a*) all sequences and (*b*) haplogroup A sequences only. The numbers on the branches are bootstrap values based on 100 replications.

(from Bonato and Salzano, 1997)

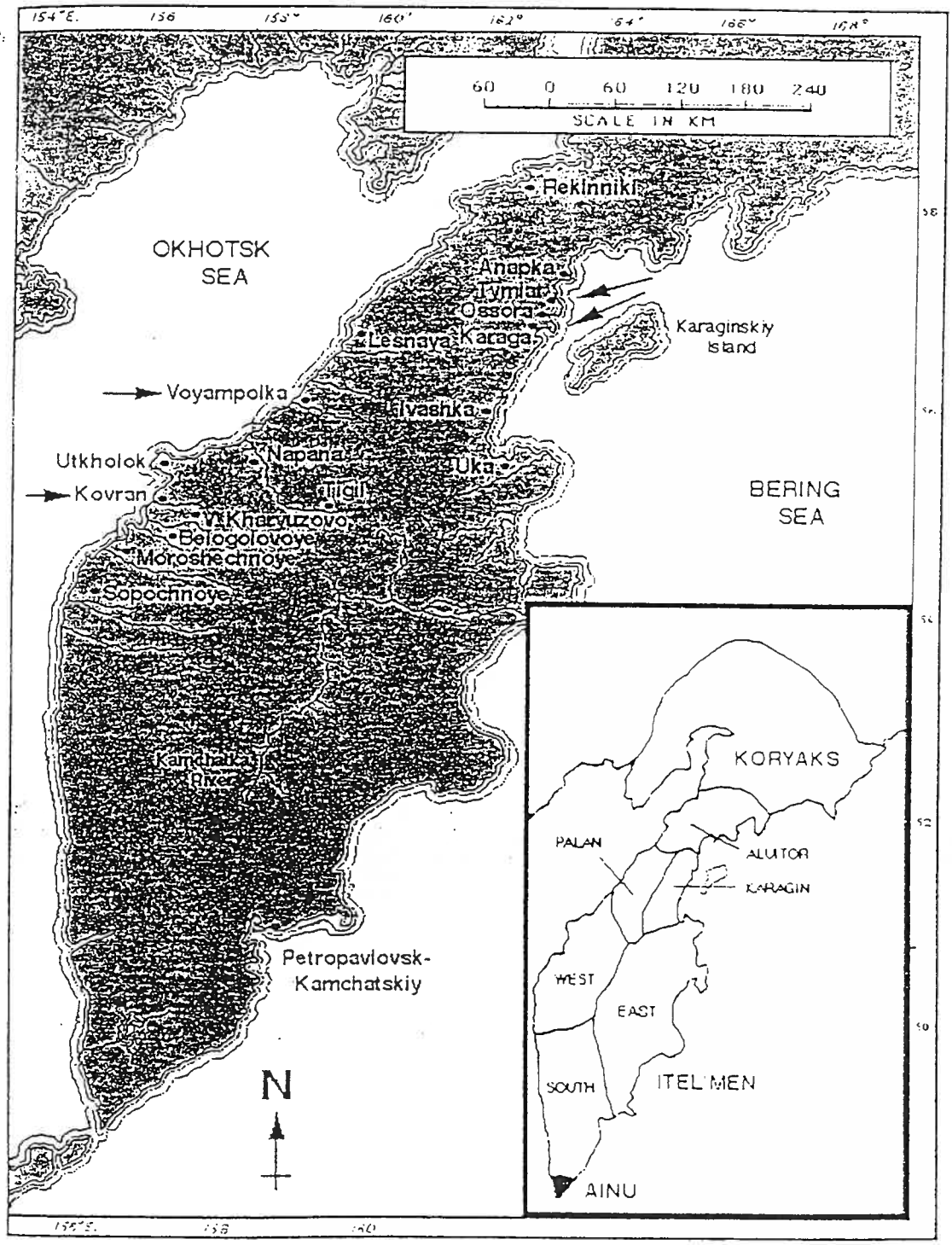


Fig.14 A map of Kamchatka showing the locations of the villages in which fieldwork was conducted in 1993 and 1996 (with arrows) and other traditional settlements of Koryak and Itel'men, the majority of which no longer exist. Inset: The traditional territories of the Koryaks (grey) and Itel'men (white) around the beginning of the eighteenth century, with the geographic locations of the dialectic subgroups for each population indicated on the Kamchatka peninsula.

