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Prehistory of Mongolian Populations as Revealed by Studies of Osteological, Dental, and Genetic Variation

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Disciplines

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The Prehistory of Mongolian Populations as Revealed by Studies of Osteological, Dental, and Genetic Variation

THEODORE G. SCHURR AND LENORE PIPES

During the past decade, researchers have made a concerted effort to characterize the biogenetic diversity of populations from East Asia. This issue has drawn attention because it is one of several world regions where the initial stages of the diversification of anatomically modern humans took place (Nei and Roychoudhury 1993; Cavalli-Sforza, Menozzi, and Piazza 1994; Jin and Su 2000). In addition, the region is marked by significant, historically documented demographic events such as wars, territorial conquests, and population relocations (Phillips 1969; Gongor 1970; Spuler 1971, 1989, 1994; Sinor 1990; Saunders 2001; Morgan 2007). One of these events was, of course, the expansion of the Mongol Empire, which at its greatest extent, in the 13th and 14th centuries, included a large portion of Eastern Europe and most of Asia (except for India and the southeast portion of the Asian continent). Although it was the most extensive, the Mongol Empire was actually one of a series of great steppe empires that expanded westward to threaten European powers (Sinor 1990; Spuler 1994; Saunders 2001; Morgan 2007).

Despite these well-known political conquests, however, relatively little is known about the Mongols before their rise under Chinggis Khan. It is generally thought that the formation of the current population in Mongolia followed a complex process that involved the mixing of ethnically different

peoples. In this regard, it should be noted that Mongols have traditionally organized themselves into a fluid and flexible system of confederations, tribes, clans, and families (Philips 1969; Gongor 1970; Spuler 1971, 1989, 1994; Badamkhatan 1987; Nyambuu 1992; Saunders 2001; Morgan 2007). This history is of great interest to biological anthropologists because the demography and social organization of these populations may have shaped the patterns of biogenetic diversity in them.

In this chapter, we review data from studies of cranial, dental morphological trait, and genetic diversity in Asian and Siberian populations in an effort to address questions about the origins and affinities of Mongolians. The information about the biological diversity of these populations is drawn from Chinese, Mongolian, Russian, and English language sources; from researchers working in different academic traditions; and from reports published during different stages of development of the field of physical anthropology, which now includes genetics. Consequently, the perspectives coming from these research reports may differ somewhat in their views on the relationships of Mongolians to other Asian populations, and their descriptions of the biological diversity within them. Regardless of these differences, the biological data obtained over the past several decades allow us to begin mapping the process by which Mongolian populations came into being, both in terms of chronology and geography.

It is this effort to map Mongolian history that allows us to discuss these biological data in terms of “-scapes.” While this concept is often used to describe specific interaction patterns that involve and transcend nation-states in the age of globalization, one can argue that such landscapes of globalization (Appadurai 1996) have been in existence and shaped Mongolian history for many millennia preceding the modern era. In fact, as we will argue, the patterns of biological variation that emerge from these studies reflect not only processes of evolution and adaptation but also social and political interactions and the exchange of ideas and technologies among populations living in the broad region that now encompasses the modern nation-state of Mongolia.

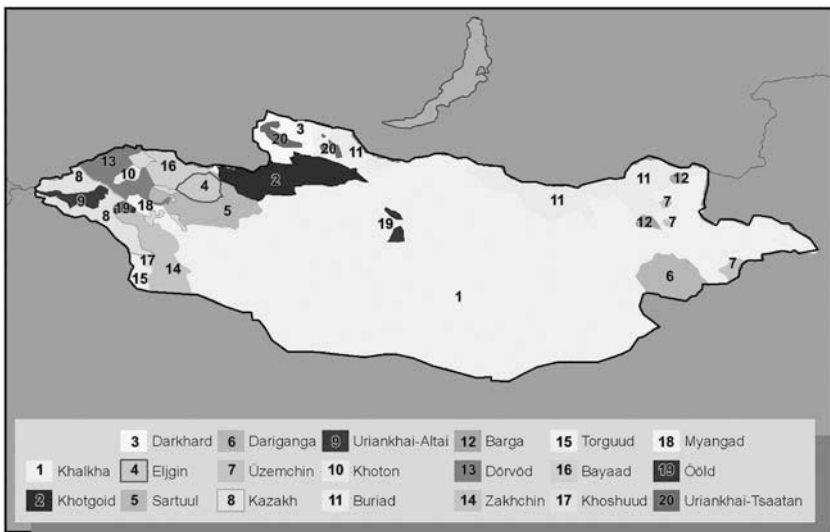
ETHNIC AND LINGUISTIC DIVERSITY IN MONGOLIA

Before describing the human biological diversity in Mongolia, we will discuss its ethnic and linguistic diversity. The presence of multiple ethnic

groups in the country—and the diversity of languages spoken there—reflect both the emergence of Turkic and Mongolic languages in Northeast Asia and the complex process of ethnogenesis that occurred over the past several millennia. Information about the ethnic identity and language practices of populations living in Mongolia also provides an important context for interpreting the biogenetic data of these groups.

Ethnic Diversity

The main ethnic group in Mongolia is the Khalkha, which is dispersed throughout the entire territory of Mongolia. There are also twenty other ethnic groups living mainly in the marginal areas of the country (Fig. 7.1). They can be generally placed in four culturally distinct clusters, including the Khalkha-Mongols, the Western Mongols or Oirats, Northeastern Mongols (Buriats, Darigangas), and Turkic speakers (Kazakhs, Khoton, Tsaatan, Tuvinians) (Badamkhatan 1987; Nyambuu 1992; Bulag 1998; Morgan 2007). The ethnic groups within the Western Mongol and Northeastern Mongol clusters have generally retained their ancient pre-Chinggis-period tribal names, and originated prior to the Mongol expansion. However, the origin of the Khalkha-Mongol cluster is more complicated, due to its being



7.1. Ethnic groups in Mongolia (modified from National Museum of Mongolian History map).

a rather recent (17th century) amalgam of intermixed tribes and families of heterogeneous origins rather than a single ethnic group per se (Badamkhatan 1987; Nyambuu 1992; Bulag 1998; Morgan 2007).

The Oirat Confederation was the most powerful group of tribes (which included the Durvuds, the Bayads, the Zakhchin, the Uriankhai, and the Torguuds) after the collapse of the Mongol Empire (Nyambuu 1992; Bulag 1998). During the late 16th and early 17th centuries, the scarcity of pasture lands and tribal warfare prompted the Torguuds and Durvuds to migrate to the steppes of western Siberia, which came under the control of Russia after the Yermak Expedition (Erdeniev 1985; Nasidze et al. 2005). In the late 17th century, the descendants of the Oirats formed the Kalmyk Khanate along the lower Volga River in Russia and became a new Mongolian-speaking ethnic group known as the Kalmyks (Erdeniev 1985; Galushkin, Spitsyn, and Crawford 2002). Today, the Torguuds live in the eastern and southeastern regions of the Kalmyk Republic, and are the numerically dominant tribe of the Kalmyk population (Erdeniev 1985; Galushkin, Spitsyn, and Crawford 2002).

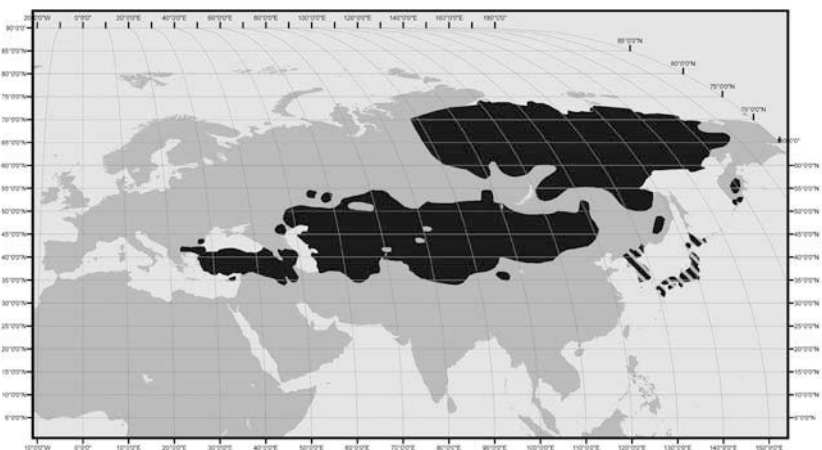
As for the Turkic cluster, the Kazakhs are the most recent arrival (late 19th century) to Mongolia. They are Sunni Muslim and trace their origins directly back to Chinggis Khan (Nyambuu 1992; Bulag 1998; Morgan 2007). The Kazakh populations consist of mostly Abak-Kerei and Naiman tribes who inhabited the Altai and Khovd regions of Mongolia after the Tarbagatai Protocol between Russia and the Qing Dynasty in 1864 (Nyambuu 1992; Bulag 1998; Paine 1996; Finke 1999). By contrast, the Tuvinians represent the ancient inhabitants (at least from the 11th century) of the far west of Mongolia (Nyambuu 1992; Bulag 1998; Morgan 2007).

Several ethnic groups are known to have non-Mongolian origins. The Khotons were brought as serfs from eastern Turkestan (now Sinkiang) to this region by Dzüngarian khans in the 17th century (Nyambuu 1992; Katoh et al. 2005b), while the Sartuuls descend from skilled slaves brought from Iran and Khorasm in the 13th and 14th centuries (Badamkhatan 1987; Nyambuu 1992). However, some groups have ambiguous, albeit non-Mongolian, origins, such as the Darkhads, a small group (~15,000 people) inhabiting the northern taiga region of Mongolia, the Uriankhai (Western Mongol cluster), and the Khamnigan (now a clan of the Buriats). In addition, a few groups were resettled partially or completely within the territory of Mongolia during Manchu rule, such as the Olet and Dariganga, with the latter forming ~300 years ago and now residing mostly in southeastern Mongolia

(Nyambuu 1992; Bulag 1998). Furthermore, the majority of Buriats lives to the south and east of Lake Baikal but also inhabits parts of northern and eastern Mongolia (e.g., Levin and Potapov 1964; Shimizu et al. 2006).

