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Unraveling the population history of the Xiongnu to explain molecular and archaeological models of prehistoric Mongolia

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UNRAVELING THE POPULATION HISTORY OF THE XIONGNU TO EXPLAIN MOLECULAR AND ARCHAEOLOGICAL MODELS OF PREHISTORIC MONGOLIA

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This dissertation explores the prehistory of Mongolia during a time when nomadic tribes created the world's first steppe empire in Inner Asia. These aggregated tribes, known to Chinese historians as Xiongnu, ruled from the 3rd century BCE to the 2nd century CE. They came to define steppe polity construction later used by the Mongol Empire under the reign of Chinggis Khan. These nomads moved extensively over the eastern steppe and interacted, both in trade and intermarriage, with peoples from southern Siberia to Xinjiang. However, the Xiongnu as a people are relatively unknown to scholars since they did not possess a written language of their own.

Although analysis on ancient skeletal remains of the Xiongnu have opened new avenues of research into their origins, scholars still do not have a comprehensive understanding of these ancient nomads. This study makes an attempt to elucidate questions of the Xiongnu's history and biological structure by examining craniofacial diversity using a methodology known as geometric morphometrics. Using a suite of multivariate statistical analyses to explain group relationships within and among the Xiongnu to groups in the region, this study explains the origins of the Xiongnu in a biological context and makes inferences about genetic exchanges. A quantitative genetic model is used to test group relationships and infer levels of gene flow between groups.

Results indicate the Xiongnu were composed of at least two biologically distinct groups. One sample from an elite cemetery in northern Mongolia shares their ancestry with a Bronze Age population from Mongolia, and possibly, to a later migration of Turks, who came to dominate the eastern steppe between the 6th and 8th centuries CE. The Xiongnu also evidence biological similarity with nomads who composed the Mongol Empire, modern-day Mongolians, and some Siberian groups. These results are similar to genetic studies suggesting a mix of Eastern and Western Eurasian haplogroups while also achieving consensus with models of steppe polity formation proposed by archaeologists, who suggest local ties to extra-local groups through interactive exchange networks. Overall, the Xiongnu nomads are very much a part of Mongolia's past with links to its modern peoples.

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CHAPTER 1

INTRODUCTION

Chinggis (Genghis) Khan (c. 1162 – 1227), founded the Mongol empire in 1206, in what would later become the largest contiguous land empire in known history (Morgan, 1986). Stretching from Eastern Europe to the Sea of Japan, the Mongols ruled and conquered a vast array of peoples, with as many different languages, religions, and cultures. Though the Mongol empire was known for rapid territorial expansion with brutal efficiency in their conquering abilities, they were not the first steppe empire to rule from horseback. In fact, the Mongols are just one in a long succession of polities that ruled over the vast Inner Asian steppe beginning in the second century BC. From around 200 BC onwards, Chinese historical records and more recent archaeological investigation indicate these small-scale societies that were scattered across the northeastern steppe aggregated into novel organizational forms as large-scale, hierarchically organized, integrated polities of pastoral peoples. These features come to define the Inner Asian zone of steppe history. This dissertation concerns the first of those steppe polities, a group of nomads that resided in what is now central and northern Mongolia, though at their height, were known to control a large territory that spanned the Xinjiang Province and Inner Mongolia in the west of China, the Baikal region of southern Siberia, and Kazakhstan at the westernmost geographic boundary.

These people, known as the Xiongnu, pronounced *Shung-nu*, (c. 209 BC – 3rd century AD), were the prototypical example of pastoral nomads who came to dominate and rule over a large swath of Inner Asia. As a confederation established at the end of the first millennium BC and disintegrated sometime during the 2nd or 3rd century AD, there

is still much that scholars do not know about Xiongnu origins and its people. As a non-literate society, much information about this group comes from historical sources written by the Chinese during the Qin (221 BC – 206 BC) and Han Dynasties (206 BC – 220 AD). However, more recent work by archaeologists, who have uncovered burial assemblages and artifact types, and molecular anthropologists who have extracted ancient DNA from skeletal remains, have contributed to our knowledge of the Xiongnu. Though much of what we do know archaeologically and genetically come from a mortuary and burial context, very little research has been undertaken using skeletal quantitative traits in an effort to answer questions surrounding their origin and regional population history.

This dissertation is an effort towards filling that gap in the Xiongnu's history. In this study I will employ a quantitative genetic model to understand microevolutionary processes of the Xiongnu by using a geometric morphometric approach to craniofacial shape variation coupled with morphometric multivariate statistical analysis. Ultimately, this dissertation seeks to answer the question of just who the Xiongnu polity were as a people by investigating their skeletal remains. Archaeologists and some physical anthropologists, through the convenience of artifact style and mortuary context, have labeled the Xiongnu an ethnic group, in a similar vein that we as modern people might associate ourselves with a particular group, such as German, Irish, or the more pan-ethnic Latino ancestry. These labels allow researchers to reconstruct their histories – both from an archaeological and biological perspective. However, these labels need to be scrutinized in order to better understand the origins and interactions of those groups in pre-history – especially those groups that do not provide a written language with inferences of self and/or group identity.

This dissertation asks several important questions of interest to both archaeologists and molecular anthropologists. If these people were composed of several diverse biological groups, will this be reflected in craniofacial morphology? If we detect an elevated level of diversity within the Xiongnu, is it feasible to use craniofacial morphology to tease out and identify those groups? Though complicated in an archaeological context, the use of some group identifier is necessary. If we construct this identity in terms in which a large, politically aggregated and opposed group came to be known as the Xiongnu, and whom the Chinese struggled against – that is, a politically cohesive unit – then we should be able to discuss this entity and its agents as a group in the archaeological record. Further, we should be able to biologically test those individuals who composed this political unit. That is, we could ask: Was there more than one biologically distinct people who composed the Xiongnu political entity if they were originally diverse tribes inhabiting the eastern steppe? And, will this diversity be detected through craniofacial morphology?

In addition to potential multiple biological histories of the Xiongnu, who did they interact with biologically? Though there is substantial historical (Christian, 1998; Beckwith, 2009) and archaeological (Honeychurch and Amartuvshin, 2006; Wright et al., 2009) evidence for trade and exchange with local and regional groups, what was the extent of their biological interaction? What extent, that is, can we detect potential gene flow with groups in the region of Inner Asia? Were the Xiongnu interacting more strongly with groups from China, due to greater exchange or geographic proximity? Or, will we be able to detect a signature of gene flow from other regions, such as Central Asia or Siberia?

This dissertation has broad impact for both the population history of Mongolia and to researchers interested in understanding complex nomadic groups in both the present and in the past. If results show a significant affinity of the Xiongnu to modern Mongolian people, then the Mongolian people can call these steppe nomads ancestors in the same way they trace their biological continuity to Chinggis Khan and the Mongol Empire of the 13th century. The greater academic community is impacted by both the collection and dissemination of geometric morphometric data on nomadic groups and the utilization of population genetic models of population history and structure. The inclusion of skeletal data is essential in these endeavors.

In addition, this research is significant to furthering the use of quantitative data as a valid and informative way to explain group relationships in the past. Though there are limitations and biases in the data, the use of quantitative characters has informed and framed some of the most important questions in human evolutionary history. Ancient DNA has become more important to answering some of these fundamental questions, as the ability of researchers and speed of technology have improved exponentially over the last several years. However, many of these studies are limited in scope to a few individuals or samples, and rarely do they achieve the scale required to answer questions of population history and structure at the regional level (though intensive research in some areas, such as Native American population history, is indeed promising). There is also the question of sample destruction. Most analyses of ancient DNA require the use of a significant bone or tooth sample (though see Bolnick et al., 2012), which could be detrimental to future researchers. The research presented in this dissertation is non-destructive and available for future use. That is why, for now, the use of quantitative

characters are the foremost accessible way to understand regional population history and biological structure in a non-destructive context.

Defining Population History and Structure

In order to potentially differentiate a single biological group using skeletal data, we need an appropriate analytical methodology that has been tested and shown to be informative in the assessment of population history and population structure. Since the terms population history and population structure will be used extensively throughout this dissertation, they should be defined here. Both of these terms have at their core an element of human biological variation that attempts to determine genetic, or biological, similarity. Using some estimate of genetic or biological distance, we can then begin to ask questions relevant to a population's history, or structure. Distances can either be calculated as quantitative measurements, such as metric characters of the skeleton, or discrete, such as nonmetric traits of the skeleton, or DNA markers (mitochondrial DNA, the Y-chromosome, single nucleotide polymorphisms to name a few). This dissertation uses biodistance (craniofacial variation) as a proxy for genetic (DNA) variation. This justification, though controversial, does have empirical evidence to support such as claim.

The underlying theoretical framework of biodistance (or genetic distance) analyses is the observation that populations that exchange mates become more biologically (genetically or phenotypically) similar over time, while those that do not become more dissimilar at a rate determined by their effective population size (reviewed in *Chapter Two*). There are several assumptions for using biodistances to reconstruct population history and structure. These include: *1)* mutation and selection are held constant in order to explain the effects of genetic drift and gene flow for geographically

proximate populations who share similar environments; *2)* the skeletal samples used are not naturally occurring populations, but rather are temporally ordered lineages; *3)* changes in allele frequencies result in measurable changes in the phenotype that can be characterized in some mathematical manner; *4)* environmental effects on phenotypic variation are randomly distributed among the samples being studied; and *5)* inheritance of phenotypic variation is additive (due to the action of multiple genes with small effect on the phenotype) and resemblance among relatives is strong (Stojanowski and Schillaci, 2006).

Of course, these assumptions (especially assumptions 3 and 4) are controversial. Evolution at the genetic level could proceed without noticeable changes to the phenotype, as well as having developmentally plastic, or epigenetic changes occur at the phenotypic level without a great effect at the genetic level (Hallgrimson and Hall, 2011). This is especially true for the human head (Lieberman, 2011a). Several studies have shown that assuming an underlying equal and additive effects model for quantitative trait evolution produces similar estimates to neutral genetic evolution in terms of genetic similarity (Relethford, 2004b; Manica et al., 2007; Betti et al., 2009, 2010; Strauss and Hubbe, 2010). An even more recent study has suggested extensive evidence for a strong correlation between genetic and biological distance matrices (Martinez-Abadias et al., 2012). These authors used the Hallstatt ossuary, located in Austria and having extensive genealogical information, to test for pervasive integration in the human skull (the functioning or constraint of organismal form – in this case, the human skull). Because of the genealogical information associated with each crania, the authors were able to directly estimate the genetic covariance matrix for cranial shape and found that the

phenotypic covariance matrix is highly correlated, indicating that the use of phenotypic covariance structure can be used as a proxy for genetic estimates of similarity and dissimilarity. Therefore, the environment, though important in many studies of craniometric evolution and development, is treated here as a nuisance parameter, and I treat biological and genetic distances equally throughout this dissertation. Biodistance can be computed and represented visually in a number of ways and are discussed further in *Chapter Five*.

Population structure is primarily concerned with those factors affecting mate choice, the genetic relationships between individuals within a population, or subdivisions within a population. Various cultural, demographic, and ecological factors contribute to mate choice, which in turn invariably affects the genetic distances between individuals and groups. What is most important are those reasons that limit or enhance gene flow in a population. *Population history* is a related theme, however, what most concerns researchers are those factors that affect the genetic impact of historical circumstance, such as invasion, migration, and other events that might affect the genetic exchange between populations. In other words, those historical factors that might affect the biological distance between populations.

For this dissertation I will explore the population structure of the Xiongnu by examining the mainly demographic factors, such as the geographic distance between groups, and the population history of the Xiongnu by examining those interactions (potential gene flow) from long-distance migration and invasion, or through cultural contact with groups residing in China, Central Asia, and Siberia. These analyses will be carried out using both an indirect (multidimensional scaling, biological distance matrices)

and direct approach (Relethford-Blangero quantitative genetic model, R-matrix, model-based clustering methods). I will restrict my interpretation of Xiongnu population history and structure to biological quantitative characters in light of genetic and archaeological research.

Craniofacial Variation, Geometric Morphometrics and Samples

In an effort to define the origins of the Xiongnu and gain insight into its population history and structure, I will use craniofacial variation as a proxy for genetic variation. The analysis of ancient populations through the investigation of skeletal features is an effective and informative way to understand modern population structure and infer relationships in the past. It has been used extensively in the anthropological literature, and I expand on this in *Chapter 6*. Briefly, craniofacial morphology has been used to assess patterns of human variation in both a bioarchaeological and forensic context, test hypotheses concerning the emergence of modern human origins, and reveal evolutionary relationships among groups (Relethford, 1994; Relethford and Harpending, 1994; Hanihara, 1996; Steadman, 2001; Hennessey and Stringer, 2002; Hanihara et al., 2008; Harvati et al., 2010; Hubbe et al., 2011).

A brief mention of how I define group in this study is necessary. Throughout this dissertation, I use the word population or group to denote a specific skeletal sample in the data. However, this is not entirely appropriate as skeletal samples are not truly meaningful biological units as described by Mayr (1963). Cadien et al. (1974) brought this issue up many years ago to address methodological and statistical interpretation of skeletal remains. In their review, these authors pointed out that the evolutionary unit under analysis in skeletal samples is not the population, but a lineage. Since skeletal

samples tend to come from a diverse set of temporal circumstances, many (if not most), of these individuals did not have the opportunity to mate, and therefore, cannot in and of itself be called a biological breeding population. Rather, the skeletal sample that is temporally ordered in some manner with presumed genetic continuity is called a skeletal lineage. These skeletal lineages are not operating under different microevolutionary processes, but rather are the product of such microevolutionary processes.

When comparing skeletal lineages, we are unable to use statistical properties similar to how we treat populations. Such biologically relevant parameters including means, variances, frequencies, etc, are incomparable and should not be expected to provide reliable estimators of any single population. Therefore, the lineage is not often an approximation of the population. However, it has been shown by a number of authors in both theoretical and methodological advancements (see Buikstra and Beck, 2006) that the analysis of temporally-ordered skeletal series can give significant insight into historical demographic processes, pathology, or evolutionary processes among human groups in the past. In this dissertation, the skeletal samples used are referred to as either groups or populations, knowing full well that they are not entirely representative of the living population from which they came. I strive to maintain that all interpretations made using these skeletal samples are for those biological lineages, and not a complete representation of the now living population, which may, or may not, be considered the descendants of these skeletal lineages.

In this study, I use geometric morphometrics (GM) in order to study size and shape differences of craniofacial morphology. The advantages of using GM methods over more traditional methods of craniometrics are discussed in *Chapter 6*. GM has been

defined as the fusion between biology and geometry. It is a useful approach to quantitative characterization, analysis and comparison of biological form (Bookstein, 1991; Marcus et al., 1996). GM is a landmark-based method in the analysis of shape in bidimensional or tridimensional space (Bookstein, 1982; 1986), and there is a growing body for advanced statistical and graphical techniques in shape analysis. There are many advantages to using GM. Briefly, GM is a robust method to describe morphological trends and detect shape differences; provides a better visualization of shape over more traditional approaches; is becoming more accessible with the availability of hardware and software programs (many freely available via the Web); and data sampling is easier, more precise and efficient.