(from Shurr et al, 1999)

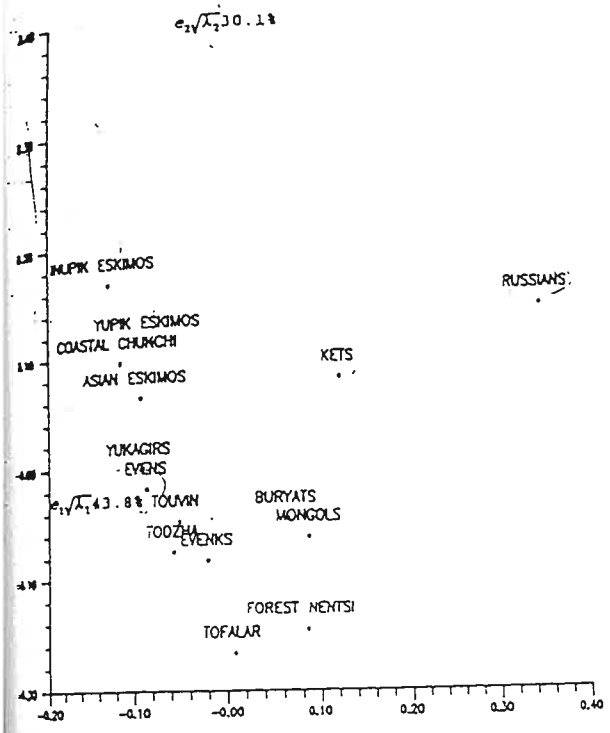


Fig.15.a A true least squares reduction of an R-matrix into a "genetic map" consisting of a plot between the first and second axes of 15 populations and 7 genetic loci. The two scaled eigenvectors account for more than 74% of the total variance.

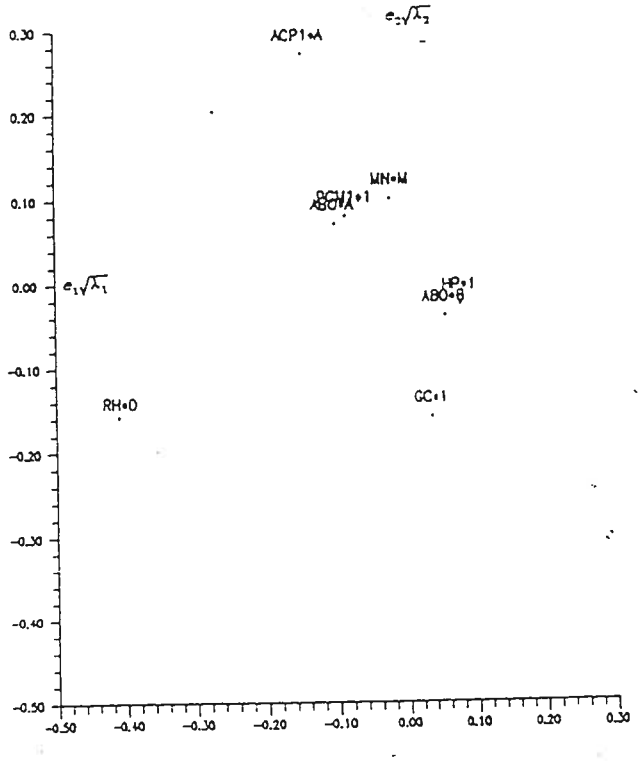


Fig.15.b Distribution of the alleles plotted along the first two scaled eigenvectors for the 15 populations shown in Fig. 2.

(from Crawford, 1997)

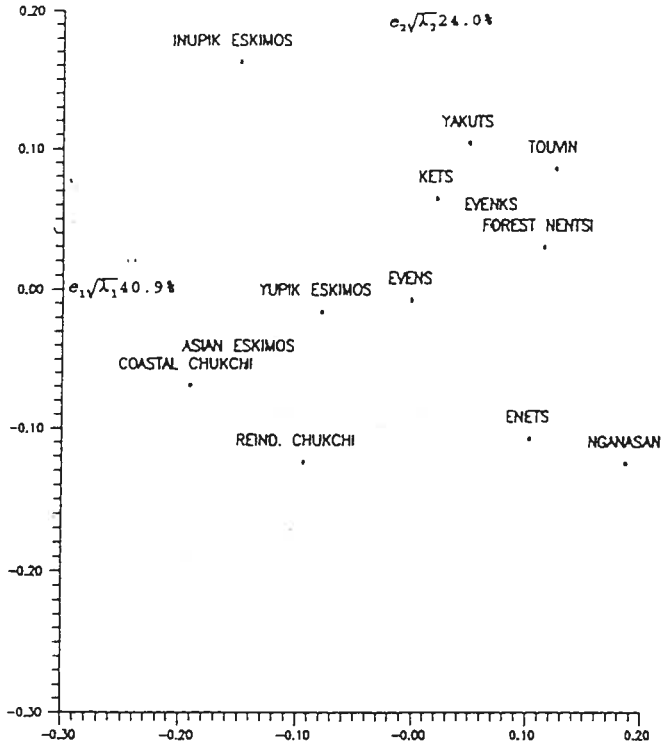


Fig.16. a Plot of the relationship between 13 populations, including 11 Siberian and 2 North American groups, reduced by least-squares from 9 alleles into two dimensions. The total variance subsumed by the first two eigenvectors is 65%.