Linguistic Groups

The diversity of human groups in Mongolia is also reflected in the linguistic diversity of the region. A total of thirteen Mongolic languages are spoken in Northeast Asia, including Buriat (Mongolia, China, and Russia), Kalmyk-Oirat (chiefly from the Kalmykia Autonomous Region), and Mongolian (Golden 1992; Janhunen 2003). The latter is the primary language of most residents of Mongolia and is spoken by around 2.5 million people in Inner Mongolia, Mongolia, and Russia. Most linguists view Mongolic languages as belonging with Turkic and Tungusic languages in the Altaic family (Poppe 1965; Janhunen 2003; Starostin, Dybo, and Mudrak 2003) (Fig. 7.2). Within the Tungusic group, Manchu (Dongbei in China) had the greatest number of speakers and was formerly the *lingua franca* between China and the outside world for over 200 years, although today it is practically extinct (Huang 1990; Gorelova 2002; Janhunen 2003). Turkic languages are spoken principally in a nearly continuous band from Turkey, Armenia, and Azerbaijan through the Central Asian republics of Kazakhstan, Uzbekistan, Turkmenistan, Kyrgyzstan, and Tajikistan to Xinjiang in China (Golden 1992;



7.2. Map showing distribution of Altaic languages in northern Eurasia (modified from http://en.wikipedia.org/wiki/File:Altaic_family2.svg).

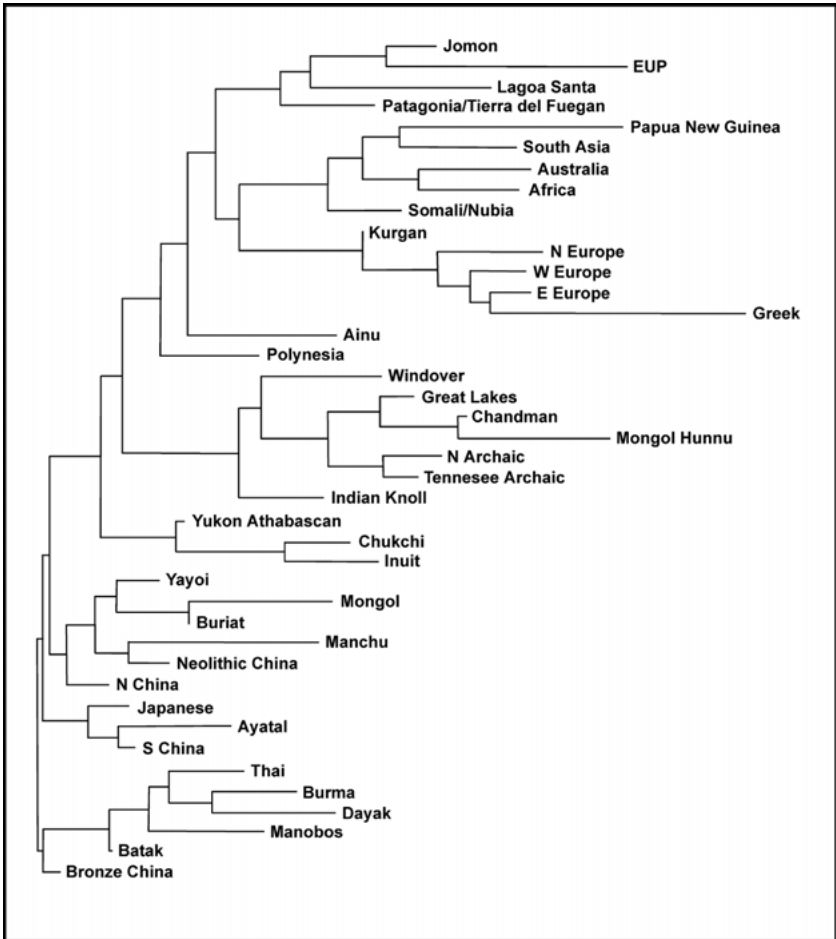
Janhunen 2003). In addition, because they share many linguistic features with Mongolic and Turkic languages, Japanese and Korean are also believed by some (but not all) linguists to be distantly related to them (Poppe 1965; Clauson 1968; Miller 1971; Starostin 1991; Vovin 2001).

CRANIO-FACIAL VARIATION IN MONGOLIA

Having outlined the ethnolinguistic diversity of Mongolia, we will now begin our discussion of the biogenetic diversity found in the country and surrounding areas. Over the past several decades, Russian and Mongolian scholars have undertaken extensive studies of the cranio-facial characteristics of human groups in northern Eurasia. These studies suggest that modern-day Mongolians inhabit the region proposed to represent the origin of the “Mongoloid” racial type,¹ which is prevalent in present-day Mongols as well as the majority of southern and eastern Siberian ethnic populations (Alexeev 1978; Alexeev and Gohman 1984; Alexeev and Trubnikova 1984; Bulag 1998).² However, almost all of the Turkic speakers in Mongolia are viewed as belonging to the Southern Siberian (Afanasiev) racial type. This type may have emerged during the introduction of steppe nomadism to East and Central Asia at the end of the Neolithic (2300–2000 BCE) by “Caucasoid” tribes of supposed Indo-European origin, who brought with them an influx of West Eurasian genes and physical traits (Phillips 1969; Alexeev, Gohman, and Tumen 1987; Thornton and Schurr 2004; Anthony 2007). The next several thousand years were marked by the appearance of a succession of dominating tribes that emerged through power struggles in the region (Spuler 1971, 1989, 1994; Sinor 1990; Morgan 2007). It has been suggested that, by the end of the Xiongnu domination of East and Central Asia (3rd century C.E.), the modern biological appearance of Mongolia’s inhabitants had probably taken form (Alexeev 1978; Alexeev and Gohman 1984; Alexeev and Trubnikova 1984; Alexeev, Gohman, and Tumen 1987).

Using a less typological approach, Western and Japanese scholars have also analyzed craniofacial metric traits in world populations³ (Fig. 7.3). They observed that the Mongolian Bronze Age Chandman and Mongol Hunnu (Xiongnu) appeared similar to modern Native Americans from the Great Lakes regions, as well as prehistoric Archaic Period individuals from North America (Brace et al. 2001; Seguchi 2004). Interestingly, the Mongolian Chandman sample was not closely related to the roughly contemporaneous

Chinese Bronze Age sample from Anyang. Nor were Mongols strongly related to the Mongolian Chandman, the Mongol Hunnu, or the late Paleolithic Chinese Upper Cave 101 sample. These patterns suggested biological differences between the Upper Paleolithic inhabitants of greater Northeast Asia and the Neolithic people who later expanded into Siberia and East Asia, as well as the influence of ancient Europeans on Bronze Age Mongolia.



7.3. Neighbor-joining tree based on Mahalanobis distances calculated from craniofacial metric data. Branch lengths reflect genetic distances, while the vertical distances between the lines have no quantitative significance. Bootstrap values represent the percentage of times per 1,000 runs that the branches appear together (modified from Seguchi 2004).

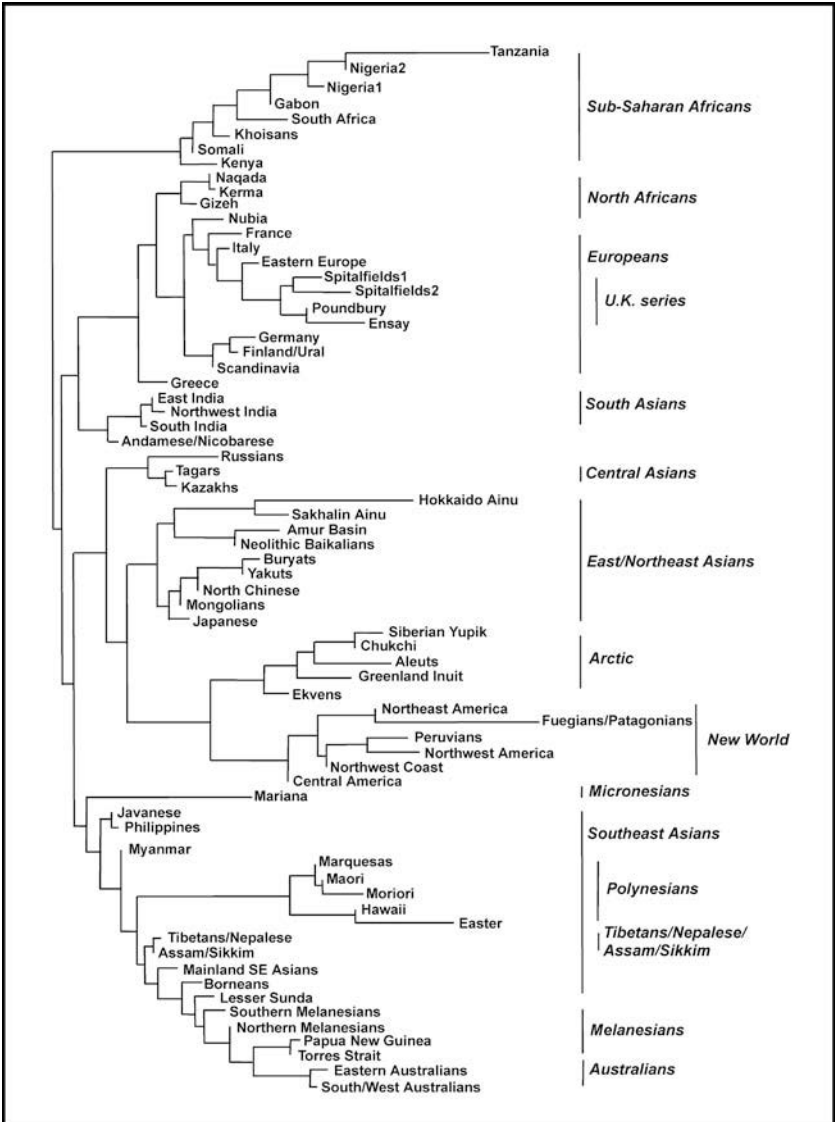
In a similar study, Hanihara, Ishida, and Dodo (2003) investigated the distribution of cranial metric traits in world populations but examined a more expansive data set than used in previous reports. In their neighboring-joining tree of mean measure of divergence (MMD) estimates, the East Asian (EAS), plus Northeast Asian (NEAS), and European samples formed two clearly discernable clusters; Mongols were situated in the former, while Central Asian (CAS) samples were located between the other clusters (Fig. 7.4). This study further revealed the clinal nature of discrete cranial trait variation across regions and the morphological discontinuity in isolated populations such as the Ainu, which probably resulted from restricted gene flow and local adaptation.