The data used in this dissertation comes from multiple sources. Craniofacial data were sampled from human populations over several time periods. Notably, Xiongnu crania were sampled from two locations in Ulaanbaatar, Mongolia: the National University of Mongolia and the Mongolian Academy of Sciences, Institute of Archaeology. Other Mongolian crania were sampled from skeletal collections housed at the American Museum of Natural History (AMNH), New York, NY; Musee de l'homme in Paris, France; and Moscow State University, Moscow, Russian Federation. I have also sampled crania from China at Jilin University, Changchun, China; from Central Asia at Musee de l'homme; from Europe and Africa at the AMNH; from Japan at the University of Tokyo and the Museum of Nature and Science, Tokyo, Japan; and from Siberia at the Institute of Archaeology and Ethnology, Russian Academy of Sciences Siberian Branch, Novosibirsk, Russian Federation and Moscow State University.

Hypotheses Tested

This study seeks to test various archaeological models of steppe polity formation through the inclusion of biological data. The use of quantitative data will be used to interpret biological evidence of Xiongnu population history and structure. Various indirect methods will be used to construct population relationships. These include principal components analysis, distance-based methods, and clustering methods to visualize group relationships. Craniofacial data will be used to test whether the Xiongnu polity arose as a consequence of its relationship to a core state, such as China, or was formed *in situ* as a consequence of mobile technology. The extent of interaction with China and other regional groups for essential resources will be tested as evidence of gene flow should be reflected in the quantitative data.

Using a model of population genetics for quantitative characters (Relethford and Blangero, 1990), I will test the hypothesis for greater than expected gene flow occurring during the Xiongnu period as a consequence of interaction. If nomadic groups were self-sustaining and had little or no biological interaction with the sedentary people of China (or other peoples of Eurasia), then we should see biological continuity among Mongolian nomadic groups through various temporal periods. That is, a biological continuity evidenced in craniofacial diversity among Mongolian groups would be the result with little to no degree of gene flow from China or elsewhere.

Though more difficult to test, this dissertation will seek to test within-group variability of the Xiongnu polity. This is made more difficult due to insufficient contextual information of each individual attributed to Xiongnu material culture. If all of the individuals who composed the Xiongnu came from a similar 'ethnic' group, then the

observed morphological diversity will be low. If these individuals came from diverse biological geographic origins, then the Xiongnu will show higher morphological diversity, and possibly multiple biological groups.

Chapter Organization

Several chapters lay the theoretical foundation for this dissertation. *Chapter Two* extensively discusses population genetic and quantitative genetic theory. As quantitative genetic theory is built upon the foundation of population genetic theory, an exhaustive review, including historical origins and fundamental concepts of population genetics, are introduced in Chapter Two. The concepts of polymorphism, genetic drift, gametic disequilibrium, and natural selection are covered. In addition, chapter two covers craniofacial development. This is necessary in order to understand why the human skull is so integral in evolutionary studies of population history and why the human head has been studied, prodded, measured, and poked by evolutionary biologists and physical anthropologists alike for so many years (Lieberman, 2011a).

Chapter Three gives background information pertaining to the origin of modern humans, the number of dispersals from Africa, the colonization of East Asia and Siberia, and migration studies related to groups residing in Eurasia. Fundamental to Chapter Three is an understanding of the various genetic systems used to study human migration (gene flow) in modern and ancient populations. This includes a brief discussion of mitochondrial DNA, the Y chromosome, autosomal DNA such as single nucleotide polymorphisms, genome-wide studies, and ancient DNA. These genetic systems are reviewed in light of the quantitative characters that help to better understand human origins and migration.

Chapter Four is an introduction to Xiongnu population history from an archaeological and genetic perspective. In this chapter, a brief discussion of the ethnicity of the Xiongnu is given, followed by the evidence of material culture, mortuary studies, and genetic research that has gone into defining these prehistoric nomads.

Chapter Five discusses the materials used in this dissertation and the landmarks used to define craniofacial morphology. As mentioned above, samples used in this dissertation come skeletal collections housed in several countries. Most of the crania sampled are considered to be 'modern', that is within the last two centuries. As the focus of this dissertation is on a group that arose during the Late Bronze Age and Early Iron Age of Mongolia (~2500 years before the present), I have sampled various temporal periods for inclusion in the analyses. These include the Chinese and Japanese Neolithic; the Bronze Age of China, Mongolia, and Siberia; and the Iron Age of China and Siberia. Unfortunately, all of the samples from Central Asia are modern, making temporal comparison more difficult to assess. The landmarks used in this dissertation are homologous between specimens and are well-defined. Importantly, the biological landmarks quantify overall size and shape differences between individual and group crania, and have been shown in previous studies to be effective in the analysis of human population history and structure.

Chapter Six outlines the analytical methods employed during the course of this dissertation. Importantly, I introduce the concept of craniofacial variation and its utility in studies of population history and structure; the concept of geometric morphometrics as applied to studies of biological form and variation, as well as a discussion of significant literature applied to craniofacial evolution and development; the use of quantitative

genetics and some fundamental concepts applied to quantitative trait variation, including the R-matrix approach and the model of Relethford and Blangero (1990); statistical approaches to the analysis and representation of biological distance, including canonical variate analysis, principal component and principal coordinate (eigenvector) analysis, and cluster analysis; and lastly, regression and distance matrix correlation analysis.

Chapter Seven outlines the results from the data. Results are discussed in a hierarchical manner. That is, I first test within-group variability of the Xiongnu for the validity of further aggregation of the sample, then compare the Xiongnu and various other Mongolian samples to a world-wide analysis of all groups, including samples from Europe and Africa. I then compare the Xiongnu and Mongolian samples separately to three regions: China, Siberia, and Central Asia. This is done in order to assess population relationships on a more localized level, in an effort to test hypotheses of population history of the Xiongnu in a quantitative genetic context.

I conclude this dissertation with *Chapter Eight*, which focuses on discussing my results in both a quantitative and genetic context by comparing the results to the literature, in addition to discussing future directions of research.

CHAPTER 2

POPULATION GENETIC THEORY, QUANTITATIVE GENETICS & CRANIOFACIAL DEVELOPMENT

Any scientific undertaking within the field of biological anthropology is grounded in evolutionary theory. Naturally, the concept of evolution - the change in inherited traits in a population of living organisms over successive generations - drives biological anthropological investigation, whether the pursuit is the understanding of human biological diversity or the reconstruction of our hominid ancestors through the analysis of the fossil record. The transformation of physical anthropology in the 1950's brought significant changes to the prevailing paradigm as researchers shifted from a typological and static view of human evolution to one ultimately concerned with the expression of natural processes and change over time. Although anthropologists today are still concerned with the *physical* variation of morphological traits and characteristics, a greater interest and understanding has been gained from the field of evolutionary genetics and the molecular age of complete genome sequencing.

In this chapter, I will explore the history of human evolution from a molecular and population genetic point of view. In its simplest terms, population genetics is the study of how evolution works as a genetic process in natural populations (Crow and Kimura, 1970). Though the scope of this dissertation falls into the realm of quantitative genetics (Falconer and Mackay, 1996; Lynch and Walsh, 1998), it is imperative to grasp how the field of population genetics has contributed to the study of human evolution and variation. This chapter is primarily concerned with explicating those mechanisms affecting human evolution and variation, such as selection, genetic drift, mutation, and

migration. Each of these microevolutionary processes will be investigated and reviewed in light of population genetic theory. I will also review how these mechanisms affect evolution.

How much has selection affected current diversity as found in modern human populations from throughout the globe? How much can be attributed to a neutral, or nearly neutral model of evolution? To answer these questions, we need to start with an appropriate historical context from which to discuss the role of population genetics' and molecular biology's contribution to the field of biological anthropology. In 1951, Sherwood Washburn published "The New Physical Anthropology", a now seminal article in physical anthropology (Washburn, 1951). In the article, Washburn discussed the future of physical anthropology.

Washburn suggested that no longer should anthropologists be concerned with meticulous measurement, calculating indices, or defining type specimens for static classification. The new physical anthropology should be concerned with mechanisms of evolutionary change and adopt a broader and more dynamic perspective. Like Lewis Binford (1962) a decade later, descriptive and speculative methods needed to be replaced with an emphasis on problems, processes, and means by which to test those problems. Just as archaeology ten years later would see a paradigm shift (Kuhn, 1970), so too would physical anthropology.

Population Genetic Theory I: Detecting Variation

Since the rediscovery of Mendel's work in 1900 by de Vries, Comes, and von Tschermak (Provine, 1971), evolutionary geneticists began to place stronger emphasis on biological change through time. In years following Mendel's rediscovery, a heated and

virulent debate was still under way between two camps, known as the Bateson's "Mendelians" and Pearson's "Biometricians". Their focus was on continuous versus discontinuous inheritance and the roles of Mendelian heredity and natural selection on the impact of evolution. The Mendelians essentially discounted the importance of natural selection, while the biometricians placed too heavy an emphasis on statistical competence.

It was not until the 1930's that Mendelian inheritance was finally incorporated into evolutionary theory in what is known today as the "Modern Synthesis" (Hartl and Clark, 2007). This synthesis was achieved through an understanding that mutation rate and genetic variation does not dissipate through the mechanism of heredity, but is actually preserved and accumulated by the Mendelian mechanism of inheritance. The future development of population genetic theory was thus clouded for many years while Bateson and Pearson antagonistically pulled one another's hair out. Fortunately, for evolutionary biology, a lieutenant of Bateson, R.C. Punnett, asked mathematician G.H. Hardy about a simple Mendelian problem of inheritance. Punnett's concern was why in a random-mating population the dominants did not over time drive out the recessives (Punnett, 1911; Edwards, 2008).

If the two camps had paid closer attention to Mendel's original work (or to Yule, 1902), they would have found that the Law of Segregation clearly answers this dilemma. Segregation does not depend on the segregates, nor does dominance or a randomly mating population have anything to do with it. Mendel's experiments with selfing showed that the expected genotypic frequencies of the offspring of two parents are equal to the frequencies of those genes in the parents themselves. Provided equal variance in

sexual reproduction, no change in gene frequency will take place from one generation to the next. The importance of segregation can not be underestimated in the development of population genetic theory. In fact, it is the framework in which population genetics operates. The mechanism of Mendelian segregation is a highly regular process with strong geometric and algebraic overtones (Felsenstein, 2009). In addition, segregation occurs whether or not selection is operating, or whether or not mutation and migration are present.

Hardy-Weinberg Proportions: Hardy's simple law (Hardy, 1908) of equal allelic proportions was derived independently of Wilhelm Weinberg (Weinberg, 1908), and as such the Hardy-Weinberg Law of Equilibrium was deduced, although neither Hardy nor Weinberg used the words law or equilibrium in their publications (Edwards, 2008). It should be noted that some believe the work of Castle (1903) deduced the rule that later became known as the "binomial square rule". However, in his work, Castle (1903) did not explicate the rule for offspring in the next generation. His concern was with calculating selection generation-by-generation, and only gave results for the percentage of dominants in the population, not the genotypic frequencies of the populations, nor any rule stating, in the absence of selection, that genotypic frequencies would remain stable over subsequent generations (Edwards, 2008). Therefore, Castle did not derive the 'law' independently later verified by Hardy and Weinberg.

The problem of equal allelic proportions seemed so obvious to G.H. Hardy he wrote "I should have expected the very simple point [of genotype frequencies] which I wish to make to have been familiar to biologists." (Hartl and Clark, 2007:52). In fact, in his peroration to *A Mathematician's Apology* (1940), Hardy wrote, "I have never done

anything ‘useful’. No discovery of mine has made, or is likely to make, directly or indirectly, for good or ill, the least difference to the amenity of the world.” I believe G.H. Hardy would be surprised if given a perfunctory glance into a biology or anthropology classroom where population genetics is being taught.

Although simple in nature and trivially obvious, the Hardy-Weinberg “law” has clear and important implications. First, under the appropriate conditions, genotype frequencies can be predicted from gene frequencies. The second implication is that Mendelian reproduction in a random-mating population has no inherent tendency to favor one allele over another, or more explicitly, it will not tend to lose genotypic variability. This point is important because without variation, selection has nothing to act upon. By working within the domain of gene, rather than genotype, frequencies, the Hardy-Weinberg law simplified calculations that allowed geneticists to focus on the important perturbations that may be involved when populations are not in “equilibrium”.

Equilibrium is only possible in a restricted sense. A better way to describe this phenomenon would be to discuss a change (or lack thereof) in gene frequencies from one generation to the next as being in proportions. If we change gene frequencies of a population, there is nothing in the Hardy-Weinberg Law that will restore the frequencies to their original value. However, we can alter the genotype frequencies without altering Hardy-Weinberg proportions. The general equation for the Hardy-Weinberg principle is:

$$p^2 + 2pq + q^2 = 1 \ ;$$

where p and q are the allele frequencies in the population.

To maintain these principles of equal proportion, one must make multiple simplifying assumptions. These include: *1)* random mating within the population; *2)* no

differential fertility of the genotypes (no selection); *3)* equal genotype frequencies among the sexes; *4)* no mutation; and *5)* no immigration or emigration so that all members of the next generation come from the present generation. There are also a few hidden assumptions. These include: *1)* non-overlapping, or discrete generations so that one generation gives rise to another, the parents do not reproduce again, and are no longer counted as part of the population; and *2)* an infinite population size that remains fairly constant.

As expected, in naturally occurring populations, these assumptions are almost never held. Only in very short-lived populations with simple life histories (such as annual plants) do we find the concept of non-overlapping generations applicable. Models that incorporate continuous time (overlapping) in generations are less tractable than discrete models, and are therefore not as often used (Felsenstein, 2009). Discrete generation models, however, are a useful approximation for organisms with more complex life histories, such as humans. Ultimately, what the Hardy-Weinberg law contributes to population genetic theory is a simple null hypothesis from which to test the likely perturbations in gene frequency.

Linkage (Gametic) Disequilibrium: Hardy-Weinberg maintains that alleles are often found in random association. If you have a gene with alleles *B* and *b* at frequencies p_B and p_b, respectively, where $p_B + p_b = 1$, the Hardy-Weinberg principle tells us that the genotype frequencies of *BB*, *Bb*, and *bb* are expected to be in proportions (i.e. $p^2 + 2p(1-p) + (1-p)^2 = 1$) provided mating is random. Thus, there is random association among the *B* allele and *b* allele. Often, however, alleles are not in random association. When alleles are in nonrandom association at two or more loci, it is called linkage

disequilibrium (LD), or gametic disequilibrium. Patterns of LD in human populations have informed migration histories (Plagnol and Wall, 2006), detected recent positive natural selection (Voight et al., 2006; Sabeti et al., 2007), and discovered distribution and evolution of recombination hotspots (McVean et al., 2004; Spencer et al., 2006). The term, however, is often a barrier to understanding as detecting LD does not ensure either linkage or a lack of equilibrium (Slatkin, 2008).