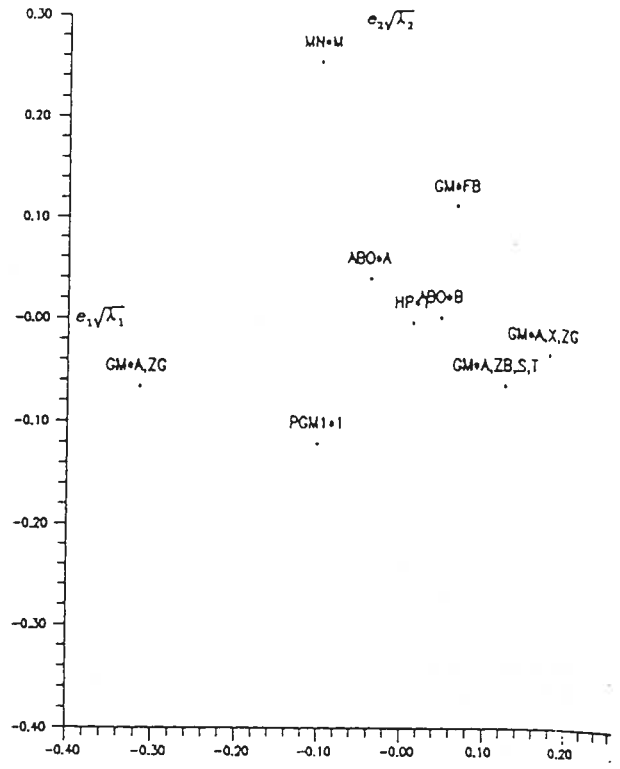


Fig.16.b Distribution of the nine alleles dispersed along the first two scaled eigenvectors in the genetic plot shown in Fig. 4.

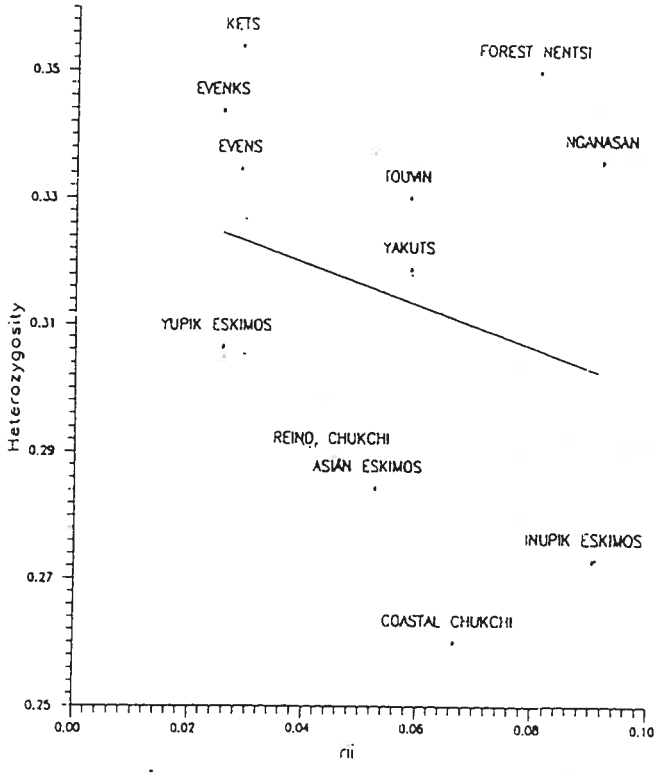


Fig. 17 A plot of the regression of heterozygosity (H) on the distance from the centroid of distribution (r_{ii}). The regression line shown is the theoretical one.

(from Crawford, 1997)

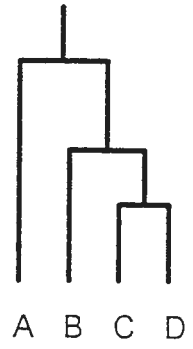
An Example of How A Branching Model and a Migration Model Can Produce the Same Genetic Distances

Model 1: Completely separate populations (A, B, C, D) of equal size diverging at different times

Dates of divergence (generations):

	A	B	C
B	1000		
C	1000	500	
D	1000	500	200

Dendrogram of genetic distances:



Model 2: All populations are of equal size begin at the same point in time, and are connected by gene flow

Migration matrix (entries indicate the probability of being in a column and originating in a row):

	A	B	C	D
A	0.99954	0.00008	0.00019	0.00019
B	0.00008	0.99924	0.00034	0.00034
C	0.00019	0.00034	0.99869	0.00079
D	0.00019	0.00034	0.00079	0.99869

Dendrogram of genetic distances:

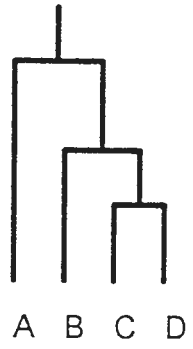


Figure 18 An example of how two different processes, a branching model and a migration model, can produce the same genetic distances. Model 1 is based on a highly simplistic model of drift in populations of equal size that have diverged at different times.[56] Genetic distance is obtained from the expected kinship after *t* generations of drift. Although overly simplistic, this model provides the type of distances expected given differential times of origin. Model 2 is based on a migration model where all populations are of equal size and have exchanged migrants according to the stated migration matrix. The genetic distances were derived from the genetic relationship matrix expected at equilibrium using the Rogers-Harpending migration matrix method.[57] Note that the two models produce the same dendrograms. Therefore, if we have only a dendrogram, we have no way of distinguishing which type of underlying model produced the distances. This type of problem is discussed in further detail by Felsenstein.[44]

(Relethford, 1995)