DENTAL TRAIT VARIATION IN MONGOLIA

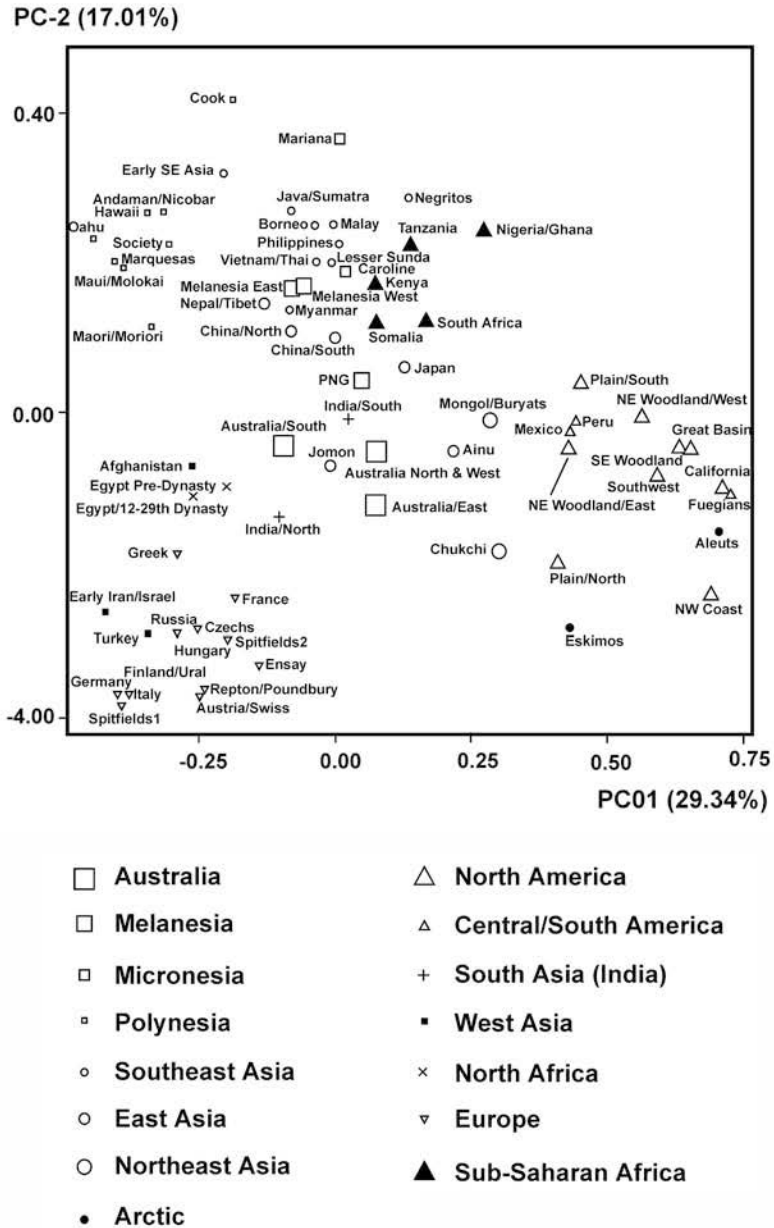
Various researchers have characterized dental variation in Asian and Native American populations in order to reconstruct the peopling of northern Eurasia and the Americas. Turner (1987, 1990, 1992; Turner, Manabe, and Hawkey 2000) interpreted the East Asian dental variation as indicating that the evolution of the Sundadont dental pattern took place in Southeast Asia. Sundadonty is a more generalized dental trait complex and was probably brought to Southeast and East Asia during the initial human expansions into these regions. Most extant populations from Southeast Asia and Australasia exhibit this dental pattern.

East Asian populations with Sundadont dental traits subsequently spread into Northeast Asia. Sundadonty likely evolved into sinodonty in northern China, Mongolia, and southern Siberia, possibly around 30,000 years ago, and certainly before 14,000 years ago, when Paleo-Siberian sinodonts had crossed Beringia into the New World (Turner 1987, 1990, 1992; Turner, Manabe, and Hawkey 2000). The Sinodont dental complex is characterized by traits such as shovel-shaped incisors, single-rooted upper first premolars, triple-rooted lower first molars, and other features. It is typical of Northeast Asian populations but also found in many Native Americans.

In their analysis of dental metric traits in world populations, Hanihara and Ishida (2005) also observed some of these same patterns (Fig. 7.5). In particular, they noted that the Native American, Arctic, and NEAS samples had relatively large mesiodistal crown diameters. Mongols and Buriats



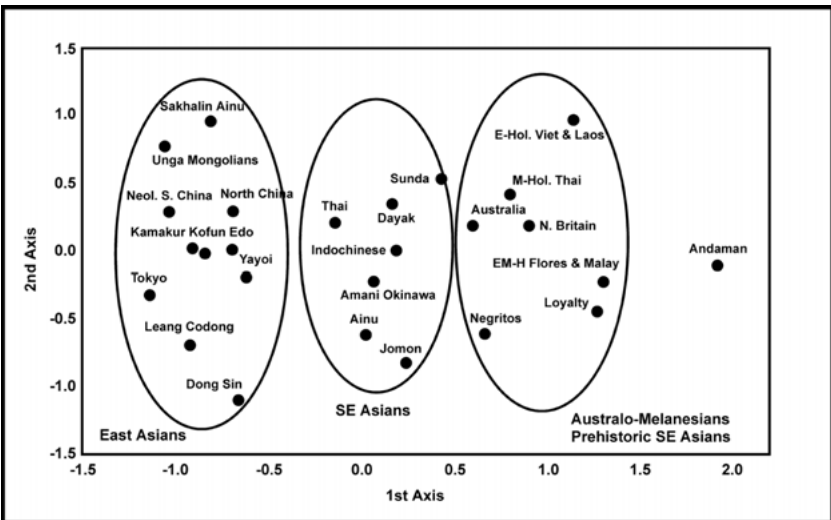
7.4. Neighbor-joining tree based on mean measure of divergence (MMD) distance estimated from cranial metric traits (Hanihara, Ishida, and Dodo 2003).



7.5. Scattergram of first and second principal components (PC) scores based on C-score data generated from dental metric traits (Hanihara and Ishida 2005).

showed similarities to Ainu, Jomon, and Japanese, but also to Native Americans. At the same time, they were slightly more distant from Chinese, Indians, and Aboriginal Australians, all of which have Sundadont traits.

In a similar study, Matsumura and Hudson (2005) examined both metric and nonmetric dental data to test the “two-layer” or immigration hypothesis for the peopling of Southeast Asia. They found close affinities between recent Australo-Melanesian and prehistoric Southeast Asian (SEAS) populations, whereas most modern Southeast Asians possessed a mixture of traits associated with East Asians and Australo-Melanesians (Fig. 7.6). In addition, for both metric and nonmetric dental traits, Mongols clustered with other EAS populations, including Buriats, Koreans, Japanese, and North Chinese populations.



7.6. Multi-dimensional scaling plot of genetic distances based on 16 nonmetric dental traits (redrawn from Matsumura and Hudson 2005).

Overall, these dental trait studies provide general support for the division of EAS populations into Sinodonts and Sundadonts. However, Matsumura and Hudson (2005) argue that Chinese and Japanese populations are more similar to SEAS populations, and that the dental characteristics of most modern Southeast Asians are a mixture of traits associated with East Asians and Australo-Melanesians. In fact, the trait variation of EAS/NEAS dental configurations is relatively large, suggesting a complex population

history marked by an early divergence of populations in this region and multiple migrations from outside sources (Hanihara 2008).