Linkage disequilibrium is important to the fields of evolutionary biology and human genetics because it provides information about past events and constrains the potential response to natural and artificial selection. When LD is found throughout the genome, it can reflect population history, the breeding system, and patterns of geographic subdivision. When found in specific genomic regions, LD can reflect the history of natural selection, gene conversion, mutation, and other forces that cause gene-frequency evolution. How these factors can cause LD between a particular pair of loci or in a genomic region depends on local recombination rates (Slatkin, 2008).

Measuring LD between alleles at two loci has been complicated and many definitions have been proposed, but all depend on the quantity:

$$D_{AB} = P_{AB} - P_A P_B ;$$

which is the difference between the frequency of gametes carrying the pair of alleles A and B at two loci (P_{AB}) and the product of the frequencies of those alleles (P_A and P_B). This was originally developed for alleles located on two chromosomes, but now is more conventional to refer to the same chromosome (Slatkin, 2008) where the allele pair AB is defined as the haplotype and P_{AB} is the haplotype frequency. This D value characterizes the extent to which two alleles are nonrandomly associated.

If D = 0, there is linkage equilibrium (LE) that has similar properties to the Hardy-Weinberg (H-W) proportions discussed above. The essential feature of H-W proportions is that equilibrium is reached in one generation of random mating. Any deviations disappear immediately and all departures mean something interesting is happening, such as extensive inbreeding or strong selection. LE differs in that the allele frequency equilibrium is not reached within one random generation, but in fact is affected by the rate of recombination. Therefore, D will decrease at a rate that depends on the recombination frequency, *c*, between two loci:

$$D_{AB}(t + 1) = (1 - c)\, D_{AB}(t) ;$$

where *t* is time in generations. Linkage equilibrium will eventually be reached, however, for loci that are strongly linked (> 0.5), it will occur slowly.

Many mechanisms of evolution can create linkage disequilibrium, such as selection, drift, population subdivision, population bottlenecks, inbreeding, gene conversions, and inversions (Slakin, 2008). The initial question that prodded research into LD was of natural selection and how linked alleles may affect reproductive fitness or how the response to selection on one locus might be accelerated or hampered by selection affecting the other. Prominent in this area of research is the concern with LD and its effect on long-term trends in evolution.

Kimura (1965), Nagylaki (1974), and others have shown that unless interacting loci are closely linked or selection is especially strong, recombination dominates and, to an extent, LD can be ignored. These theories support Fisher's (1930) depiction of natural selection gradually increasing the average fitness of a population. Selection alone can create LD when fitnesses are multiplicative, in that the average fitness of an individual

who carries the AB haplotype exceeds the product of the average fitnesses of individuals who carry just the A allele or B allele alone. This pattern is easiest to detect in diallelic loci in haploid organisms (Slakin, 2008) by examining the relative fitnesses of each gametic combination.

Genetic drift coupled with selection (*both discussed in greater detail below*) as well as drift alone can create LD between closely linked loci, with an effect that is similar to taking a small sample from a large population. If two loci are unlinked (linkage equilibrium) but are only sampled by a few individuals, some LD will be created. Drift interacting with selection has a surprising effect. Drift causes closely linked loci that are under selection to be slightly weakened, thus reducing the response to selection. This effect has been called the Hill-Robertson effect (Hill and Robertson, 1966; Felsenstein, 1974; Comeron et al., 2008). Notably, linkage between sites under selection will reduce the overall effectiveness of selection in finite populations. The most important find was by Felsenstein (1974), who was the first to recognize this effect and how it might have a role in the evolution of recombination and sexual reproduction. What he found was that the Hill-Robertson effect causes selection to be inefficient in purging deleterious mutations in a species with low recombination rate (Slakin, 2008). Therefore, natural selection tends to favor any mutation that increases recombination rates (Barton, 1995).

Selection tends to affect one or a small number of loci. By contrast, changes in population size, subdivided populations, and gene flow affect LD throughout the entire genome. Genome-wide patterns of LD are helping to explain the history of changes in population size and patterns of genetic exchange. Of note for human population history, an extreme reduction in size (often a population bottleneck – *explained below*), can

increase LD. Generally, after a bottleneck occurs, haplotypes will be lost resulting in increased LD. A period of subpopulation increases will augment LD by also increasing the effect of genetic drift. Long distance LD in humans may indicate a bottleneck that occurred after an initial migration out of Africa. Some studies (Zhang et al., 2004; Schmeger et al., 2005) have shown higher levels of genome-wide LD in some populations, which would indicate a past bottleneck .

Significantly, higher resolution studies of linkage disequilibrium in humans will allow for the investigation of genome-specific regions in which LD variation may indicate the nature of archaic introgression from extinct ancestors, or the investigation of subdivision as humans migrated from Africa.

Population Genetic Theory II: Mechanisms of Evolution

> *A comprehensive theory of evolution, one which does not yet exist, would integrate ecological processes (which determine the range of environments and the fitnesses of phenotypes), developmental processes (which determine the effect of genotype on phenotype), and population genetics (which tells us the changes in genetic composition of a population when the fitnesses of the genotypes are known).*
>
> - Joseph Felsenstein (2009)

Selection: Viewed narrowly, selection is just another deviation from Hardy-Weinberg proportions. Viewed more broadly, selection is the primary force that causes evolution to become adaptive and is the creative and progressive element in the evolutionary process. Because selection operates on the phenotype, rather than the genotype, a population genetic discussion of natural selection ultimately is concerned with quantifying and measuring fitness, *w*, or the capability of an individual or a specific genotype to reproduce and contribute to the next generation. Invariably, we want to be able to detect natural selection in human evolution. A useful starting point for this discussion ultimately

will begin with the neutral theory of molecular evolution, as proposed by Motoo Kimura (1968, 1983).

Kimura (1968) suggested that most polymorphisms observed at the molecular level are selectively neutral. Therefore, the genetic variations that have accumulated and exist within and between populations are largely the result of neutral or nearly neutral processes (Ohta, 1992; 2002) rather than from selection. The neutral model predicts that the loss and fixation of alleles is the result of genetic drift across populations and rate of evolution is simply a function of the mutation rate (Tishkoff and Verrelli, 2003). In other words, genetic polymorphisms that exist within a population are balanced between the effects of mutation and random genetic drift.

The original formulation of neutral theory posited mutations whose fate is determined solely through random genetic drift, known as the strictly neutral model. Of course, Kimura knew that mutations could have various effects upon the fitness of an individual, but developed the theory so that mutations either are deleterious and eliminated, or in the more rare case, are advantageous and become fixed (Hartl and Clark, 2007:318). As new mutations are introduced, random drift determines whether a neutral allele will become fixed or lost. At equilibrium, this balance that is created by drift and mutation results, on average, in each new allele gained by mutation becomes balanced by an existing allele that eventually becomes lost. Although many of the central principles of the strictly neutral model have been disputed, nearly neutral and neutral theories have provided population geneticists with a powerful null hypothesis by which to test natural selection.

Though it has been shown that most genetic polymorphisms are under weak selection or neutral in their behavior, researchers have continuously worked on finding genes that are the target of natural selection, especially those that have historically or recently played a role in disease susceptibility. The population of humans has increased over the last ~ 50,000 years and with this population increase, vast changes in culture and ecology during the Late Pleistocene and early Holocene (~ 10,000 years ago) created new opportunities for accelerated adaptive evolution (Hawks et al., 2007). According to Fisher (1930), rapid population growth should accompany an increase in the rate of adaptive substitutions, or an acceleration of new positively selected alleles.

In other species, the size of the population affects that species response to adaptation. For example, natural insect populations often show a resistance to pesticides in the wild, whereas small laboratory populations under similar selective pressures develop less effective polygenic adaptations (Roush and McKenzie, 1987). During the last 10,000 years, humans have witnessed rapid skeletal and dental evolution and the appearance of many new genetic responses to diet and disease (Armelagos and Harper, 2005).

To approach the study of natural selection in human populations, recent technological advances (whole genome sequencing) coupled with extremely large datasets (HapMap Project, Perlegen, Human Genome Diversity-CEPH Panel, 1000 Genomes Project) have enabled researchers to detect the signatures of recent positive selection among candidate alleles for evolutionary and medical purposes (Hinds et al., 2005; Kelley and Swanson, 2008; Hardy and Singleton, 2009). To date, a number of studies have detected positive selection in the human genome (Biswas and Akey, 2006;

Kelley et al., 2006; Voight et al., 2006; Wang et al., 2006; Kimura et al., 2007; Sabeti et al., 2007; Barreiro et al., 2008; Pickrell et al., 2009; Chen et al., 2010). Many of these studies have identified loci that are important for human adaptation, including genes related to disease resistance (Hamblin and Di Rienzo, 2000; Wang et al., 2006), dietary and subsistence changes associated with lactase persistence (Bersaglieri et al., 2004; Tishkoff et al., 2007), genes involved in skin pigmentation (Harding et al., 2000; Williamson et al., 2007) and the *EDAR* gene involved in hair morphology (Kelley et al., 2006; Fujimoto et al., 2008).

One of the drawbacks to many of the above studies is the reliance on datasets that represent only a fraction of human diversity (ascertainment bias). Although datasets like the HapMap project have characterized over 3.1 million single nucleotide polymorphisms, or SNPs (a DNA sequence that differs between members of a population), only a few populations have been sampled to derive this data. These include one European, one African, and one or two East Asian populations. If selection is to be detected, a higher geographic resolution in the sampling of human genomic diversity is required. More recently, a larger dataset for analyzing genome scans has become available and is known as the Human Genome Diversity-CEPH Panel and uses SNP data containing 938 individuals from 53 populations (Li et al., 2008).

Pickrell et al. (2009) used this dataset (657,143 SNPs) to search for patterns of haplotype sharing and putative selective signals between genetically similar populations, and identify novel candidate loci that have experienced recent positive selection and relate them to phenotypic variation. These authors found several genes that are associated with developmental pathways, pigmentation, and type II diabetes to be under selective

pressure, but find a lower rate of selected genes than Hawks et al. (2007) found in their paper. Hawks et al. (2007) found approximately 7 percent of the human genome appears to have genes associated with selection. This hypothesis stems from rapid population growth during the Neolithic in which agriculture was starting to be adopted. Pickrell et al. (2009) found that the geographic patterns of selection (haplotype sharing) to be strongest in genetically similar populations, especially in Europe, the Middle East, and Central Asia.

These authors suggest this pattern of geographical selective structuring in loci is also characteristic of patterns of neutral loci, and therefore distinguishing true cases of selection from the tails of a neutral distribution may be more difficult than often assumed. However, in response, Hawks (personal blog: http://johnhawks.net/weblog?page=1), suggests the geographical pattern of selection is entirely what we would expect because those signals of selection date to the Neolithic with the complementary rapid population expansion and novel adoption of agriculture. Particularly, the adoption of agriculture from West Asia into Europe, nomadic incursions from Central Asian, and spread of languages across the steppe and south into the Indian subcontinent are all characteristic of the last 10,000 years. If the strong selective patterns now exhibited in these regions required extensive migration and interaction, we should expect other areas of the world with less migration and interaction, to share fewer haplotypes. In this case, long distance dispersal has had a higher impact upon genetic variation, selection, and population history.

Genetic Drift: Much of population genetic theory depends on an idealized population, or a simple representation of a population. Like the basic assumptions held by the Hardy-

Weinberg law, this idealized population assumes nonoverlapping generations of individuals, random mating among individuals, a constant population size of *N* diploid individuals, and random reproduction over individuals resulting in a Poisson (normal) distribution of progeny (Hey and Machado, 2003). This idealized model was developed by Sewall Wright (1931) and R.A. Fisher (1930) and is known as the Wright-Fisher model. The model – random genetic drift with binomial sampling – was developed for the use of only two alleles, but can be extended to include three or more alleles.

If a population contains 2*N* alleles among which two alleles *A* and *a* are present, then the state of the population can be described by the number of *A* alleles in the population. There are two possible states – fixation and nonfixed allele frequencies that are allowed to drift to any other possible allele frequency. The fixed states are known as absorbing states while the probability of the population drifting from a state having *i* copies to *j* copies of allele A is known as the transition probability (Hartl and Clark, 2007:102). This model, expressed in terms of discrete states with fixed probabilities of going from one state to another is known as a Markov chain (Felsenstein, 2009:198).

Both Fisher and Wright pioneered the study of population genetics and we should view their contributions in turn. R.A. Fisher (1890 – 1962) worked on the development of new statistical techniques, such as the derivation of the exact distribution of the correlation coefficient, created one of the most statistically useful measures known as analysis of variance, and invented the technique of maximum likelihood, a method used for fitting a mathematical model to real-world data (Aldrich, 1997). Fisher showed how continuous traits were not only compatible with Mendelian inheritance but was also predicted by it. He also explained how individual characters were not simply "blended

away" through the process of crossing over. He used stature data from human populations to show how a continuous distribution follows a Mendelian inheritance scheme through the interaction of multiple factors (although he concluded environment did not play a significant role in stature variance).

Sewell Wright's (1889 – 1988) major contributions to the field are in the areas of inbreeding, mating systems, and genetic drift, and in the field of statistics, path analysis (Provine, 1986; Crow, 1988). Wright was the creator of both the inbreeding coefficient (the probability that two alleles at a locus in an inbred individual are identical by descent) and *F*-statistics (frequently used to describe the presence of population structure), both standard measurements in population genetics. Contra to Fisher, Wright believed in the importance of interaction systems among genes and the possibilities of drift having large consequences in small populations. Fisher thought selection had more of an effect on larger populations while drift had little significance.

Wright also contributed to population genetics the concept of shifting balance theory. This theory states that a large population becomes subdivided into a set of small, semi-isolated subpopulations, or demes, which he envisaged would occur in three stages. First, initial random genetic drift upon a small subpopulation (allowing the population to explore their adaptive topography – a measure of relative fitness against allele frequency) would then be followed by intrademe and interdeme selection. Following random drift, a phase of mass selection occurs, whereby favorable gene combinations become incorporated into the genome by the act of natural selection. This phase is then followed by between-population selection, in which the more successful demes increase in size and rate of migration, where this increase is also associated with higher fitness, and ultimately

these favorable genotypes spread throughout the entire population in ever-increasing distribution (Hartl and Clark, 2007:245-246). Evolutionary geneticists have debated whether these delicate conditions associated with shifting balance theory hold in natural populations, and thus have remained largely untested (Chouteau and Angers, 2012).

Wright's emphasis upon random genetic drift should not be understated in the field of biological anthropology. The concept states that random changes in gene frequency in a population that occur when a finite number of progeny are formed by the random sampling of gametes from the parents. This random sampling of genes will cause the composition of the offspring and parental generations to differ. In the Wright-Fisher model, ideal populations that have been evolving for a long time are in a 'steady state' or 'equilibrium' pattern of variation in and between sub-populations that have arisen from the balance between random genetic drift (which tend to make populations different) and gene exchange (which makes them more similar). This model is especially useful when trying to understand variation in subdivided populations with fluctuations in population size.

Population size is inherently linked to the effects of random genetic drift. Often, populations have intermittent small population sizes, due to any number of circumstances, such as disease epidemics or other mechanisms that might cause population collapse. These fluctuations in population size cause bottlenecks, which are periods when only a few individuals survive to produce offspring (Amos and Hoffman, 2009). Conversely, small population size is important if a population grows from only a few founding individuals, a phenomenon known as founder effect. As a consequence of the founder affect, genetic variation may be low due to those initial founders, or by

chance have a high or low frequency of particular alleles. The effects of genetic drift in a small population can cause populations to undergo significant fluctuations in only a few generations in unpredictable patterns resulting in chance fixation or loss of a particular allele, or alleles. This concept of fixation, under the assumption of molecular neutrality (absence of differential selection), means that the proportion of populations expected to go to fixation for a given allele is equal to the initial frequency of that allele. This makes intuitive sense given a simple example. If the initial allele frequency is 0.1, only 10% of the time will a population become fixed for that allele. Likewise, if the initial allele frequency is 0.9, 90% of the time it will become fixed (Hedrick, 2005:306). Of course, if other evolutionary mechanisms are at play (such as genetic hitchhiking) these assumptions do not hold true.

Effective Population Size: Underlying the concept of random genetic drift is a measure population geneticists have theoretically developed known as the effective population size, or N_e. The concept of effective numbers in a population underlies another important observation: overall genetic diversity within a species. Recent studies have shown that humans have reduced overall genetic diversity compared to the great apes (Kaessmann et al., 2001; Yu et al., 2004; Charlesworth, 2009). This observation is interesting given the large census size of humans today. What is accounting for the low variation found among human populations?

Effective population size was introduced by Wright (1931) as he considered the increase in identity by descent in various situations. Its purpose is to provide a way of calculating the rate of evolutionary change caused by the random sampling of allele frequencies in a finite population, otherwise due to genetic drift (Charlesworth, 2009).

Another way of describing the estimated effective size is to say that N_e of a population is the number of individuals in an ideal population that would lose genetic variation at the same rate as the actual population (Crow and Denniston, 1988; Leberg, 2005). Of course, this ideal population is assumed to have a stable population size (not changing), and free of mutation, natural selection, and migration. As we know, however, no natural population perfectly fits this ideal population, and as such, it is expected that N_e will differ from N_c (the census size of the population). N_c hardly ever accounts for the effects of inbreeding and drift, and as such N_e correctly reflects those effects. Many factors can contribute to census size being unequal to effective size. These include unequal sex ratios, variation in offspring number, inbreeding, mode of inheritance, age and age-class structure, variance in population size, and spatial and genetic structure (Charlesworth, 2009).

Effective population size can answer a number of important biological and evolutionary considerations. First, the product of mutation rate and N_e determines the equilibrium level of neutral or weakly selected variability in a population. Second, the product of N_e and effectiveness of selection determine whether a favorable mutation spreads or a deleterious mutation is eliminated (Charlesworth, 2009). The implications for humans, who have a low effective size (see below), show evidence for decreased genetic variability and reduced effectiveness of selection in comparison with other species.

Crow (1954) made a further theoretical advance by pointing out there is more than one way of defining an effective size for the population. Kimura and Crow (1963) applied a way to measure effective population size by calculating the rate of change in

variance in allele frequency among subpopulations. Later, Crow and Denniston (1988) explicated the concepts of inbreeding effective size, variance effective size, and eigenvalue effective size (Ewens, 1982). The two most common measures are inbreeding (N_{eI}) and variance (N_{eV}) effective size. Inbreeding effective size estimates the probability of homozygosity due to common ancestry (or the size of an ideal population losing heterozygosity due to increased relatedness), while variance effective size estimates the amount of allele-frequency drift per generation (or is the size of an ideal population experiencing drift) as the same rate as the actual population (Wright, 1931, Crow, 1954; Crow and Denniston, 1988).

The effective size of a population, N_e is actually the harmonic mean of the actual numbers – the reciprocal of the average of reciprocals (Hartl and Clark, 2007:122). This is important because a single period of population decrease (resulting in a bottleneck) can result in a serious loss of heterozyogosity. This loss of diversity is often the result of one subpopulation splitting from the larger population and founding a new subpopulation. The accompanying random genetic drift is then known as a founder effect (Chakraborty and Nei, 1977). Founder effects in humans have important implications such as in medical genetics because a population derived from a small number of founders (such as Ashkenazi Jews) may have an elevated incidence in an otherwise rare genetic disorder (Bray et al., 2010). Other genetic effects of a bottleneck include a reduced number of alleles, a distorted equilibrium of allele frequencies, and an increase in linkage disequilibrium (Hartl and Clark, 2007:123).

A brief hypothetical example should show how the harmonic mean is dominated by the smallest terms, often resulting in a small effective population size over time. An approximation for the harmonic mean can be written as:

$$\frac{1}{N_e} = \frac{1}{t}\left(\frac{1}{N_0} + \frac{1}{N_1} + \ldots + \frac{1}{N_{t-1}}\right)$$

where N_e is the effective population size, t is the generation interval, N_0 is the initial generation, and t-1 is the ith generation. Suppose a population went through a bottleneck as follows: $N_0 = 1000$, $N_1 = 10$, $N_2 = 1000$. If we calculate N_e across all populations using the above equation, we get $1/N_e = \left(\frac{1}{3}\right)\left(\frac{1}{1000} + \frac{1}{10} + \frac{1}{1000}\right) = 0.034$, or $N_e = 1/0.034 =$ 29.4. The average effective number is only 29.4, whereas the arithmetic average number of individuals would be $\left(\frac{1}{3}\right)(1000 + 10 + 1000) = 670$.

It is important to distinguish (and for researchers to make known) which estimate of effective size is being used. As environmental changes have a direct affect on genetic diversity, the reduction or increase in population size also impact allele frequency fluctuation and level of inbreeding. However, population changes do not impact N_{eI} and N_{eV} in a similar manner. A rapid decrease in population size has a concurrent effect upon variance effective size, but although inbreeding effective size will also decrease, it is likely to remain large for many generations (Leberg, 2005). In our example above, the generation that went from 1000 to 10 would have a large effect upon N_{eV} but have a small impact upon N_{eI}. This becomes important when the effective population size of humans is estimated.

Long term estimates for N_e in humans have suggested approximately 10,000 (Nei and Graur, 1984; Takahata, 1993; Stoneking et al., 1997; Eller et al., 2004; Yu et al., 2004; Blum and Jakobsson, 2011; Gronau et al., 2011; Li and Durbin, 2011). Some authors have attributed this low effective size to suggest that humans have recently expanded from a small number of ancestors, a view that fits with a recent African replacement model, as opposed to the regional continuity model (Harpending et al., 1993). Although this small size fits with an explanation of an expansion of a small group of humans from an initial population in Africa, the situation should be apparent that this scenario is overly simplified (Relethford, 2008a). Archaeological evidence suggests a *census* population size for early archaic humans during the Pleistocene to be approximately 500,000 to 1,000,000 (Hassan, 1981). Therefore, the effective size of 10,000 seems incommensurate with the census size if at least one-third of the 500 to 1 million census size are of reproductive age (167,000 to 500,000 effective population size).

This value of around 10,000 N_e seems to contradict the regional continuity model, which implies a large population occupying the Old World. However, models proposing a scenario of a small African population have not considered the effects of population extinction and recolonization. Eller et al. (2004) have shown that a model of extinction and recolonization of local populations with reasonable population parameters (high genetic deme variation, low interdeme migration, kin-structured colonization) could result in a long-term census size magnitudes larger (thus implying the possibility of a regional continuity model) with an effective size of about 10,000. Eswaran et al. (2005) have also shown that a wave-of-advance model could explain low estimates of effective

size given a larger census size. Although these models may not reflect the reality of past human population size, it does show that multiple interpretations of human origins could be made in light of low N_e.

More recently, Premo and Hublin (2009) have proposed the low effective population size seen in humans, Neanderthals (Noonan et al., 2006) and our archaic ancestors, is the result of selection acting upon culturally mediated migration. The idea is that cultural differences between populations usually results in the impediment of gene flow between them. This effect is similar to inbreeding. Coupled with a demographic force acting upon subpopulations – natural selection (advantageous mutations) – genetic diversity is reduced, and has been reduced since the Pleistocene.

Population Structure

Migration and Gene Flow: Almost all natural populations are grouped into smaller subpopulations where mating usually takes place. All species are distributed over space, but few explore their panmictic potential. In other words, most species, like humans, do not visit every part of their natural range in one generation and usually do not choose a mate randomly among all potential mates. These subpopulations are referred to as the population structure. Genetic studies have shown a correspondence between the geographic location of samples and the associated genetic diversity (Goldstein and Chikhi, 2002; Charlesworth et al., 2003; Hey and Machado, 2003; Novembre and Ramachandran, 2011).

When subpopulations become divided, often they are limited or completely isolated from migration. In this case all individuals found within that subpopulation will mate with each other. Population subdivision, then, results in inbreeding because all

individuals within the subpopulation share remote ancestors, even if they choose their mates at random (Hartl and Clark, 2007:276). Wright (1943) was the first to point out that many populations are structured in a hierarchical fashion, meaning that subpopulations can be grouped into progressively inclusive levels, where, at each grouping, the lower levels are included within the higher ones. This is known as nested structure.

Relethford (2002) used a nested approach to partition human global genetic diversity for two different quantitative traits: craniofacial traits and skin color. Like others (Lewontin, 1972; Barbujani et al., 1997) who have partitioned genetic variance in a similar way, Relethford found that the majority of human diversity (using craniometric traits) exists within local populations (~ 85%), with progressively less among local populations (~ 15%) and among major geographic regions (~ 10%). This means that genetic divergence based upon craniometric traits is not great among large continents but is actually found within local populations residing on those continents, implying a model consistent with neutral traits under an isolation by distance model (*see below*).

In stark contrast, a global analysis of skin color showed the vast majority of total variation (88%) is occurring among geographic regions, with much less occurring among (3%) or within (9%) local populations (Relethford, 2002). This pattern, which tells us nothing about population history, is not unexpected given the evidence for selective pressures affecting global variation in skin color (Relethford, 1997b; Harding et al., 2000). Several studies have shown a direct correlation between skin color and latitude, and the amount of ultraviolet radiation, with darker average skin color in populations

living at or near the equator and increasing lighter skin with distance from the equator (Relethford, 1997b; Jablonski and Chaplin, 2000).

One of the most important consequences of population structure is a reduction in the average proportion of heterozygous genotypes relative to that expected under random mating (Hartl and Clark, 2007:276). Although subpopulations may remain relatively isolated resulting in increased levels of autozygous alleles (identity by descent) over time, most subdivided populations are rarely completely isolated. This is where the process of migration becomes important. Migration in this context can refer to the movement of organisms or their gametes (genetic exchange) among subpopulations. Migration results in an increase of gene flow between populations, thus limiting how much genetic divergence can take place. Genetic drift can work in combination with gene flow to homogenize a population. There have been models developed for both one way and two-way migration patterns (Wright, 1951; Kimura and Weiss, 1964; Harpending and Ward, 1982).

One of the most common models of migration is the island model (Wright, 1951). This model assumes a large population that splits into many subdivided populations that are dispersed geographically. Unlike one-way migration where one subpopulation is migrating into another without an equal amount coming in the reverse direction, the island model receives an equal proportion of migrants from all subpopulations. These subpopulations then form a pool which then disperses among the subpopulations. In this way, all migrants contribute equally, where the expected allele frequency among the migrants must equal the average allele frequencies among the subpopulations (Hedrick, 2005).

The unidimensional stepping-stone model incorporates spatial structure (Kimura and Weiss, 1964). In this model, there are an infinite number of subpopulations, each with effective size N, organized along a linear habitat. Each subpopulation exchanges migrants with its two adjacent populations at a rate of m_1, with a symmetric number of migrants going to each adjacent subpopulation. There is also a small rate of exchange (m_∞) between all subpopulations and an external population of infinite size. Crow and Kimura (1970) have shown that if m_∞ is small relative to m_1, then the gene frequency correlation between subpopulations separated by s spatial units is

$$r_s = \exp\left(-s\sqrt{\frac{2m \propto}{m_1}}\right)$$

This equation indicates a decreasing gene frequency correlation with increasing spatial distance, which leads to an expectation that biological distance should increase with increased geographic distance (Malécot, 1969; Morton, 1977).

The migration matrix method provides for the most general model of human migration among a finite number of subpopulations within a region (Harpending and Ward, 1982; Rogers and Harpending, 1983, 1986; Relethford, 1986). This model can be used to represent finite versions of the island model and the stepping stone model, as well as general migration patterns. Under this model, a matrix M, represents the probability that an individual in subpopulation j came from subpopulation i, where the matrix is used in conjunction with a diagonal matrix of deviations resulting from drift to predict a variance-covariance (R) of standardized gene frequencies between groups (Konigsberg, 1990). In addition to exchange between subpopulations, this method can model long-

range migration either by a scalar common to all subpopulations or a vector of immigration rates specific to each subpopulation (Jorde, 1980).

Migration can have a significant homogenizing effect with very few migrants. F_{ST} as a measure of genetic divergence is profoundly affected by migration rate, or *m*, per generation. F_{ST} is a fixation index originally developed by Wright (1921) to quantify the inbreeding effect of population subdivision. This fixation index is equal to a reduction in heterozygosity by comparing the least inclusive to the most inclusive level of the population hierarchy and measures all effects of population structure combined:

$$F_{ST} = \frac{H_T - H_S}{H_T}$$

where H_T is the average heterozygosity assuming Hardy-Weinberg proportions (HWP) among organisms within the total area, and H_S is the average heterozygosity assuming Hardy-Weinberg proportions within random-mating subpopulations. We can measure how migration affects identity by descent by using an equilibrium value of F_{ST}:

$$\hat{F} = \frac{1}{1 + 4Nm}$$

where N is the number of diploid organisms and *m* is the migration rate. The product of N*m* can be interpreted as the absolute number of migrant individuals that come into each subpopulation in each generation. As the number of migrants increases, the equilibrium value (or genetic differentiation) decreases. Only a few (1 or 2) migrants per generation are needed to significantly decrease $\hat{F}$ and thus homogenize the subpopulation.