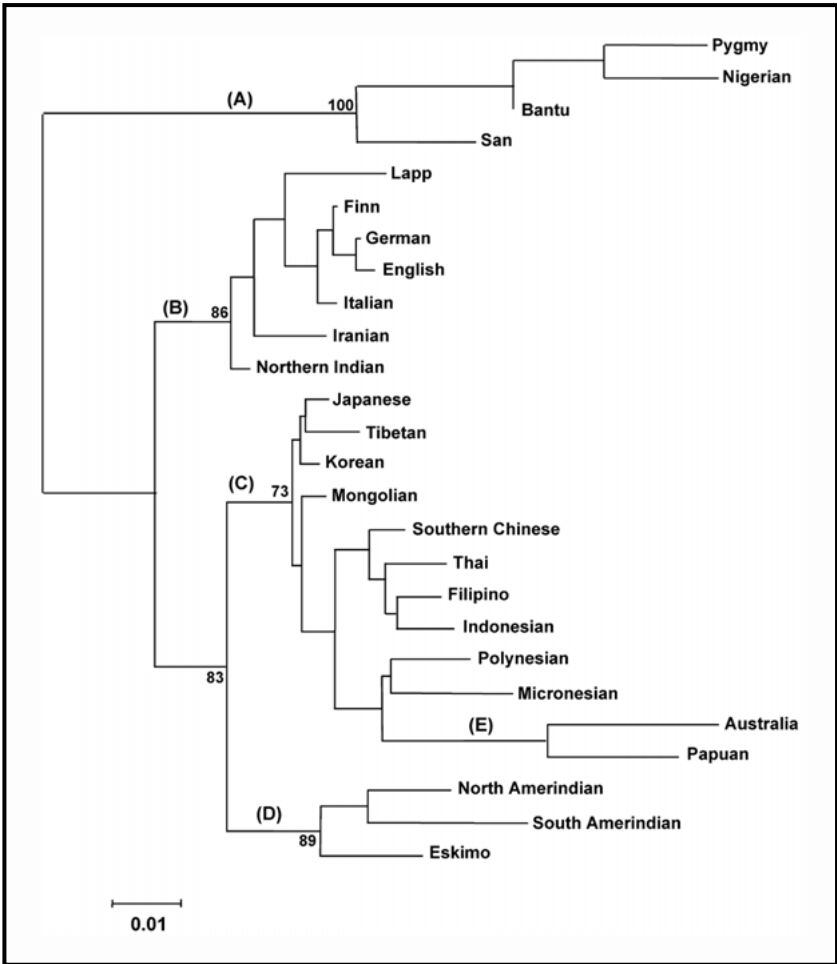
GENETIC VARIATION IN MONGOLIA

In the previous section, we discussed morphological traits that are variable in different human populations and across different regions. Because these traits are under considerable genetic control, they are quite useful for population history studies. In this section, we discuss data from a number of genetic studies of Asian populations and what they indicate about the history of Mongolian populations. The loci discussed include classical genetic markers, immune system genes, and molecular genetic markers such as the mitochondrial DNA (mtDNA) and the Y-chromosome. This review of genetic studies is by no means exhaustive, but does provide a reasonably thorough overview of patterns of diversity in Mongolian and other Asian populations.

Classical Genetic Markers

Since the early 1970s, populations from different regions of Asia, including Mongolia, have been extensively analyzed for variation in some blood group systems, serum proteins, and red blood cell enzymes as well as immunoglobulins and HLA class-I and -II loci (Goedde et al. 1987; Matsumoto 1988; Nei and Roychoudhury 1993; Novoradovsky et al. 1993; Cavalli-Sforza, Menozzi, and Piazza 1994). In the early studies, Mongols were observed to cluster together with NEAS populations (i.e., Tibetans, Koreans, and Japanese) and were next closest to SEAS populations. They were less similar to Australo-Melanesian and New World populations (Nei and Roychoudhury 1993) (Fig. 7.7).

However, relatively few of the studies involving biochemical markers or polymorphisms were conducted with the aim of determining the genetic interrelations of Mongolian ethnic groups. Studies analyzing these markers have revealed a high genetic diversity within Mongolian populations but statistically insignificant values of genetic differentiation for regional populations (Rychkov and Batsuuri 1987; Batsuuri 1995; Batsuuri, Ganbold, and Sodgerel 1995; Chimge and Batsuuri 1995, 1999; Chimge et al. 1997; Ganbold, Batsuuri, and Tamjidmaa 1995; Ganbold, Batsuuri, and Sharav 1995; Galushkin, Spitsyn, and Crawford 2002). They also showed a decreasing



7.7. Neighbor-joining tree for 26 human populations based on genetic distances, (D_A), estimated from classical genetic markers, with bootstrap values indicated for each interior branch. Major groups of human populations are Africans (A), Caucasians (B), Greater Asians (C), Amerindians (D), and Australopapuans (E) (modified from Nei and Roychoudhury 1993).

cline of alleles from biochemical and immunological loci typically seen in European and West Asian populations from east to west.

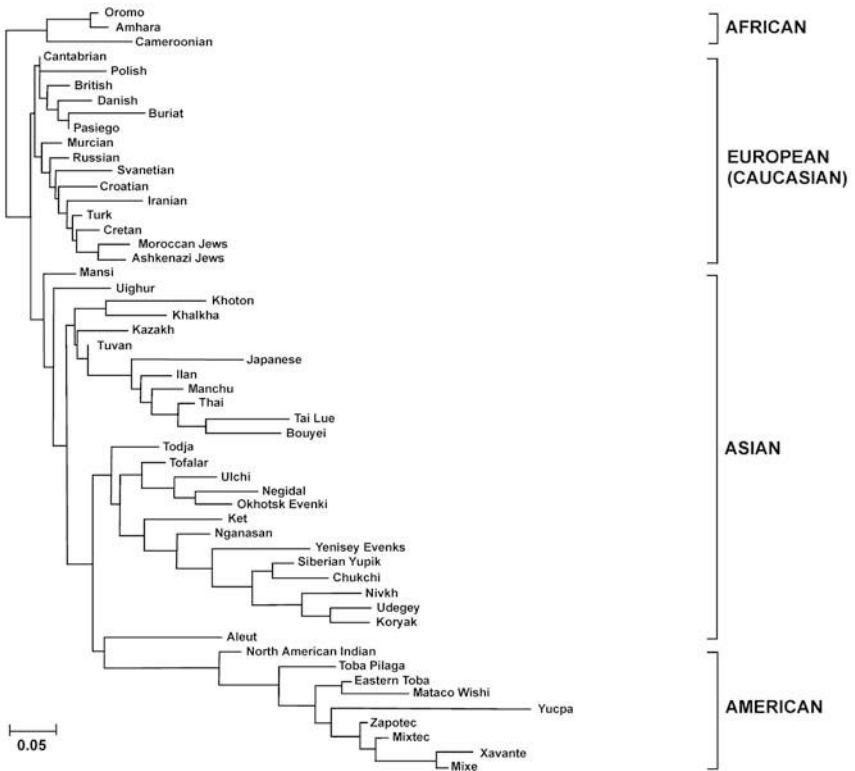
Immunoglobulin (antibody) data have also yielded interesting insights into Asian population history. Matsumoto (1988) summarized the distribution of immunoglobulin allotypes (G_m) in populations from different

regions of Asia. In general, Northeast Asian populations were characterized by the presence of four Gm haplotypes—ag, axg, ab3st, and afb1b3. They were further divided into two groups based on genetic distances estimated from Gm haplotype frequency distributions. The southern group was characterized by a high frequency of Gm^{afb1b3} and a low frequency of Gm^{ag}, while the northern group was characterized by a high frequency of both Gm^{ag} and Gm^{ab3st} and an extremely low frequency of Gm^{afb1b3}.

Populations in China, mainly Han but also minority ethnic groups, exhibited a remarkable heterogeneity of Gm allotypes from north to south. By contrast, Korean and Japanese populations were considerably more homogeneous with respect to these genetic markers. The center of dispersion of the Gm^{afb1b3} gene characterizing SEAS populations was identified as the Guangxi and Yunnan area in southwest China (Matsumoto 1988).

The Gm^{ab3st} haplotype, which occurred at its highest frequency in northern Baikal Buriats, appeared widely in Northeast Asia. While dropping sharply in frequency from mainland China to Taiwan and Southeast Asia, this haplotype was present at high frequency among Koryaks, Yakuts, Tibetans, Olunchuns, Tungus, Koreans, Japanese, and Ainu, and was probably also introduced into the Hui, Uighurs, Indians, and Iranians through the Mongol expansion. Based on these findings, Japanese and Koreans clustered within the Northeast Asian group, which likely originated in the Baikal area of Siberia (Matsumoto 1988).⁴

Other immune gene data provided a somewhat different picture of Mongol genetic ancestry. In their analysis of HLA class-I and -II loci from CAS, EAS, and NEAS populations, Uinuk-ool, Takezaki, and Klein (2003) identified a set of alleles and haplotypes that differentiated all of the indigenous Asian populations from other Old World populations. This set was primarily responsible for the grouping of the Asian populations in a single cluster within their population tree (Fig. 7.8). Mongols clustered with Turkic-speaking groups in the part of the tree that also included Japanese, Han and Manchu, and SEAS populations. They were separated from the cluster that included nearly all native Siberian populations, and were also distant from Native American populations. The ethnic groups that did not fall into the native Siberian cluster (Buriat, Mansi, and Tuvan) showed clear evidence of admixture with European/West Eurasian or CAS populations.



7.8. Neighbor-joining tree of human populations based on genetic distances calculated from allele frequencies at the HLA-DRB1, -DQB1, and -DQA1 loci (Uinuk-ool, Takezaki, and Klein 2003).

Mitochondrial DNA Diversity

Numerous studies over the past twenty years have defined a variety of mitochondrial DNA (mtDNA) lineages, or haplogroups, in different human populations. Because the mtDNA is strictly maternally inherited and does not recombine, one can reconstruct female genetic genealogies using mutational information contained in haplotypes defined in different human populations. Based on their mutational characteristics, these haplotypes⁵ and the haplogroups to which they belong can be arranged in a network or a tree reflecting their sequence of evolution and phylogeographic relationships.