Wright (1943) developed one of the most widely cited models for continuously distributed populations known as the isolation by distance (IBD) model. This phenomenon, where it is common to find a correlation between pairwise genetic

differences and pairwise geographic distances in samples, has defined human population structure (Malécot, 1969; Konigsberg, 1990; Manica et al., 2005; Prugnolle et al., 2005; Handley et al., 2007). IBD indicates the tendency for most individuals to migrate between neighboring populations, which often results in a smooth increase in genetic differentiation with increasing geographic distances between populations (otherwise known as a cline). This model is in contrast to the island model, which assumes non-overlapping generations and that all migrants can be pulled from a larger "island" rather than coming predominantly from neighboring populations. Making such an inference (that the migrant could be from a larger gene pool) could affect interpretations of diversity, population structure, or even effective population size. Anthropologists need to use and base inferences on more realistic, geographically explicit models (Handley et al., 2007).

Relethford (2004b) has demonstrated, rather remarkably, a strong pattern of isolation by distance with correlating geographic distance for variation in cranial morphology and genetic distance (measured by F_{ST} and estimated from microsatellite data). Further studies (Manica et al., 2005, 2007; Ramachandran et al., 2005) have shown loci that are largely neutral are distributed continuously among human populations, with very little evidence for genetic discontinuities, although the work by Pritchard et al. (2000), Rosenberg et al. (2002; 2005) and Jorde and Wooding (2004) have shown there to be distinct biological clusters among human populations. These authors have suggested that these 5 or 6 clusters are genuine (and not due to sampling) and attributed their presence to slight discontinuities in previously identified patterns of IBD, consistent with a model of reduced gene flow at geographical barriers such as the Himalayas and Sahara

(Rosenberg et al., 2005; Gayden et al., 2007; Henn et al., 2010). However, researchers still do not understand the biological processes that have shaped these clusters.

The debate between continuously distributed genetic differentiation (clines) and distinct clusters of human population structure is important both from an evolutionary and epidemiological perspective. Most of the debate has been over sampling schemes within the HGDP-CEPH dataset, however, it is useful to understand where the majority of the variance originates. To use a clinical example, if a pharmaceutical company wanted to test a new drug for some condition among Africans, they would need an appropriate sample. If genetic diversity can be identified within major clusters (the continent of Africa being one of them), then the company may only have to sample among a small group of African groups. If however, variation is mostly clinal and attributable to geography, then sampling among sub-Saharan groups may not be appropriate to test in individuals residing in Morocco.

Quantitative Genetic Theory

Invariably, any discussion of human variation using fossil evidence, such as this dissertation presents, necessitates the use of metric characters that have a continuous distribution. Konigsberg (2000) outlines the use of quantitative variation in biological anthropology by exploring how we use a Mendelian system of inheritance to understand continuous trait variation. Briefly, we need to understand the simplest model of quantitative genetics known as the *equal and additive effects model*, which states that the phenotype reflects the *net* effect of polygenic inheritance, with each locus (usually many) having an equal effect on the genotype, and where these effects become additive, in the sense that the distribution of phenotypes will begin to reflect a normal, continuous

distribution of traits. The result being that the trait in question can take on a number of possible values and is not limited to a finite number of discrete classes. In order to characterize the dispersion (or spread) of these values, we use the variance of the distribution, which is the average of the squared values around the average value. From this variance, we can partition the genetic additive and environmental variance of a trait in order to estimate heritability. Heritability is normally defined as the proportion of phenotypic variance due to additive genetic effects. These compromise the main parameters gleaned from continuous quantitative genetic data. How are these parameters estimated?

First, we need to understand the notion of genetic kinship. In genetics, this term is equal to the probability that two individuals share alleles at a locus that are identical by descent, meaning the two alleles are identical because the copies were passed onto each other by a common ancestor. This expected additive genetic correlation between pairs of individuals is often referred to as the correlation of relatedness (or coefficient of relationship). For example, the coefficient of relationship between a parent and his child is one-half, because the child has received half of his or her genes from that parent. Correlations between pairs of individuals is normally assessed using a scale between 0 – 1. A value of zero indicates no genetic relationship, while a value of 1 indicates a perfect correlation of variables (or traits) and is normally measured for within self estimates or used for estimates of monozygotic twins. Another way to estimate this measure is to use the covariance between relationships (normally unrelated individuals). The covariance measure is not scaled between -1 and 1, rather it is an average product of deviations around the average value. The additive genetic variance is normally calculated for

pedigree (genealogical) groups. Correlation and covariance methods are often used on sibling relationships to estimate heritability values.

Another method of estimating quantitative genetic parameters is to use regression analysis for parent-offspring groups. Regression refers to the fitting of a straight line that shows the relationship between two (or more) variables. Therefore, a measurement is made on a parent, and the same measure is made on the offspring. Galton (1889) used regression to calculate the stature of offspring from stature of the parent. After calculating a best-fit line between offspring and parent, Galton concluded that a narrow-sense heritability for stature is moderate (meaning that there is a significant additive genetic component to stature). A more general method for estimating parameter values in a model is maximum likelihood. Likelihood methods incorporate parametric models and are defined as proportional to the probability of obtaining the observed data given unobserved parameter values. Maximum likelihood uses values (the log-likelihood) that estimate parameter values that most likely generate the observed data. Though complicated, this method has become more common to estimate trait heritability (Carson, 2006a, 2006b, Martinez-Abadias et al., 2009). The use of quantitative genetics in studies of selection, drift, and mutation is beyond the scope of this dissertation (see Lynch and Walsh, 1998, for a more comprehensive treatment of the subject).

Relethford and Lees (1982) and Relethford (2007) review the basic approaches to quantitative trait theory and variation. These approaches are referred to as either *indirect* or *direct*. Indirect applications involve the indirect application of models of population structure in the assessment of biological differences among populations. Such methods include various multivariate statistical tests used for exploratory reasons, such as

principal components analysis. Direct applications of population structure have as it's goal an estimation of specific parameters that often require more assumptions than model-free methods. Direct applications in the assessment of population structure include the use of the Relethford-Blangero method described below.

Population genetic theory as a quantitative approach studies how genes, or phenotypes, are distributed within and across populations and how gene distributions pattern against time and space (Haldane, 1929; Fisher, 1930; Wright, 1943, 1951, 1977; Mielke et al., 2006:47). Because patterns of genetic (or phenotypic) distribution are affected by evolutionary forces, population genetic theory is inherently tied to evolutionary theory. Population genetics observes changes in allele frequency and makes direct observations of the genotype; however, studies have indicated that quantitative traits are useful to assess similar variables (such as evolutionary forces affecting the phenotype) in a number of anthropological settings (Jantz, 1973; Droessler, 1981; Konigsberg, 1988, 1990; Relethford and Blangero, 1990; Williams-Blangero and Blangero, 1989, 1990; Relethford, 1992, 1994; Konigsberg and Ousley, 1995; Relethford and Crawford, 1995; Hanihara, 1996; Hemphill, 1999; Powell and Neves, 1999; Stefan, 1999; Jantz and Owsley, 2001; Reddy, 2001; Steadman, 2001; Ross, 2004; Stojanowski, 2004; Brace et al., 2006; Stojanowski and Schillaci, 2006; Hanihara, 2008; Schmidt et al., 2011; Seguchi et al., 2011).

A quantitative trait, in its broadest sense, can be defined as a genotype and/or phenotype distribution that is considered continuous, such as head length or height, rather than discrete, such as a blood type or a DNA haplotype (Relethford and Lees, 1982; Konigsberg, 2000; Crawford, 2007:187). The majority of quantitative traits studied in

anthropology are known as ‘complex traits’ due to the observation that many are also polygenic, where a continuous distribution (reflecting a normal distribution) is a function of multiple genes and environmental influences (Falconer and Mackay, 1996; Varela and Cocilovo, 2007).

Complicating the polygenic model of equal and additive effects are nongenetic variables, such as environmental variance, that could potentially influence phenotypic variation. Quantitative variation is the net effect of both genetic and environmental influences, and as such a more complex model needs to consider variation due to dominance (Mielke et al., 2006:236). Often, quantitative genetics contains important statistical aspects, such as the mean and variance of a particular trait. Importantly, we want to understand the variance associated with quantitative trait information. Variance is a useful tool in order to partition genetic and environmental influences. Specifically, we can partition the total phenotypic variance (V_p) into three components: i; The additive genetic variance (V_a) resulting from the equal and additive effects model; ii; The non-additive genetic variance resulting from dominance effects (V_d); and iii; Environmental, or non-genetic, variance (V_e). This can be expressed mathematically as

$$V_p = V_a + V_d + V_e$$

Importantly, partitioning the phenotypic variance allows for an estimation of heritability, often expressed as h^2, which is the measure of the relative proportion of total phenotypic variation that is due to genetic variation (Relethford, 2007). Known heritability can be deduced from living populations through family historical information (such as sibling relationships); however, this estimate is often difficult (if not impossible)

to express in prehistoric populations, since biological relationships among sample individuals are unknown (Sjovold, 1984; Devor, 1987; Varela and Cocilovo, 2007).

Anthropological geneticists often use what is known as a narrow-sense heritability that gives the proportion of phenotypic variance that is explained by transmissible genetic effects (additive genetic variance from parent to offspring). This can be expressed as

$$h^2 = \frac{V_a}{V_p}$$

Considered in the narrow sense, heritability provides a proportion of variance in a trait explained by genetic transmission and is a key parameter in models of evolution of quantitative traits (Konigsberg, 2000). It must be kept in mind that estimates of trait heritability are relative and should not be taken as absolute and fixed. In addition, trait heritability is population specific. If the environmental variance in a population declines, the relative amount of genetic influence will increase by definition. This point is significant because the use of craniofacial morphology (as measured by craniometrics) used in population genetic studies requires an estimation of trait heritability (Sparks and Jantz, 2002; Carson, 2006a). Accordingly, an average heritability for cranial traits (influenced by genetics) is often used to understand the underlying evolutionary processes and relationships within and among groups under analysis (Relethford and Blangero, 1990; Relethford et al., 1997).

This issue of heritability of craniofacial traits has been taken up by a number of authors (Sjovold, 1984; Devor et al., 1986; Devor, 1987; Konigsberg and Ousley, 1995; Sparks and Jantz, 2002; Arya et al., 2002; Carson, 2006a, 2006b; Martinez-Abadias et al., 2009). The general conclusion of these studies is that human craniofacial traits have a

moderate to high degree of genetic variation. Unfortunately, the estimation of heritability for these studies is difficult for comparative purposes, as trait heritabilities have been computed on different kinds of samples (living and skeletal remains) from different geographical regions accounting for different familial relationships (twins, nuclear, extended families), and using a variety of statistical approaches (regression, ANOVA, path analysis, maximum likelihood [ML]). The biggest problem for these studies (except Carson, 2006a and Martinez-Abadias et al., 2009) is that a large, suitable, pedigree-structured skull series is almost non-existent.

One exception is the Hallstatt ossuary collection, located in Austria (Sjovold, 1984). This collection provides for a structured pedigree with a well-known genealogical relationship among skulls. Carson (2006a) used a ML method to provide a more nuanced estimate of cranial heritabilities and found, in agreement with Sjovold's estimates based on Howell's measurements, that craniometric traits show a low to moderate narrow sense heritability. However, Carson (2006a) pointed out some differences and concluded that facial dimensions and cranial breadth measurements are less heritable characters of the skull, though this partly stems from the different statistical approaches used to estimate the traits.

Martinez-Abadias et al. (2009) also studied at the Hallstatt collection, however, they applied geometric morphometric methods to pattern genetic correlation among various cranial traits. As integration is pervasive in the human skull (Bookstein et al., 2003; Martinez-Abadias et al., 2012), it is important to estimate this correlation, as integration between characters can limit the evolvability of traits and determine their evolutionary response (McGuigan, 2006). Using a modular perspective to estimate

cranial trait heritability (facial, neurocranial, and basicranial), the authors test for patterns underlying genetic variation in these regions by applying maximum likelihood methods. They find, similar to Carson (2006a), that trait heritabilities are low to moderate, with the face having the highest number of significantly heritable traits, followed by the basicranium and the neurocranium. However, a comparison of amounts of genetic variation among regions was not statistically significant, implying no significant differences among cranial regions. This, again, indicates a high component of integration for craniofacial traits.

These and other studies have shown a direct correlation between phenotypic and genetic relationships for anthropometric traits (Cheverud, 1988; Williams-Blangero and Blangero, 1989; Konigsberg and Blangero, 1993; Konigsberg and Ousley, 1995). Therefore, metric relationships among groups should reflect an ancestral or phylogenetic relationship with environmental variance having minimal effect across subpopulations under analysis (Williams-Blangero and Blangero, 1990; Ousley and McKeown, 2001). This observation was empirically verified in the Martinez-Abadias et al. (2009) study. These authors tested the correlation of specific suites of craniofacial traits within and among functional regions of the skull by exploring genetic and phenotypic patterns. The null hypothesis (no correlation between generic and phenotypic matrices) was rejected in favor of an observed pattern of high genetic and phenotypic correlation, suggesting that genetic and environmental effects on development produce similar patterns of phenotypic variation. Thus, in cases where a genetic (**G**) covariation matrix is unavailable, a phenotypic (**P**) covariance matrix could be used as a proxy to **G** in population genetic models (Cheverud, 1988; Roseman, 2012). As a correlate finding, however, Martinez-

Abadias (2007) found that the proportionality of **G** to **P** is not a straightforward consequence of the similarity between the correlation matrices. This suggests that phenotypic data may introduce a potential bias in population and quantitative genetic studies unless the sample size is sufficiently large, or pedigree information is available.

Craniofacial Evolution & Development

Physical anthropology has long been fascinated by the evolution, development, and morphological variation of the human head (Lieberman, 2011a). To understand this fascination, we need only look to questions of human evolution, and how the human head differs from other primate species. There are, of course, distinctive human features that might emphasize the evolutionary intensity of the head, which range from a large brain to smaller front teeth and a protruding nose. What selective (or non-selective, i.e., neutral) pressures arose to drive these evolutionary changes? This question can be answered by studying how the head develops and functions in an evolutionary context. However, this is difficult because heads are highly integrated, and the way one regions grows and functions can have significant changes to other parts. One example is the increase in brain size, which triggered how the braincase and face grow, which, in turn, changed the biomechanics of chewing, range of sound frequencies, and changed the overall balance of the head. Due to the structural integration of the human head, it is often difficult to distinguish whether specific shifts were adaptations, by-products of other shifts that were selected for different reasons, or simply stochastic changes (Lieberman, 2011a).

To begin, we need to understand why heads are so complex. This complexity stems from three main sources: the critical functions the head performs (Gilbert, 2006); the number and diversity of its components (Wagner, 1996; Klingenberg, 2008b); and the

degree and extent of integration (Bruner et al., 2010; Martinez-Abadias et al., 2012). First, the head performs a range of critical functions, including housing and protecting the brain, participating in respiration, thermoregulation, vocalization, locomotion, vision, chewing, swallowing, tasting, smelling, hearing, and balance. Therefore, almost anything that enters the body or provides information about the world gets routed through the head. And because the diverse components that perform these functions in the head share the same space and structural supports, they must also grow and change together without loss or compromise of function – from embryo to adult.

As to the second source of complexity, the head is composed of a number and diversity of components that are considered modular. Modularity, being comprised of distinct, partially independent units, is a hallmark of all organisms (Klingenberg, 2008b). Though modularity tends to be less abundant and discrete at higher levels of organization (i.e., the genotype is more modular than the phenotype, cells more modular than organs), it is clear that the human head has an impressive number of modules. In a typical adult human skull, there are diverse structures that include 22 bones that derive from hundreds of ossification centers, 32 teeth, dozens of muscles, the brain, eyes, olfactory bulbs, organs for balance and hearing, as well as component modules at finer levels of structure, including glands, nerves, veins, arteries, and sinuses that supply, drain, and innervate these structures – all packed into a comparatively small space (Lieberman, 2011b).

The third source of complexity – integration – is evident in the fact that form and function of the head are closely intertwined. Integration can be considered complementary to modularity, both being characteristics of complex systems (Klingenberg, 2008b). Modules are partially independent. Integration describes the way

different components, or modules, of a system are combined into a whole. Integration is a general property of all organisms and is manifest throughout the entire body both functionally and developmentally.

At a simple structural level, for example, the roof of the oral cavity is also the floor of the nose; the top of the face is also the floor of the brain; and the pharynx is an important vessel for air, food, liquid, sound, and mucus (Lieberman, 2011a). Therefore, any changes in form or function of any one component of the head inevitably affect the form and function of others. Integration is also apparent in the way heads develop and grow. Various genes that regulate growth and development in the head also influence different organs and tissues at different times of ontogeny. Further, integration is apparent in that functions of the head are not restricted to local processes, but affect disparate areas of the head. As an example of an integrated functional complex, consider chewing. In the act of chewing, one generates force not only in the tooth crowns but in the tooth roots, the periodontal ligament that attaches the roots to the jaws, places where muscles attach to the skull, the temporomandibular joint, and elsewhere (Lieberman, 2011b).

The key to understanding how the human head has evolved, we need to consider the many modules of the head and how these modules function and are integrated. However, this complexity has both good and bad consequences. For example, the multifunctionality, modularity, and integration can make any effort of inferring or testing whether a given feature was favored by natural selection, was a byproduct of selection, or evolved from random evolutionary changes, difficult at best. These features may also complicate and frustrate efforts to test hypotheses about evolutionary relationships, such

as whether two species share a similar feature due to common ancestry, or to independently evolving features that correlate with a convergently evolved feature(s).

The benefit of head complexity lies in the fact that each module that comprises integration is integrated in a special way. What this means is that every component of the head interacts with its neighbors throughout life, especially during growth and development. They stimulate each other in various ways – to grow faster, slower, differently, etc. Sometimes these are due to simple proximity, as in the case of the bony wall of the face with tissues interacting in the same functional space, so that one region can accommodate growth in a neighboring regions, and vice versa. Other times these interactions are more indirect, as when the angle of the cranial base alters the orientation of the upper face.

This high integration and complexity of the head raises a paradox. Because these components are tightly constrained, one might imagine that any change would disrupt how the head grows and functions, leading to a loss of integration, and a decline in performance, and thus overall fitness. Yet, it appears that heads are extremely evolvable (capable of generating a wide range of heritable, phenotypic variation), as seen from the variation and diversity of mammalian skulls in terms of size, shape, and function. For example, the human head is probably more evolvable than other parts of the body, which may be more conservative. Look at the differences in the *Australopithecines*, which are more cranial than postcranial (Green et al., 2007). Another example of a major modification to the head with relative unaffecting consequences of function is the change in the larynx that allows for the improved ability to speak (Negus, 1949). Dropping the larynx resulted in the rearrangement of various components of the throat that function

during swallowing, thus making humans swallow differently (and thus less safely) from other mammals. Given this scenario, one would imagine that selection would have acted against any hominin with a slightly lower larynx (more likely to die at a young age) in spite of potential acoustic advantages (McCarthy and Lieberman, 2001; Lieberman, 2011a).

There are two nonexclusive hypotheses that might account for this type of paradox. The first considers that heads evolved such variation because of the considerable intensity of natural selection on heads. That is, the more intricate, functionally vital parts of the body are more evolvable than other, less complex, regions. Given the head's vital roles in such things as cognition, vision, and smell, it is possible that selection may have acted more strongly on the head than in other regions, such as the knees. However, this hypothesis has not been verified in empirical studies, rather suggesting that other evolutionary forces, such as drift, have a greater effect (Relethford, 1994, 2010; Roseman and Weaver, 2004; Betti et al., 2010).

The second nonexclusive hypothesis suggests that the complex regions of the head derive their evolvability (ala McGuigan, 2006) from the way they are integrated during development through the many interacting layers of epigenetic interactions (Lieberman, 2011a,b). According to this model, complexity facilitates evolvability by high level epigenetic interactions during development to allow the head to tolerate and adjust to a wide range of variation. This formulation is similar to Moss' functional matrix hypothesis (Moss and Young, 1960), which posits the head comprises a number of mutually accommodating functional units. If evolvability theoretically occurs in proportion to the degree of modularity (i.e., the genotype is more evolvable than the

phenotype), and the head is composed of many modules, then heads may be especially prone to evolutionary modification. Because variation is an essential component of evolution, changes to the size, shape, or timing of development in each of the head's modules, can add to opportunity for change.

Lieberman (2001a) likens these changes to *tinkering*, creating and modifying objects opportunistically, using whatever is available and convenient at the time, but not having any inherent design functions. This is especially appropriate if one considers the process of natural selection, which takes advantage of heritable variation made available from random mutation. In his words, "tinkering takes advantage of modularity and leads to integration" (Lieberman, 2011a:15). This integration stems from epigenetic interaction.

How does epigenetic interaction contribute to craniofacial development? First, what is epigenetics? Originally coined by Waddington (1942) as the casual interactions between genes and their products which help to produce the phenotype, the term can be more explicitly defined as the sum of the genetic and nongenetic factors acting upon cells to control selectively the gene expression that produces development and evolution (Hall, 2011). The broad definition of the term has come to define the vast set of processes by which alternative, variable phenotypes derive from a given genotype (Haig, 2004). In cell biology, the term epigenetic has come to refer to heritable changes in a cell's genomic function that do not alter DNA sequences. Gene regulation is influenced by a multitude of factors, such as other genes (transcription factors), other organisms (population density, predators), and environmental factors such as temperature or the uterine environment. All of these levels are included in the study of epigenetics (for further treatment of the subject see Jablonka and Lamb, 2005; Hallgrimsson and Hall, 2011).

Here, the original definition espoused by Waddington (1942) is preferred when discussing the evolution of the human head.

Lieberman (2011b) outlines three major steps to epigenetic integration in the human head: patterning, morphogenesis, and growth. Patterning helps to create discrete units in the right place at the right time. *Hox* genes pattern the first four somites in the head from the axial skeleton (Deschamps and van Nes, 2005), while the rest of the head is patterned via numerous cell lines. These cell lines are intricately integrated with one another in an architectural arrangement from the start so that skeletal units grow around and between functional spaces. Morphogenesis allows precursor cells in particular units to differentiate into cell types that form distinct tissues and organs, via inductive interactions with neighboring tissues. Teeth are an example of inductive interaction (Stock, 2001; Gomez-Robles and Polly, 2012). Epithelial cells (cells that line the cavity of the body and cover flat surfaces) transplanted from the incisor to the molar regions of the jaw cause local mesenchymal cells (multipotent stem cells) to develop into incisors rather than molars (Jernvall and Thesleff, 2000). The special properties of the head via morphogenesis are related to the density of the organs and the functional spaces, which share a common architecturally complex skeletal framework where few walls of bone or cartilage are unique to any single organ or space.

Lastly, epigenetic interaction in morphological integration and function is involved in growth and can generate significant levels of phenotypic variation via mechanisms such as heterochrony, which is the developmental change in the timing of events leading to changes in size and shape. These epigenetic interactions during growth are essential to various functions and development of the head. The masticatory system

provides an example of how functional integration is maintained in response to mechanical and other stimuli (Herring, 1993). In order for proper occlusion to occur, the upper and lower jaws must fit into one another precisely. Ideally, the upper and lower jaws would be mirror images of each other, however, evolution has required more complex 'tinkering' due to several reasons. The mandible and maxilla have different embryonic origins, are patterned differently, and grow through a different set of processes. This sets up particular integrative challenges, as the mandible articulates with the cranial base while the maxilla grows downward from the nasal cavity. As the cranial base is not a stable platform – the cranial base changes angle and length, flexion occurs during ontogeny, facial elongation causes the cranial base to extend – and these processes occur at different rates, in different ways, and with different effect, many epigenetic interactions are necessary in order for the maintenance of proper occlusion (Herring, 1993; Lieberman, 2011a).

Building on the functional matrix hypothesis, Lieberman (2011b) considers additional reciprocal epigenetic interactions among both skeletal and nonskeletal components to account for craniofacial change in human evolution. Using his more integrated hypothesis, in which interactions occur between organs, spaces, and tissues, he accounts for those epigenetic interactions that were most important in the ~7 million history of hominids since our last common ancestor, and especially in the last 2 million years when the genus *Homo* started to see increased encephalization (Holloway et al., 2004). He outlines three sets of epigenetic mechanisms of integration that were most important. First are the effects of intracranial pressure on neurocranial growth that cause components of the braincase to grow superiorly, laterally, and posteriorly as the brain

grows in volume. Signaling growth factors such as *Fgf2* activate osteoblasts in sutures and cause expansion within the cranial cavity, in turn causing drift to occur in the cranial fossae and an expansion of the synchondroses of the cranial base (Enlow, 1990). These growth mechanisms and constraints seem to have accommodated increased brain size in human evolution.

A second mechanism of epigenetic integration stems from the angulation of the cranial base, in which the basicranial platform can accommodate a larger brain relative to the length of the cranial base by being more flexed (Weidenreich, 1941). A number of hominids and within the genus *Homo* show considerable variation for this trait (Lieberman, 2011a). A final aspect of epigenetic integration is skull width. Increases in brain size have been accompanied in humans by a wider posterior, middle, and anterior cranial fossae. As the face grows downward from the anterior cranial fossa and forward from the middle cranial fossa (Enlow, 1990), a wider neurocranium and basicranium have led to a wider face, especially in the upper portions around the orbits and in the middle and lower portions of the face (Lieberman et al., 2004; Bastir et al., 2008). Width dimensions are among the strongest sources of correlation in the mammalian skull (Hallgrimsson et al., 2007), including in humans (Polanski and Franciscus, 2006). These changes most likely were accompanied by a number of epigenetic mechanisms.

In summary, the development of the human head from an evolutionary and developmental perspective is extremely complex, owing to the nature of the complexity of the head itself. It should be no surprise that the human skull has been so well studied – fossil, functional, genetic, and developmental – and that using the skull is in many ways still one of the best approaches to studying human evolution and modern population

history and structure. However, we must keep in mind to not use reductionist explanations as a model to understanding human head evolution. The integrated and modular complexities of the human head mean we need as many lines of evidence to explain models of human evolution, and to test microevolutionary processes.

Concluding Remarks

The overarching theme within population genetic theory deals with elucidating those evolutionary forces that have shaped underlying patterns of genetic diversity within and among groups of humans. To this end, genome-wide studies and evolutionary-development studies have begun to answer those evolutionary processes most responsible for the observed patterns of variation found in natural populations. What is less clear, however, are questions of how genetic variation affects phenotypic traits. As a skeletal biologist working within the confines of bioarchaeological sampling schemes, an understanding of how genetic processes shape phenotypic variation are imperative if we are to gain a more significant understanding of how morphological variation changes through time. What roles do genetic drift or various forms of natural selection play in determining the amount and pattern of phenotypic trait variation?

The polymorphisms responsible for the observed phenotypic variation may be evolving neutrally, or could be transients on their way to being eliminated because they are deleterious, or on their way to fixation due to being adaptive (Mitchell-Olds et al., 2007). Are deleterious mutant alleles with short persistence times explaining the variation within populations, as hypothesized by a mutation-selection balance (Haldane-Muller Principle)? If so, these allelic variants might be very different from the alleles that are responsible for adaptive evolution, with much of the observed variation actively

maintained by natural selection, either in individual populations by balancing selection, or throughout the entire species by local adaptation.

Complex-trait variation, whether in the form of craniofacial variation or in the formation of sickle-cell polymorphisms, is better understood and facilitated by identifying those genes that underlie phenotypic variation. In other words, a goal toward a better understanding of the evolutionary processes affecting phenotypic variation, is knowing the causal relationship between genotype and phenotype. Complex traits and associated diseases are influenced by many factors other than genetic variation. The increasing availability of genomic and developmental tools and a synthesis of human population genetics with plant and animal species have given us the ability to answer important, yet complex, questions relative to human adaptation and evolution.

CHAPTER 3

HUMAN ORIGINS, MIGRATION & EURASIAN COLONIZATION

The colossal geographic expansion and subsequent colonization of the globe by modern *Homo sapiens* over the last approximately 150,000 to 100,000 years has been characterized by debate, complexity, and even controversy (Tattersall, 2009). Most researchers seem to disagree about the origins, timing, and processes of migratory, colonization, and re-colonization events in human history. The changes associated with the human diaspora are well-documented but are still poorly understood (Jobling et al., 2004). This lack of consensus among researchers of human migration studies may be attributable to various artifacts of the research endeavor, which involves data gathered from diverse linguistic, archaeological, paleontological, anthropological, and genetic sources.

There are obvious gaps in research and the literature that need to be filled with increased and varied data: larger samples are required to better understand genetic and morphological diversity and multiple theoretical paradigms are needed in which to interpret the data. However, given these caveats, anthropological, linguistic, ethnohistorical and genetic data are consistently converging to tell a vastly improved account of how humans came to inhabit various areas of the globe. Human migration studies have been important to understanding not only the global colonization process, but ultimately, are important in the context of human evolutionary history. Through these studies, whether in a global, regional, or local context, the realization is apparent that migration is affected by both biological and cultural factors, which in turn have an impact on the population's biology and culture (Relethford, 1997:332).

In a genetic sense, the study of human migration is concerned with those principal microevolutionary forces affecting human diversity, such as natural selection (cultural or biological), or stochastic processes that have shaped human variation, such as genetic drift or gene flow. Although these processes are acting over relatively short temporal and spatial scales, the patterns of migration observed in modern populations should inform us about longer term evolutionary mechanisms affecting our species (Fix, 1999:203). However, anthropological geneticists have usually taken a phylogenetic approach (Lynch, 1989) to the understanding of ancient migrations (Hey and Machado, 2003), and therefore are more interested in a longer time span of evolution that attempts to reconstruct histories according to population genetic divergence occurring in isolation. The problem with this approach is that seldom do human groups evolve in isolation, nor are we reproductively isolated species that can be "split" from one another using typological branching (i.e. cladistic) methods.