The initial examination of mtDNA variation in Mongolian populations began in the early-to-mid 1990s. Studies by Sambuughin, Petrishev, and

Rychkov (1991, 1992) provided a general impression of mtDNA diversity in the region, but the data were of sufficiently low resolution to reveal any phylogeographic patterns there. Kolman, Sambuughin, and Bermingham (1996) expanded on these findings through the analysis of an expanded set of markers in and sequencing of the hypervariable region I (HVSI) of the mtDNA genome. A similar analysis of Mongolian populations by Merriwether et al. (1996) was directed toward locating the ancestral homeland of Native American populations. Together, these two studies demonstrated that the four major mtDNA haplogroups commonly seen in Native American populations (A–D) were also present in Mongolia (e.g., see Schurr 2004). These maternal lineages comprised roughly half of the mtDNAs in the Mongolian populations sampled, with most of the rest likely belonging to East Eurasian haplogroups.⁶ However, due to methodological constraints, these researchers were unable to further define these lineages.

Subsequent studies have provided much more detail about the mtDNA composition of Mongol populations from Xinjiang, Mongolia, and Inner Mongolia, as well as a tentative picture of diversity in other ethnic groups in Mongolia (Kong et al. 2003; Yao et al. 2004; Gokcumen et al. 2008; Pipes, Labuda, and Schurr 2008). All of them have generally shown that the haplogroup distribution in Mongolians consists of mostly East Eurasian lineages (A, B, C, D, F, G, M7, M8, M9, N9a, Y, Z), with a low to modest frequency (4–20 percent) of West Eurasian lineages (H, J, T, U, and W) (Fig. 7.9; see DVD). Most of the East Eurasian haplogroups are observed in indigenous Siberian and NEAS populations (Torroni et al. 1993, 1994; Starikovskaya et al. 1998, 2005; Schurr et al. 1999; Derbeneva et al. 2002a,b; Derenko et al. 2002, 2003, 2004; Pakendorf et al. 2003; Schurr and Wallace 2003; Gokcumen et al. 2008), while M7 haplotypes are much more common in Chinese, Japanese, and Korean populations (Yao et al. 2002; Kivisild et al. 2002; Tanaka et al. 2004; Lee et al. 2006). In addition, haplogroups B and F are commonly seen in East and SEAS populations as well as Tibetans (Ballinger et al. 1992; Torroni et al. 1994; Melton et al. 1995; Qian et al. 2001; Schurr and Wallace 2002; Yao et al. 2002; Kong et al. 2003; Tanaka et al. 2004; Macaulay et al. 2005; Li et al. 2007), but represent a small proportion of the mtDNAs in Mongolians (Fig. 7.9; see DVD).

Recent work with SEAS populations has provided insights into the antiquity and diversification of some of these maternal lineages (Macaulay et al. 2005; Li et al. 2007). Haplogroup ages were estimated for the high

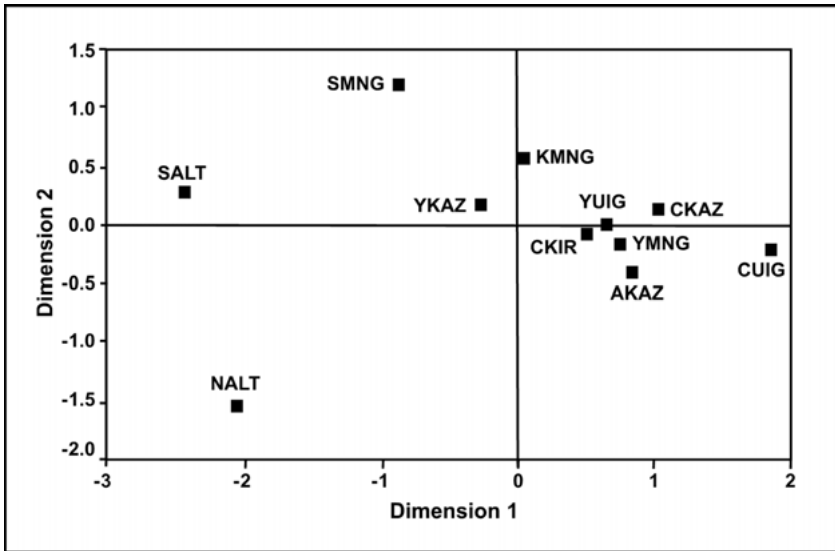
frequency haplogroups (B, F, M7, R) in the region, and they were found to originate about 50,000 years ago. In contrast, subhaplogroups appearing within specific regions or language families arose some 10,000–20,000 years ago. These findings confirm the early human colonization of Southeast Asia and the later emergence of regional gene pools, along with gene flow between them. This same process of genetic differentiation undoubtedly took place in Northeast Asia, giving rise to the distribution of maternal lineages observed there.

By contrast, the West Eurasian haplogroups are much more common in populations inhabiting regions to the west of Mongolia (Comas et al. 1998, 2004; Quintana-Murci et al. 2004; Gokcumen et al. 2008). All of these maternal lineages probably originated in Central or West Asia (Torroni et al. 1996, 1998; Macaulay et al. 1999; Richards et al. 2000; Capelli et al. 2003) prior to their being brought to East Asia by Neolithic steppe populations some 5,000 years ago (Golden 1992; Hiebert 1994; Frachetti 2002; Renfrew 2002; Thornton and Schurr 2004; Anthony 2007). Overall, the frequency of East Eurasian haplogroups decreases in an east-to-west direction, whereas the opposite trend occurs for West Eurasian haplogroups. This pattern likely emerged through the complex population interactions occurring across this region over the past several millennia.

The end result of these dynamic interactions is that most Turkic- and Mongolic-speaking groups possess a common set of maternal haplogroups (C, D, G2a, and H) and a minimal number of haplotypes from these lineages at appreciable frequencies. However, the overall patterns of haplotype diversity in these groups vary considerably, based on their local histories. These findings imply that most contemporary Turkic and Mongolic ethnic groups emerged from a common mtDNA pool that was widely distributed in Central and East Asia (Gokcumen et al. 2008). This interpretation is supported by the statistical analysis of mtDNA sequence diversity in which Mongol, Kazakh, Kirghiz, and Uighur populations cluster together (Fig. 7.10).

Y-chromosome Variation

Numerous studies over the past dozen years have defined a variety of paternal lineages, or Y-chromosome haplogroups, in different human populations.⁷ The Y-chromosome evolves more slowly than the mtDNA but has accumulated sufficient variation to permit the delineation of specific branches of the paternal genealogy for human populations.



7.10. A multidimensional scaling plot (MDS) of genetic distances estimates based on mtDNA data for Central Asian populations and Altaian Kazakhs. Population abbreviations: Altaian Kazakhs (AKAZ) and Mongolians (SMNG) from Gokcumen et al. 2008; Mongolians (KMNG) from Kolman, Sambuughin, and Bermingham 1996; Northern Altaians (NALT) and Southern Altaians (SALT) from Zhadanov et al. 2006; Kazakhs (CKAZ), Kirghiz (CKIR), and Uighurs (CUIG) from Comas et al. 1998; and Uighurs (YUIG), Mongolians (YHMG), and Kazakhs (YKAZ) from Yao et al. 2004. The stress value for this plot is 0.0583 (Gokcumen et al. 2008).

These informative markers occur in the nonrecombining region of the Y-chromosome (NRY). Based on their mutational characteristics, these NRY haplogroups can be arranged in a network reflecting their sequence of evolution and phylogeographic relationships.

One of the first studies of Y-chromosome variation in Mongols involved the survey of CAS populations (Zerjal et al. 2002). This analysis revealed a wide array of haplogroups, as well as clinal distribution of West Eurasian haplotypes from west to east, in this broad region. Mongols had primarily haplogroups C and K along with low to moderate frequencies of other haplogroups present in Central/East Asia (D, N3, O3, R1a1) and low frequencies of paternal lineages more commonly seen in populations from the Caucasus and West Asia (J, L, R1a). This pattern of Y-chromosome diversity is generally consistent with the findings of other studies of CAS/EAS populations (Tajima et al. 2002; Katoh et al. 2005a; Xue et al. 2006). The

multidimensional scaling (MDS) analysis of genetic distances estimated from NRY data in these populations showed Mongolians to be similar to Uighurs and Uzbeks and somewhat more distant from Kazakhs, Kirghiz, and Tajiks to the west of them (Fig. 7.11; see DVD).

A similar analysis of the distribution pattern of Y-chromosome haplogroups revealed statistically significant genetic differences between the populations of the Baikal and Altai–Sayan regions (Derenko et al. 2006). These findings were attributed to the differential contribution of CAS/EAS and West Eurasian paternal lineages to the gene pools of modern South Siberians. In this regard, populations from the Baikal region demonstrated a higher prevalence of CAS/EAS lineages, while the populations of the Altai–Sayan regions had a more substantial paternal contribution from populations originating to the West.