According to Moore (1994) populations may fission, become geographically separated but continue to exchange genes, or even merge again at a later time. Although this model of reticulation may be more appropriate for explaining population history in the present, the complex nature of unraveling human interaction in the past may make any ethnogenetic attempt of reconstruction impossible. Likewise, use of classical population genetic models to interpret ancient and modern migration are ultimately too simplistic – often making unrealistic assumptions (for mathematical purposes) about equilibrium between migration rates and patterns over long periods of time.

Fix (1999) suggests using a synthetic model that is more conceptual in nature than either a strictly phylogenetic or population perspective. This approach is based on an

evolutionary framework that would ideally include behavioral ecology, anthropology (intensity of land use, population density, and social stratification), population genetic models (kin structured, post-marital residence), and even the use of computer simulations (Fix, 2005; Hellenthal et al., 2008) that have the potential to analyze a large number of variables that can then be modeled. Fix (1999:213) believes that the incorporation of ecological, economic, and socio-cultural variables into more encompassing models will lead to a better understanding of the migration process, even if these variables are unable to be directly obtained from populations now far removed in time.

This chapter will discuss both a local and regional perspective to account for past migrations, differential demographic processes, origins, and timing of the ancient colonization process in North Eurasia. Migration, in the context of this dissertation, needs to be defined, as it is understood differently depending on the anthropological perspective. For archaeologists, the term migration has been used generally to refer to whole population replacements with long-range movements, with the expectation that these movements cross socio-cultural boundaries (Adams et al., 1978). Within biological anthropology, and more specifically within anthropological genetics, the term migration usually refers to the movement of peoples, with the end result being the movement, or transfer of genes, in what is called gene flow (Crawford, 2007). This chapter will focus on the genetic perspective of migration.

Questions about Eurasian colonization and timing continue to be debated by anthropologists, linguists, archaeologists, and geneticists alike. When did modern humans first move into and colonize northern Eurasia? How have ancient and modern migrations shaped the biological and genetic diversity of this vast region? What are the present

biological and genetic relationships among these various groups? What do these relationships tell us about ancient or modern demographic processes?

To understand the broader picture of human migration, we need the ability to elucidate questions of local patterns of demographic change, kinship, and residence patterns (Burmeister, 2000). To tell this story, diverse datasets need to be analyzed and integrated into a synthetic whole, including the use of molecular data, such as the uniparentally-inherited markers of mitochondrial DNA and the Y chromosome, or bi-parental markers such as autosomal DNA and the X chromosome. Archaeological data, including the use of radiocarbon and isotope evidence, and quantitative biological skeletal markers, such as odontometrics and craniofacial diversity, will be reviewed to better understand the complex nature and process of colonization and migration in northern Eurasia.

For the purpose of this chapter, North Eurasia will be defined by the territory that spans from the Artic Ocean in the north, Ural Mountains in the west, the Sea of Okhutsk in the east, and the Altai mountains to the south. This area covers all of Siberia, part of East Asia (historical Manchuria), and northern Mongolia. To understand how northern Eurasia was colonized, we will, of necessity, have to include a large swath of research into the colonization process of East Asia. In addition, a brief discussion of the initial migration out of Africa will be undertaken as a means to place the origins and timing of the colonization of north Eurasia in an appropriate time-depth context. To further characterize the demography of northern Eurasia, this chapter will explore more recent migrations.

An emphasis in studies of human migration is placed upon local changes that subsequently affected regional diversity. To this end, the quantification of migratory events occurring throughout Asia spatially and temporally must be interpreted within a cultural-specific context. For example, when making any interpretation of genetic diversity and population structure using uniparentally inherited molecular markers (mtDNA and Y chromosome), context-dependent issues such as patrilineal clan association, matrilocality, or other social organization practices that might affect rates of migration must be considered (Seielstad, 1998; Perez-Lezaun et al., 1999; Oota et al., 2001, 2002; Destro-Bisol et al., 2004; Wood et al., 2005; Wilkins and Marlowe, 2006; Chaix et al., 2007; Hammer et al., 2008; Kumar et al., 2008; Segurel et al., 2008).

Researchers who study human migrations often use multiple sources of data and methods in order to make accurate and reliable interpretations. Various inquiries using genetic, biological, archaeological, ethnographic, and linguistic data have been used collectively or in isolation to interpret ancient migration events. There are advantages and disadvantages to all of these types of data, however, when used in conjunction with one another they can provide robust and powerful tools for interpretation. This chapter will privilege the biological (quantitative markers) and genetic data as the most applicable sources of information to infer ancient and recent migration events.

Genetics and Migration Studies

During the course of research into human diversity, origins and migrations, the study of classical genetic markers (Cavalli-Sforza et al., 1994) has eventually given way to more widespread usage of DNA markers (Cann et al., 1987; Bowcock, 1991; Jorde et al., 2000; Rosenberg et al., 2002, 2005; Weber et al., 2002; Watkins et al., 2003; Bastos-

Rodrigues et al., 2006; Conrad et al., 2006; Li et al., 2008; Durbin et al., 2010). Until recently, the most widely used molecular marker in population genetic studies were the maternally inherited mitochondria (Cann et al., 1987). More recently, the paternally inherited Y chromosome has increasingly been used in population genetic studies to investigate human demographic histories (Underhill and Kivisild, 2007). Both mitochondrial DNA and the Y chromosome contain rapidly evolving markers that are most informative for reconstructing human evolutionary history over the past 10 to 500 thousand years, while the more slowly evolving nucleotide variability of autosomes (including insertion/deletion polymorphism) and the X chromosome are more informative over time depths ranging from 0.5 to 2 million years, the most recent common ancestor of autosomal and X-linked genes (Tishkoff and Verrelli, 2003).

Mitochondrial DNA (mtDNA) is a short stretch of DNA that forms a continuous circle and is only found outside the nucleus of a cell in organelles known as mitochondria and is inherited as a single locus (Anderson et al., 1981; Stoneking and Soodyall, 1996; Andrews et al., 1999; Pakendorf and Stoneking, 2005). Migration studies have employed the use of mtDNA extensively because it is maternally inherited (Giles et al., 1980), essentially lacks recombination (although see Kraytsberg et al., 2004), has a low effective population size, and has a higher mutation rate than nuclear DNA (Comas et al., 1996; Kolman et al., 1996; Yao et al., 2002; Quintana-Murci et al., 2004; Black et al., 2006; Nasidze et al., 2008; Achilli et al., 2008; Qin et al., 2010).

Alternatively, the Y chromosome is a molecular marker that is exclusively inherited in male offspring (Casanova et al., 1985; Jobling and Tyler-Smith, 1995). The nonrecombining portion of the Y chromosome (NRY) acts much like mtDNA to reveal

the structure among human populations and possibly to infer the order and timing of their descent (Underhill and Kivisild, 2007). Y chromosome makers tend to show restricted regional distribution, or population specificity, making them ideal in being able to mark unique migration events in the past (Hammer, 1994; Hammer et al., 1997; Jin and Su, 2000).

In the past few years, there has been significant progress in reconstructing the detailed genealogical branching order of the tree topologies for both mtDNA and the nonrecombining portion of the Y chromomsome (Jobling and Tyler-Smith, 2003; Underhill and Kivisild, 2007). Like mtDNA, use of the Y chromosome has been extensively studied and used in migration studies and in the analysis of population history and origins (Quintana-Murci et al., 1999a; Wells et al., 2001; Cinnioglu et al., 2004; Zegura et al., 2004; Nasidze et al., 2005; Pakendorf et al., 2006; Simms et al., 2011). More recently, studies have started to emphasize the need to include both mtDNA and the Y chromosome (among other genetic markers) in analyses of population history, structure, and migration (Matukusa et al., 2010; Yunusbayev et al., 2012).

Both mtDNA and the Y-chromosome studies define population relationships through the use of the haplogroup and haplotypes. Haplotypes are defined as a combination of alleles or DNA sequences at adjacent loci on a chromosome that are transmitted from generation to generation together. A haplotype may be one locus, several loci, or an entire chromosome depending on the number of recombination events. A haplogroup is a group of similar haplotypes that share a common ancestor. Haplogroups are generally assigned letters of the alphabet, and refinements consist of a combination of letters and numbers.

Both genetic systems have advantages and disadvantages. Though both have given researchers significant insight into human population history, neither can fully infer some basic parameters of human demographic history because each provides only a single window into the past. mtDNA, as described above, does not recombine and mutates rapidly, allowing for high resolution of population specific haplogroups (a group of similar haplotypes that share a common ancestor with a single nucleotide polymorphism mutation) that are usually geographically oriented. Both mtDNA and the NRY are hemizygous, or haploid in nature, meaning their effective population size is much smaller than autosomal DNA. Further, the Y chromosome may even have a more limited effective population size due to mating practices such as polygamy or variance in reproductive success.

There are a few severe drawbacks to using these markers. mtDNA is limited in its genome size and there is greater ascertainment bias (error introduced sampling scheme) in the distribution of polymorphic sites along the mtDNA genome. These sites are rare in coding regions, but rich in non-coding regions (hypervariable regions), however, this richness is often associated with recurrent mutations that make interpretation more difficult (Jin and Su, 2000). There are also potential problems with the Y chromosome. Selection acting on Y chromosomes will influence age estimation for common ancestors, in addition the NRY is subject to the effects of genetic drift and differential male success in producing offspring, which can critically affect the haplotype frequency (Jobling and Tyler-Smith, 1995).

Over the last several years, technology has allowed larger-scale studies to be performed using genome-wide data, using both large SNP (single nucleotide

polymorphisms) arrays and whole-genome sequencing – for both ancient and modern genomes (Lalueza-Fox and Gilbert, 2011; Stoneking and Krause, 2011). Though the scope of this research is beyond the aim of this chapter, a brief introduction is necessary since, most likely, in the future, studies of migration and population history will continue to employ genome-wide data. In the year 2010 alone, three ancient hominid nuclear genomes were sequenced (Green et al., 2010; Rasmussen et al., 2010; Reich et al., 2010), initial results from the 1000 Genomes Project made available (Durbin et al., 2010), among several other human genome and exome sequences published (Fujimoto et al., 2010; Li et al., 2010; Schuster et al., 2010). Genome-wide data have a greater potential to give a more accurate rendering of human population history, thus enabling more detailed demographic processes.

In addition to genome-wide data, the number of studies employing the use of ancient DNA to answer questions about both modern and archaic humans have also significantly increased over the last several years (Keyser-Traqui et al., 2003; Haak et al., 2008; Adachi et al., 2009; Kim et al., 2010; Krause et al., 2010; Adachi et al., 2011; Lacan et al., 2011; Raff et al., 2011; Rasmussen et al., 2011; Reich et al., 2011). Genetic analyses from ancient skeletal remains have the greatest potential to better understand kinship practices, mating patterns, social structure, and burial practices among ancient populations.

Origin of Modern Humans and Dispersals from Africa

Based on genetic and morphological data, a small subset of modern humans migrated out of Africa around 100 to 50 thousand years before the present (yr BP) and dispersed into Arabia and southern Asia sometime before 50,000 yr BP (Lahr and Foley,

1994; Kivisild et al., 2002; Mellars, 2006a, 2006b; Balter, 2011; Rose et al., 2011). The evidence for a recent African origin comes from many sources and is well supported by the data. Africa harbors the deepest genetic lineages and has the most observed diversity (Cann et al., 1987; Vigilant et al., 1991; Underhill et al., 2000; Jobling and Tyler-Smith 2003). Genome studies have also confirmed this view (Jakobsson et al., 2008; Li et al., 2008; Henn et al., 2011), while also showing how the San of southern Africa have the deepest population divergence of any modern group (Schuster et al., 2010). The close correlation between the amount of genetic diversity in a population and the geographic distance from East Africa, known as a serial bottleneck model, also demonstrates and strongly implies an African origin for modern humans (Prugnolle et al., 2005; Ramachandran et al., 2005; Manica et al., 2007; DeGiorgio et al., 2009). This observation has also been shown using phonemic (language) diversity (Atkinson, 2011), though has shown to be controversial.

Although research has now shown a high likelihood for a recent African origin, the question remains as to how many times, and the direction of dispersal of modern humans from Africa. The sequencing of the Neanderthal genome has contributed to this debate (Green et al., 2010). According to these authors, all modern non-Africans share about the same amount of gene flow from Neanderthals. That is, non-African populations seem to share approximately 1-3% of their DNA with the Neanderthal population. The striking observation that Chinese and Papua New Guineans share the same amount of DNA as the French would suggest that a single dispersal took place out of Africa, and then admixture occurred with various Neanderthal populations before the divergence of Asians and Europeans. This would seem to favor a single dispersal, rather than multiple

dispersals. An alternative explanation for the observed genetic similarity between Neanderthals and non-Africans would be deep (ancient) population structure within Africa before a dispersal into Eurasia and/or the joint effect of ascertainment bias (sampling bias of SNPs that inflate polymorphisms, overestimating genetic variation) and genetic drift (Gunz et al., 2009; Wall et al., 2009; Blum and Jakobsson, 2011).

However, these scenarios seem less likely with the finding that an ancient hominin found in Denisova cave, located in the Altai mountains of southern Siberia, is related to populations in Southeast Asia and Oceania (Reich et al., 2010, 2011; Skoglund and Jakobsson, 2011). These authors have found a genetic signal of gene flow occurring from the archaic Denisovan specimen into various island Southeast Asian populations, but not mainland East Asians. These findings would suggest that an admixture event occurred in Southeast Asia with this archaic population and then spread out into various other geographic locales. The authors interpret this evidence to mean that, rather than ancient population structure within Africa or genetic drift, a recent African origin was followed by a few admixture (or assimilation) events with non-African hominins.

Though researchers continue to debate the number of dispersals out of the Africa, one theory that is beginning to gain prominence is the so-called “coastal express”, or southern route of colonization (Macaulay et al., 2005; Field et al., 2007; Atkinson et al., 2008). More traditional models hold that humans first dispersed via the Levant (Stringer, 2000; Luis et al., 2004; Prugnolle et al., 2005) around 45,000 yr BP and spread north-eastwards toward Europe, Siberia, and the northern portion of south Asia (Foley, 1987; Cavalli-Sforza et al., 1993). However, the coastal model contends an earlier dispersal via

the Horn of Africa 60 to 75,000 yr BP along the tropical coast of the Indian Ocean to southeast Asia and Australia (Quintana-Murci et al., 1999b).

Recent work (Rose et al., 2011) has shown concrete evidence for human settlement in Arabia. These authors report on a buried site in Oman (southern Arabia) with a surface scatter that appears to belong to the regionally-specific African lithic industry known as the late Nubian complex in the Horn of Africa. The site dates to around 106,000 years ago, providing evidence for a distinct Northeast African Middle Stone age technocomplex in southern Arabia. These archaeological results appear to confirm a possible earlier migration out-of-Africa into southern Asia.