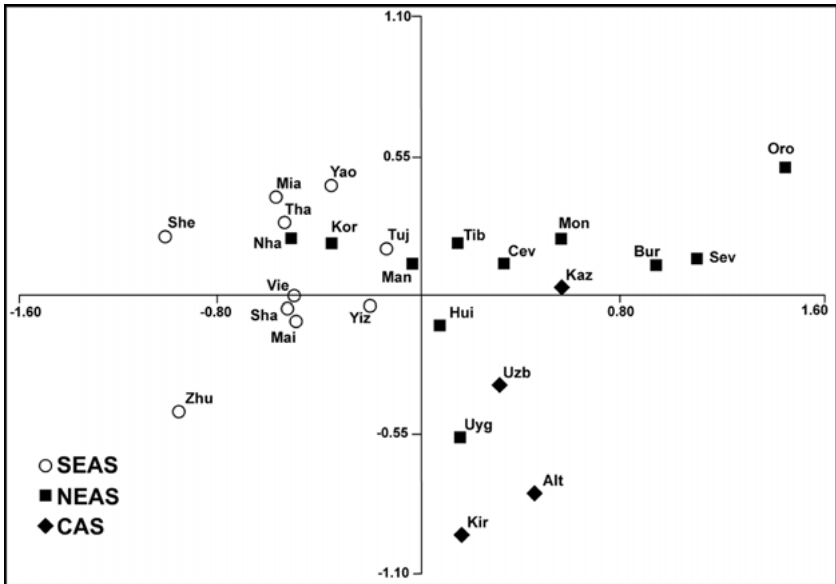
A comparable NRY study of CAS, NEAS, and SEAS populations also elucidated aspects of Central and East Asian history (Karafet et al. 2001). This study revealed that NEAS and SEAS populations shared a number of haplogroups, and NEAS and CAS groups also shared a number of paternal lineages not seen in SEAS, while very few were shared between all three of them. The MDS analysis of the F_{ST} values estimated from these Y-chromosome data revealed a broad clustering of CAS and NEAS groups and a general closeness of certain NEAS to SEAS groups (Fig. 7.12). It also placed Mongols between Tibetans, Kazakhs, Evenks, and Buriats, suggesting a strong East Asian genetic background with some relatedness to Turkic-speaking populations. These data further indicated that NEAS populations had formed through the mixing of East and Central Asian populations beginning some 30,000–40,000 years ago.

GENETIC INSIGHTS INTO MONGOLIAN POPULATION HISTORY

Having provided a basic background on biological diversity in Northeast Asia, we will now discuss how genetic data have been used to address specific questions about Mongolian population history.

Prehistoric Ancestors of Modern Mongolians

Further insights into the genetic history of Mongolian populations may come from the study of ancient DNA extracted from the skeletal remains

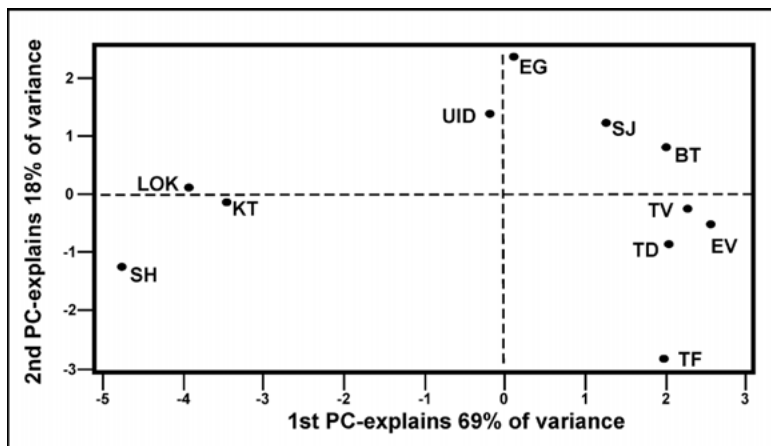


7.12. MDS plot of 25 Asian populations, based on F_{ST} genetic distances estimated from Y-chromosome data. Population abbreviations: Altai (Alt), Buriats (Bur), Chinese Evenks (Cev), Hui (Hui), Kazakhs (Kaz), Kirghiz (Kir), Koreans (Kor), Malaysian (Mal), Manchu (Man), Miao (Mia), Mongolians (Mon), Northern Han (Nha), Oroqen (Oro), Siberian Evenks (Sev), She (She), Southern Han (Sha), Taiwanese Han (Tha), Tibetans (Tib), Tujians (Tuj), Uighurs (Uyg), Uzbeks (Uzb), Vietnamese (Vie), Yao (Yao), Yizu (Yiz), and Zhuang (Zhu) (Karafet et al. 2001).

of archaeological populations. Ongoing study of Neolithic populations from the Cis-Baikal region has provided this kind of evidence. To ascertain the genetic ancestry of the Cis-Baikal populations, researchers compared mtDNA diversity in the early Neolithic Kitoi and late Neolithic Serovo-Glazkovo populations with that of modern populations from the Lake Baikal region (Mooder et al. 2003, 2005, 2006; Schurr 2003; Schurr et al. 2010). They observed distinct patterns of mtDNA diversity in the two prehistoric Cis-Baikal populations, the extent of which also seemed to mirror differences in mortuary practices, nutritional status, social organization, and other features (Mooder et al. 2005, 2006).

Mooder et al. (2006) also noted differences in population affinities for the Kitoi versus the Serovo-Glazkovo (Fig. 7.13). The Kitoi clustered near the Shors and Kets, whereas the Serovo-Glazkovo was located closer to the populations currently residing around the Baikal region. In addition, when

compared with the prehistoric Egiin Gol population from Mongolia, the Serovo-Glazkovo showed genetic similarities to this Xiongnu population. These data supported the distinctiveness of the two Neolithic peoples of the Cis-Baikal region and also implied that the Kitoi had left the region to be replaced by populations ancestral to contemporary Turkic- and Mongolic-speaking groups.



7.13. PC map of mtDNA haplogroup distributions for prehistoric and modern populations as estimated by F_{ST} . Population abbreviations: LOK, Lokomotiv; UID, Ust'-Ida; SH, Shorians; KT, Kets; TF, Tofalars; TD, Todjins; TV, Tuvinians; EV, Evenki; BT, Buryats; SJ, Sojots; EG, Egiin Gol (Mooder et al. 2006).

Furthermore, the mtDNA analysis of skeletal remains from the Egiin Gol site revealed that the populations of the Xiongnu period (3rd century BCE–2nd century CE) were characterized by mixed genetic ancestry, as 11 percent of their mtDNAs belonged to West Eurasian haplogroups (Keyser-Tracqui, Crubézy, and Ludes 2003; Keyser-Tracqui et al. 2006). The moderate frequencies and diversity of West Eurasian mtDNA lineages within the populations of southern Siberia and Mongolia (Derenko et al. 2002, 2003; Yao et al. 2002; Comas et al. 2004; Gokcumen et al. 2008) support the early penetration of ancient European steppe cultures into this region during the Neolithic, Bronze Age, and early Iron Age. The genetic results are consistent with physical anthropological data from the Pre-Scythian Period that show the presence of crania with “Caucasoid” and mixed “Caucasoid-Mongoloid” features in the south Siberian steppe and Western Mongolia

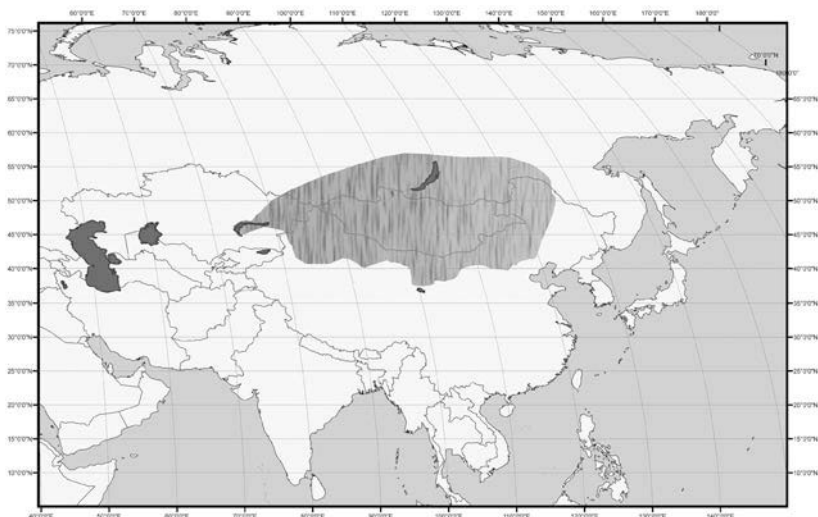
(Alekseev and Gohman 1984; Alekseev, Gohman, and Tumen 1987; Bruyako and Ostroverkhov 2004; Thornton and Schurr 2004). Since there are virtually no West Eurasian mtDNA lineages in the gene pools of northern and southern Han Chinese (Yao et al. 2002; Kivisild et al. 2002), Koreans (Horai et al. 1996; Derenko et al. 2004; Lee et al. 2006), or Japanese (Horai et al. 1996; Kivisild et al. 2002; Tanaka et al. 2004), Mongolia can be considered the eastern border of the distribution of maternal lineages that originated in West Eurasia.

As indicated above, relatively few studies have explored the genetic affinities of Mongolian ethnic groups. The ethnolinguistic and anthropological affiliations of groups that contributed to the formation of the Mongolian population are still unclear because numerous ethnic groups (including the Xiongnu, Syanbi, Jujan, Uighurs, and Kidans) could have been involved (Nyambuu 1992; Badamkhatan 1987; Bulag 1998; Morgan 2007). In fact, as noted above, the dominant ethnic group, the Khalkha, may have formed as a result of the admixture of different tribes that diverged only recently, perhaps ~300 years ago.

To investigate this question, Changchun et al. (2006) examined the mtDNA HVS-I sequences from sixteen Tuoba Xianbei remains excavated from Qilang Mountain Cemetery in Inner Mongolia and compared the resulting data with those from surrounding populations⁸ (Fig. 7.14). Interestingly, the mtDNAs of the Tuoba Xianbei ethnic group belonged mainly to haplogroups C (31.3 percent) and D (43.8 percent) and showed the closest genetic similarity to populations of Oroqen, Mongols, and Ewenki (Evenki). This result was not surprising given the high frequencies of these same maternal lineages in Tungusic-speaking populations from eastern Siberia and the Amur River region (Pakendorf et al. 2003; Schurr and Wallace 2003; Starikovskaya et al. 2005) and East Asia (Schurr et al. 1999; Kivisild et al. 2002; Yao et al. 2002; Kong et al. 2003). However, the Xianbei data came from a small sample that probably does not represent the total genetic diversity that was once present in this population, especially since the Xianbei state is known to have incorporated numerous non-Han groups during its reign (Changchun et al. 2006).

Genetic Diversity of Mongolian Ethnic Groups

Recent molecular genetic studies have begun to reveal the nature and extent of diversity of different ethnic groups within Mongolia. The mtDNA



7.14. Map showing the expanse of the Xiongnu Empire (modified from <http://en.wikipedia.org/wiki/File:Hsiung-nu-Empire.png>).

composition of most ethnic groups consisted of maternal lineages commonly seen in Siberia and East Asia, such as A, B, C, D, F, G, and others (Gokcumen et al. 2008; Pipes, Labuda, and Schurr 2008). However, some of these groups, in particular the Khoton, had low to moderate frequencies of West Eurasian lineages (e.g., H, J, and K), which are not commonly observed in East Asian populations, even at low frequencies. The largest ethnic group in Mongolia, the Khalkha, was more variable with respect to mtDNA haplogroup diversity compared with the other smaller populations, perhaps because of its recent complex origin (Badamkhatan 1987; Nyambuu 1992; Bulag 1998) and was also significantly genetically different from the Khoton and Zakhchins.

Similarly, recent Y-chromosome data have provided insights into the genetic relationships among different Mongolian ethnic groups (Katoh et al. 2005a,b). As noted above, haplogroup C*(x C3c) and its subhaplogroup C3c are the most common lineages in Mongolians, being seen commonly in the Khalkha, Uriankhai, and Zakhchin, but at much lower frequency in the Khoton. Between the two haplogroups, C*(x C3c) is more frequent in Khalkha and EAS populations (Northern Han, Korean Chinese, Korean, and Japanese), while C3c is more frequent in the Uriankhai, Zakhchin, and

Khoton from West Mongolia. Haplogroup D, which occurs at high frequencies in Japanese (Hammer and Horai 1995; Tajima et al. 2002), is present at very low frequencies in these groups. In addition, haplogroups F*(xJ,K), J, N3, and P appear at lower frequencies in Mongolian populations—and to a greater extent in Khalkha, Zakhchins, and Khoton—than other populations. By contrast, the common West Eurasian haplogroup R1a1, which may have been brought to Mongolia during the expansion of early nomadic groups in Central Asia, occurred at a very high frequency in the Khoton population and much lower frequencies in the Khalkha, Uriankhai, and Zakhchin.

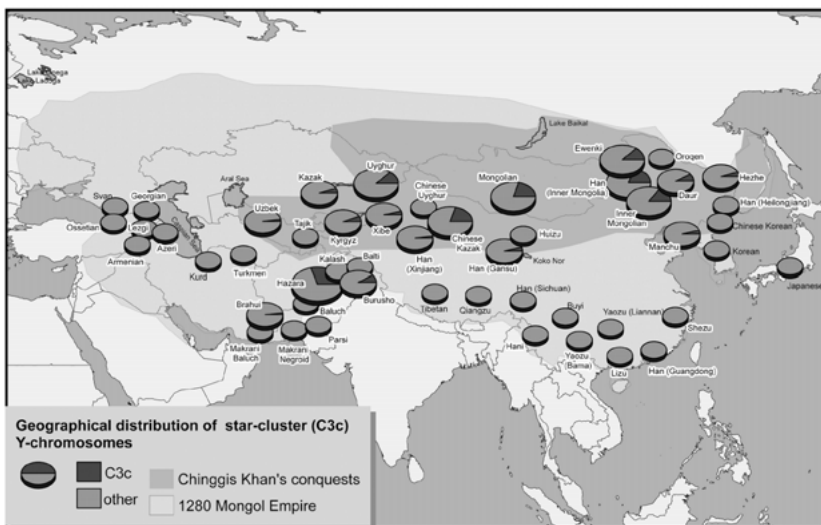
Overall, the distribution of male genetic lineages indicated that the Khalkha, Uriankhai, and Zakhchin were genetically similar to both EAS and Siberian populations. By contrast, the Khoton showed affinities with CAS populations based on both mtDNA and Y-chromosome data (Katoh et al. 2005a,b; Pipes, Labuda, and Schurr 2008) and may have arisen from a population of Turkic origin that recently migrated into the present territory of Mongolia (Batsuuri 1977; Nyambuu 1992). Interestingly, an earlier analysis of X-chromosome variation in Mongolian groups revealed strong similarities between the Uriankhai and Zakhchin populations (Katoh et al. 2002). These data, in the context of historical and ethnographic evidence (Nyambuu 1992), point to genetic admixture as the source of this close genetic relationship.

Legacy of Chinggis Khan

Chinggis Khan left a lasting political and cultural impact on Eurasia through the rapid expansion of his empire across a vast area in a matter of several hundred years. Beginning in eastern Mongolia, he and his male relatives led armies that conquered much of Central and East Asia, including the Western Xia in northern China and Khwarezmid Empire in Persia, ultimately expanding to Anatolia and Central Europe (Spuler 1971, 1989, 1994; Morgan 2007). As a result of their expansive and prolonged rule during the reign of the Mongol Empire, Chinggis Khan, his sons, and his grandsons also appear to have made a significant genetic impact on this region. This interpretation is affirmed by the pattern of genetic diversity in subhaplogroup C3c which, based on STR haplotype variation, was estimated to have originated in Mongolia some 1,000 years ago (Zerjal et al. 2002, 2003). In addition, approximately 8 percent of the men (~16 million individuals) from sixteen Asian populations living within the former Mongol Empire carried this unique Y chromosome lineage or closely related types (Fig. 7.15). The

historically documented events related to the Mongol Empire also support the genetic evidence, which suggests that this paternal lineage was spread by Chinggis Khan and his male kin as they established a long-lasting male dynasty (Zerjal et al. 2002, 2003).

Further evidence for the genetic impact of the Mongol expansion can be seen in the Mongolian-speaking Kalmyks, an ethnic group that descended from those Oirat who migrated to the lower Volga River in Russia during the Mongolian Expansion (Katzner 1986; Nyambuu 1992; Janhunen 2003; Nasizde et al. 2005). For approximately 300 years, the Kalmyks have lived interspersed among other ethnic groups, which were the original settlers of the region. However, genetic research has begun to show their connections to populations living farther to the east. For example, polymorphisms of glutathione S-transferases M1 and T1 (GSTM1 and GSTT1) revealed similarities between Kalmyks and Buriats (Popova et al. 2002). Likewise, classical genetic markers showed the Kalmyks to have close affinities with Buriats of the Baikal region and with Mongolians (Novoradovsky et al. 1993; Shimizu et al. 2006). A later analysis of mtDNA and Y-chromosome variation



7.15. Geographical distribution of star-cluster (C3c) Y-chromosomes. Populations are shown as circles with an area proportional to sample size; star-cluster chromosomes are indicated by gray (red in the DVD version) sectors. The shaded area represents the extent of Chinggis Khan's empire at the time of his death (1227) (Morgan 2007; Zerjal et al. 2003).

in Kalmyks also suggested their generally close relationship with Mongols (Nasidze et al. 2005). Interestingly, the NRY haplotypes associated with subhaplogroup C3c in Kalmyks were almost identical to those reported in Mongols, with the high frequency (31.3 percent) of duplicate alleles for the *DYS19* locus being associated with the “Chinggis Khan” Y-chromosome STR haplotype (Zerjal et al. 2003). Thus genetic data clearly demonstrate biological links between the Kalmyks and Mongolian populations.

SUMMARY OF MONGOLIAN BIOGENETIC DIVERSITY

In this chapter, we have reviewed and summarized a number of different data sets from Mongolian, Siberian, and EAS populations in an effort to map the biological affinities of Mongols. What emerges from this analysis is the observation that the biogenetic diversity seen in Mongolian populations is layered, or stratified, and reflects the long human occupation as well as several major prehistoric and historic expansions of human groups across the region. While more recent events, such as the expansion of the Mongol Empire, have left a clearly identifiable genetic imprint on populations outside of the region, they are only the most recent to do so.

In general, there is a significant East Asian genetic background for most Mongolian groups, due to the early expansion of human populations into the region. Mitochondrial DNA and Y-chromosome data, as well as archaeological evidence, suggest that Northeast Asian groups formed through the mixing of ancient CAS and EAS populations by around 30,000–40,000 years ago. The populations that initially settled the region probably possessed craniofacial traits similar to populations across northern Eurasia and dental traits similar to Sundadont (ancestral) features seen in SEAS populations.

In the Late Pleistocene, perhaps due to changing climatic conditions associated with the Last Glacial Maximum (LGM), the cranial traits linked to the “Mongoloid” racial type and the dental traits characterizing the Sinodont dental complex arose in Northeast Asia, probably 20,000–15,000 years ago. Populations bearing these physical traits began expanding across Northeast Asia and eventually to the Americas, meeting populations bearing ancestral features in East Asia. This dispersal of Mongoloid groups led to the emergence of regionally differentiated populations in different parts of East Asia.

In the post-LGM Early Neolithic, populations re-expanded into the formerly glaciated areas (e.g., Forster 2004), including the northern reaches

of Eurasia. These expansions brought both older lineages representing the earliest colonization of the region and newer lineages that evolved from them (e.g., mtDNA haplogroups G, Y, and Z and NRY haplogroups N3 and R1a1). It also appears that the regional gene pools now distinguishing different parts of Northeast Asia (Amur-Okhotsk, Southeast Asia, East Asia, Altai-Sayan, etc.) began to take form at this time, based on the ages of the genetic lineages seen commonly in those areas (e.g., Schurr et al. 1999; Schurr and Wallace 2003; Starikovskaya et al. 2005).

Later, the invention of agriculture in Southeast Asia led to significant demographic growth of populations there, as well as their expansion north out of East Asia. This expansion may be reflected in the distribution of both mtDNA and Y-chromosome haplogroups in NEAS and EAS populations, which reveal the northward spread of mtDNA haplogroups B and F and NRY haplogroup O3, in addition to the influx of SEAS craniofacial and dental traits into Korea and Japan.

The last major influence on the biogenetic makeup of ancestral Mongol populations appears to have been the influx of ancient Indo-European steppe nomads into Central/East Asia. The steppe nomads brought with them mtDNA and Y-chromosome haplogroups that were not indigenous to East Asia, as well as “Caucasoid” cranial types seen in Bronze and Iron Age burials in Central Asia and western Mongolia.

The contact and merger of eastern and western populations produced the gene pool that is seen in today’s Mongolian populations. This gene pool has largely persisted over the subsequent 3,000–4,000 years, despite the rise and fall of the Xiongnu and Xianbei Empires in the region and the struggle for control of this region by nomadic tribes and Han Chinese dynasties. The later expansion of Turkic speakers and rise of the Mongol Empire led to the spread of genetic lineages formerly confined to East Eurasia into Central and West Asia, along with the dispersal of some of the West Eurasian lineages acquired through admixture with ancient Indo-European populations in the previous millennia (e.g., Wells et al. 2001; Zerjal et al. 2002).

To further clarify the patterns of genetic diversity in Mongolia, it would be useful to analyze more expansively the DNA markers described above in the different ethnic groups present in the country. These data would complement ethnographic, historical, and linguistic evidence about the origins of these populations. The acquisition of more ancient DNA data from archaeological populations in Mongolia and adjacent territories would also

help elucidate the biological and cultural affinities of the nomadic steppe tribes that dominated this region for the past several thousand years. Finally, the continued analysis of mtDNA, Y-chromosome and autosomal loci in Mongolians, as well as populations from across Northeast Asia, will expand our understanding of the process of biological differentiation that has occurred in ethnic groups from South Siberia, Central Asia, and East Asia.

CONCLUDING REMARKS— THE GENO-“SCAPE” OF MONGOLIA

As noted at the beginning of this chapter, the patterns of biological variation in Central and East Asia that can be ascertained from studies of osteological, dental, and genetic diversity reflect not only processes of evolution and adaptation but also social and political interactions and the exchange of ideas and technologies between populations living in the broad region that now encompasses the modern nation-state of Mongolia.

We have shown that this diversity is layered or interwoven, and a product of over 40,000 years of human occupation of NEAS. This diversity has resulted from population expansions, cultural interactions, and the rise and fall of empires, as well as changing climatic conditions creating new selective conditions that have taken place during this period. We are also able to glean details of the formation of the gene pool for modern Mongolians from the different biological data sets, and the same can be done for the linguistic and archaeological evidence (see Fitzhugh and Bayarsaikhan, and Honeychurch and Amartuvshin, in this volume) from this region. Thus, when attempting to understand the broad temporal dimensions of Mongolian history, an area studies approach is actually very useful, even if the specific area under consideration (NEAS, Central-East, Inner Asia, etc.) varies subtly depending on the biological data set being examined.

At the same time, there are clearly aspects of these demographic events that allow for an approach employing the “-scape” concept, particularly with regard to network analysis. We can literally see such networks in the Y-chromosome lineage that appears to have arisen in the ancestors of Chinggis Khan and was disseminated by his male relatives, and also the interactions of the Xiongnu and Mongol empires with Han Chinese and Manchu empires, as recorded in imperial Chinese documents. The colonization of Mongolia and Inner Mongolia by Russian and Chinese powers, respectively,

further altered the ethnic landscape of Mongolia, and often forced minority populations to move from nation-state to nation-state (as seen, for example, in the genetic data from Altaian Kazakhs [Dulik et al. 2008; Gokcumen et al. 2008; Dulik, Osipova, and Schurr, forthcoming]). In addition, there are deeper networks of connections implicated by the linguistic ties between Altaic-speaking populations from across Central and East Asia, the spread of nomadic pastoralism from west to east in the 2nd and 3rd millennia BCE, and the entry of Buddhism from Tibet in the 13th and 14th centuries. However, as indicated above, these networks extend beyond the immediate confines of Mongolia proper and indicate that Mongolian identity and history have emerged through interactions extending across northern Eurasia and into East Asia over many millennia. This perspective is clearly consistent with the view that researchers need to be freed from geographic, or territorial, constraints to trace interactions around the globe (Smart and Smart 2003:266), even prehistoric ones.

The relevance of this point can be seen in the representation of modern Mongolian identity, which, in part, is traced to the emergence of the Mongol Empire and rule of Chinggis Khan. The link to this imperial past is made obvious by the presence of statues of Chinggis Khan at the National



7.16 New façade of the Mongolian Parliament with statue of Chinggis Khan in the middle. (Photo courtesy of Theodore G. Schurr)

Parliament building in Ulaanbaatar (Fig. 7.16), and the use of his name on everything from soft drinks to snack shops in Mongolia itself. However, while Chinggis Khan may have literally been the father of his country in a genetic sense, the characteristic features of Mongolian biology and culture required many thousands of years of evolution, adaptation, and cultural innovation and exchange to take their current form. Thus, it is by examining the Geno-scape of Mongolia that these different facets of its history come into clearer focus.

NOTES

1. Some of the general characteristics associated with the Mongoloid racial type (crania) are very forward-projecting malar (cheek) bones and comparatively flat faces, large circular orbits, and a moderate nasal aperture with a slightly pointed lower margin. In other words, Mongoloid peoples tend to have a larger and more gracile braincase as well as a broader skull, broader face, and flatter roof of the nose than non-Mongoloid populations (Alexeev 1978; Alexeev and Gohman 1984; Alexeev and Trubnikova 1984).
2. Mongoloid traits are thought to have emerged within an area defined by the southern steppe regions of Transbaikalia, the central and eastern regions of Mongolia, and several regions of Northern China (Jin and Su 2000). These include the presence of an epicanthal fold, lack of brow ridges, shallow mandibular fossa, small mastoid processes, stocky build, later eruption of full dentition (except second and third molars), less hair, fewer sweat glands, and a long torso (Phillips 1969; Fiedel 1992).
3. Cranial metric (discrete) traits include the dimensions of the skull and face (e.g., bizygomatic breadth, head length, nasal height). The features of cranio-facial shape and size show a high heritability (e.g., Neves and Hubbe 2005), hence are under considerable genetic control. In addition, nonmetric traits on the skull and mandible, including canals, foramina (openings), toruses, grooves, and sutures, are also used to assess the biological affinities of human groups. These nonmetric traits have been successfully used by other biodistance studies involving human populations (e.g., Konigsberg 1988; Lahr 1996; Prowse and Lovell 1996; Ishida and Dodo 1997) and their scoring procedures and descriptions are well established (Hauser and DeStefano 1989; Buikstra and Ubelaker 1994).
4. Similarly, studies of protein and nuclear DNA markers show that Koreans have close genetic affinities with Mongolians among Northeast Asians (Goedde et al. 1987; Saha and Tay 1992; Hong et al. 1993). They were also closely related to the Japanese but somewhat more distant from the Chinese. These genetic data support linguistic evidence suggesting that the ancestors of present-day Korean populations have a common origin with NEAS groups from the Altai-Sayan and Baikal regions of southeastern Siberia (e.g., Kim 1970). By contrast, mtDNA variation indicates that the Koreans are more closely related to the Chinese and Japanese among EAS populations (Harihara et al. 1988; Horai et al. 1996; Schurr et al. 1999; Lee et al. 2006). Recent studies of Y-chromosomal diversity also show that the Koreans possess lineages originating in both Northeast and Southeast Asia (Kim et al. 2000; Karafet et al. 2001). Thus, the peopling of Korea appears to have been a complex process with an initial northern Asian settlement followed by several migrations, mostly from southern to northern China (Jin et al. 2003). This process also likely reflects the population history of much of Northeast Asia in general.

5. A haplotype is a unique combination of mutations or polymorphisms present in a mitochondrial genome. MtDNA haplotypes that share phylogenetically important mutations are assigned to a specific haplotype group, or haplogroup. The terms “haplogroup” and “lineage” are used interchangeably to denote a distinct cluster of phylogenetically related mtDNAs, while the smaller branches of a haplogroup are usually called “subhaplogroups” or “sublineages.”
6. When referring to mtDNA and Y-chromosome haplogroups or lineages, we use the terms “East Eurasian” and “West Eurasian” to refer to their putative geographic origin. Here, East Eurasia encompasses the region including Northeast, East, and Southeast Asia, whereas West Eurasia extends from Europe to the Near East and West Asia. Central Asia appears to be a crossroads where populations bearing these different lineages came into contact and contributed to their genetic makeup.
7. A haplotype is a unique combination of mutations or single nucleotide polymorphisms (SNPs) present in the nonrecombining region of the Y-chromosome. NRY haplotypes that share phylogenetically important SNPs are assigned to a specific haplotype group, or haplogroup. The terms “haplogroup” and “lineage” are used interchangeably to denote a distinct cluster of phylogenetically related Y-chromosomes, while the smaller branches of a haplogroup are called “subhaplogroups” or “sublineages.”
8. The Xianbei (Dongbei or ancient Manchus) were a nomadic people in ancient China that succeeded the Xiongnu and included a sizeable federation of non-Han groups. They first became a significant part of Chinese culture during the Han Dynasty, when they occupied the steppes in Manchuria and Eastern Mongolia (Changchun et al. 2006). After the fall of the Han Dynasty, the Xianbei formed a number of empires of their own, including the Yan Dynasty, Western Qin, Southern Liang, and the Northern Wei (see Honeychurch and Amartuvshin, this volume).

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