Archaeological sites in India and Sri Lanka show technological and lithic assemblages that are very similar to those of eastern and southern African sites that date to the time around when modern humans first dispersed from Africa (Kennedy, 1999; James and Petraglia, 2005). Genetically, southwest Asia is characterized by deep genetic lineages for mtDNA and the Y chromosome (Kivisild et al., 1999, 2003; Metspalu et al., 2004), and genome-wide analyses also confirm this view (Reich et al., 2009). Indian-specific mtDNA haplogroups M, N and R and Y chromosome H, L, and R2 lineages show that groups from southern India share a genetic heritage with settlers from the Late Pleistocene (coalescent ages ranging from 30 to 70,000 yr B.P.). These results would suggest that anatomically modern humans had reached southern Asia by at least 60,000 yr BP and that there is genetic continuity between early migrations into southern Asia and modern people.

Of course, the coastlines in question during this period are now submerged below the rapidly rising sea levels of the past 15,000 yr BP (Field et al., 2007) making a more

definitive tie for strong archaeological evidence challenging. However, archaeological (O'Connell and Allen, 2004) and genetic (Ingman and Gyllensten, 2003; Hudjashov et al., 2007) evidence suggest colonization of Australia and New Guinea by as early as 45,000 yr BP (range 30-70,000 yr BP). This would suggest the possibility of modern humans reaching the southern part of East Asia around the same time, despite a lack of direct archaeological evidence for such a claim.

Wollstein et al. (2010) used genomic data (approximately 1 million SNPs) from populations residing in Borneo, New Guinea, Fiji and Polynesia. The authors used advanced statistical approaches to test the southern dispersal route hypothesis by evaluating three models of dispersal (Wollstein, et al. 2010). These scenarios included a single dispersal of modern humans from Africa, followed by a single migration to Asia and New Guinea; a single dispersal from Africa followed by separate migrations from a non-African source population; and a multiple dispersals model (Lahr and Foley, 1994) whereby separate migrations occurred from Africa in the ancestry of Eurasians and New Guineans. The model that received the strongest support was for a single dispersal followed by migrations from a non-African source population, while the weakest model was for multiple dispersals. Although not definitive evidence, this does point strongly to an earlier migration out of Africa with subsequent migrations from a non-African source population.

Migration and Colonization of East Asia

The study of East Asian population history is relevant to further disentangling the complex evolutionary history of northern Eurasia (Stoneking and Delfin, 2010). East Asia is a vast territory that encompasses a wide variety of environments, peoples,

cultures, and languages. This diversity makes reconstructing the history of this territory a challenge. This section will review the initial colonization of East Asia, the direction of migrations between SE Asia and northern Asia, and the genetic relationships and social practices that have impacted the genetic diversity seen in the region today. The reconstruction of migration patterns and processes in East Asia is particularly important and of interest because: *1)* although there is an abundance of hominid skeletal material from this area of the world, the routes of the earliest dispersals into East Asia are not well understood; and *2)* this region serves as a point of origin for later migrations into Japan, Siberia, and the Americas (Karafet et al., 2001).

One hypothesis for the colonization of East Asia has been for a single-dispersal of early migrants as opposed to multiple dispersals (Durbin et al., 2010). A recent study of an Aboriginal Australian genome has the potential to clarify the number of dispersals into East Asia (Rasmussen et al., 2011). If there was a single dispersal into Asia, then the Aboriginal Australians are predicted to have diversified from within the larger Asian population cluster. Studies have shown a split between Europeans and Asians at between 17,000 and 43,000 yr BP (Keinan et al., 2007; Gutenkunst et al., 2009), which is not compatible with an Asian population continuity for Australians if there is archaeological evidence found as far back as 50,000 yr BP. Some suggest an independent migration out of Africa before population expansion, whereby those individuals were assimilated or replaced by later migrations, with a few exceptions, including Aboriginal Australians (Lahr and Foley, 1998). Using high-throughput genome sequencing on a lock of hair from an Aboriginal Australian male, Rasmussen et al. (2011) have shown that they are

descendants of an early dispersal into East Asia, ~ 62-75,000 years ago separate from the population expansion that gave rise to all other East Asians.

Surprisingly, even though there is an abundance of hominid remains found throughout East Asia spanning the last several hundreds of thousands of years, little can be gleaned in terms of hominin migration (Trinkaus, 2005; Shang et al., 2007; Fu et al., 2008). Specifically, early modern humans dating from the Late Pleistocene have been uncovered in various contexts within East Asia, including the Zhoukoudian Upper Cave remains dated to 24-29 ka ^{14}C BP (Matsumura and Pookajorn, 2005), the Minatogawa sample from Okinawa dated to approximately 18 ka ^{14}C BP, Tianyuan Cave in China dated to 42-39 ka using direct accelerator mass spectrometry radiocarbon (Shang et al., 2007) and the Qianyang hominid sample from Liaoning Province in China dated between 16 and 22 ka using the uranium-series (U-series) method (Fu et al., 2008).

Harvati (2009) described the Upper Cave specimens (101 and 103) using a 3D geometric morphometric approach in order to place them into a larger context that includes modern East Asian morphology. The results from this analysis indicate that the Upper Cave specimens show a particular morphological affinity to Upper Paleolithic Europeans while exhibiting important aspects of modern human ancestral morphology. This observation seems to be in accordance with the Single Origin model for the origin of modern humans.

More recent excavation has uncovered fossil remains (two molars, an anterior mandible) from the Late Pleistocene in southern China. Liu et al. (2010) described several fragmentary remains found in Zhiren Cave in South China that have been firmly dated to the Late Pleistocene (>100 ky BP). This find pre-dates some 60 thousand years

the oldest previously known modern human remains in this region. The mandible in particular is interesting due to several traits that appear to be derived in modern humans, though conjointly found with corpus robustness often attributed to Late Pleistocene archaic humans. These authors suggest the origin of modern humans in Eurasia came about through population continuity with existing archaic groups, similar to older 'Assimilation' models of East Asian population history (F. Smith, 2009). They suggest that the emergence of modern humans in Eurasia preceded the behavioral complex seen during the Upper Paleolithic and question the relationship of modern humans with our archaic ancestors in Eurasia. These various fossil finds, though allowing greater insight into population history, continue to leave questions as to the timing and routes of colonization and migration within East Asia.

Traditionally, East Asia has been characterized genetically by populations now residing in the modern People's Republic of China (Chu et al., 1998; Ding et al., 2000; Yao et al., 2000; 2002a, 2002b; Kong et al., 2003a, 2003b; Deng et al., 2004; Zhong et al., 2011) with particular focus on Han ethnicity (Wen et al., 2004a, 2004b). However, more recent studies have included populations found throughout East Asia (northern and southern groups), South and Southwest Asia, Central Asia, and northern Eurasia (Su et al., 1999; Karafet et al., 2001; Wells et al., 2001; Oota et al., 2002; Comas et al., 2004; Jin and Su, 2006; Derenko et al., 2007).

Two major routes into East Asia have been suggested based on genetic evidence. The first, using classical genetic markers, contends that modern humans migrated out of Africa and settled in either southern East Asia and/or central Asia before moving into China and Siberia (Nei and Roychoudhury, 1993; Cavalli-Sforza et al., 1994). More

complex scenarios have been proposed that involve multiple migrations from Southeast and Central Asia during various historical periods (Ding et al., 2000; Karafet et al., 2001), also referred to as the "pincer" model. There is also a competing model based upon dental data (Turner, 1987) that posits all northern East Asian populations derive from peoples dispersing from Sundaland (a biogeographical region that comprises island Southeast Asia). Hanihara (1993, 1996) showed a clear separation of East Asian from Southeast Asian populations based on craniofacial morphology.

These studies also posit in one way or another a clear distinction between northern and southern East Asian populations (although see Chu et al., 1998). Karafet et al. (2001), examined variation at the non-recombining portion of the Y chromosome (NRY) in the framework of various population genetic and statistical models, and suggest an ancient clinal pattern for northern East Asian groups, but little structure in southern East Asian groups. They also found close similarity between their northern samples and Central Asian populations, as championed by Cavalli-Sforza et al. (1994), however, close sharing of haplogroups between northern and southern East Asian populations would also suggest subsequent short-range and long-range migration processes, perhaps associated with the advent of agriculture and animal domestication.

Shi et al. (2005), in agreement with Su et al. (1999) and Jin and Su (2000) presented evidence for a southern to northern migration occurring during the Last Ice Age. Both of these studies, using Y chromosome biallelic markers, suggest southern populations residing in East Asia are more polymorphic than northern populations. In particular, Shi et al. (2005) screen for the Asian-specific haplogroup O3-M122 (not found outside East Asia with average frequencies of 41.8% in Han populations) and suggest

because this marker is more diverse in the southern populations sampled, it would indicate a northward migration that occurred ~25,000-30,000 yr BP. However, as critiqued by Karafet et al. (2001), Su et al. (1999) did not include a large enough sample from northern East Asia and none from Central Asia.

The evidence presented by Shi et al. (2005) is informative, but their use of one marker, although highly informative, may only trace the history of that particular haplogroup (i.e. allele[s]) and not necessarily whole populations. These conflicting studies may not be incompatible, as more recent demographic events such as the Neolithic expansion and contacts along the Silk Road in central Asia, in addition to subsequent isolation of the northern populations may have erased any trace of an early Paleolithic dispersal coming from southern Asia.

More recent work using a large number of SNPs (autosomal variation) has given strong support for a north-to-south direction of migration (Tian et al., 2008; HUGO Pan-Asian Consortium 2009). These authors agree with studies using uniparental markers that the greatest variation is seen in southern East Asia (Ke et al., 2001; Oota, 2002; Kivisild et al., 2002; Li et al., 2007). These studies found a strong and significant correlation between haplotype diversity and latitude (clinal structure), with greater diversity in the south than in the north. Coupled with a maximum-likelihood approach to population relationships, the HUGO study also indicated a direction of population spread from south to north for the direction of colonization within East Asia.

Xue et al. (2006) further explored male demography in East Asia and posit a north-south contrast in human population expansion times. These authors, using a combination of paternal short-tandem repeat (STR) and binary markers, conclude that the

northern populations (including Mongolians, Evenks, Oroqen, and Han) started to expand 34 to 22 yr BP, before the last glacial maximum at 21-18 yr BP. In contrast, the southern groups expanded later (between 18 and 12 yr BP) but grew exponentially faster. Zhong et al. (2011) found similar results in their analysis of the Y chromosome on a large number of Chinese populations. These authors concluded the observed genetic divergence is due to a small contribution from Western Eurasian populations prior to the Paleolithic expansion from the south.

These explanations for contrasting expansion times involved the ability of northern groups to exploit megafauna during the Upper Paleolithic period, which for this region (northern China and southern Siberia) was a highly productive environment with an abundance of large animals (Kuzmin and Orlova, 1998; Goebel, 1999). The southern East Asian groups did not experience a similar environment due to the LGM, and expanded only after temperatures became warmer and more stable and humans could exploit plant resources such as tubers (~ 15,000 yr BP).

On a more local level, genetic studies have attempted to explain more recent demographic events, such as the demic expansion during the Neolithic of agricultural communities occurring ~10,000-8,000 yr BP, the southward expansion of the Han population, and more recent events such as migrations along the Silk Road (Comas et al., 1998; Su et al., 2000; Hanihara, 2004; Wen et al., 2004a, 2004b; Black, et al. 2006). For example, Wen et al. (2004a) investigated the expansion of the Han culture and supported a demic diffusion model similar to European agricultural expansions out of the Near East (Cavalli-Sforza et al., 2004). Interestingly, these authors also found differing sex-specific demographic histories as revealed from mtDNA and Y chromosome analysis. They found

that there is no significant difference between northern and southern Han groups when examining the male lineage, but significant and substantial differentiation according to the maternal history. These findings are indicative of sex biased population admixture in southern Hans, meaning the expansion process of the Han population was dominated by males.

Clarifying the Han contribution to modern East Asians, Gao et al. (2007) analyzed ancient DNA taken from dental remains from the Laija site located in northwestern China, which dates to 3,800 to 4,000 yr BP. Archaeological research of the Laija site suggests it was associated with the Qijia culture, a major culture that flourished during the late Neolithic Age to the early Bronze Age and whom were a branch of the tribal peoples known as Di-Qiang (Ren et al., 2002). During a process of tribal integration the Qijia culture evolved to become the Huaxia civilization, which later developed into the Han. The Di-Qiang also migrated southwest and eventually developed into part of the Tibeto-Burman speaking populations (according the geneticists), who are now widely distributed throughout areas of central and southern China, and Tibet (Wen et al., 2004b). Haplogroup comparison for the two ancient individuals (found in the same house) reveals a consistency and continuity of geographical distribution to modern populations, although different haplotypes were found. The haplogroup diversity would exclude the possibility of a matrilineal social structure; however, the ancient sample did reveal continuity meaning they most likely have contributed to the modern gene pool of people now residing in Northwest China. These results are supported in a larger ancient DNA study from Qinghai province (Zhao et al., 2011).

Overall, these authors reveal the complex nature of population history and structure in East Asia that can be characterized as multilayered, multidirectional, and a continuous history of recurrent gene flow with subsequent genetic admixture between groups. This trend has further consequences and similar patterns when attempting to unravel the complex nature of migrations into the far northern reaches of Asia, and ultimately into the Americas.

Migration and Colonization within Siberia

Siberia is a vast expanse of territory (over 12 million km^2) that Russian geographers define as extending from the Ural Mountains in the west to the Pacific Ocean in the east, including massive watersheds of the northern-flowing Ob, Yenisei and Lena rivers. Ecologically, Siberia is characterized by three latitudinal zones – southern, subarctic, and arctic Siberia, with each zone presenting challenges to human occupation and further migration during the prehistoric and recent periods (Goebel, 1999). Southern Siberia is differentiated from northern Siberia as being more mountainous, and mantled by an array of diverse vegetation communities, while the north is typified by relatively flat, featureless terrain and comparatively homogenous biomes (boreal forest or taiga). Goebel (1999) believes that during the Pleistocene, southern Siberia was not as productive as northern Siberia. He characterizes the north as the "Mammoth-steppe" that presented more opportunities for humans than did the boreal forest to the south.

The Paleolithic of Siberia: The Siberian Paleolithic has been organized by archaeologists into three sequences (Lower, Middle, Upper) that match the Eurasian archaeological record. Although scarce evidence suggests an early colonization during the Lower Paleolithic (Middle Pleistocene), most archaeologists agree that more reliable dates and

artifact industries come from the Middle Paleolithic (130,000 – 40,000 yr BP) (Kuzmin and Orlava, 1998; Vasil'ev et al., 2002). Firm dates for the Middle Paleolithic are from 70,000 to 40,000 yr BP and are characterized by the Mousterian lithic tradition, probably manufactured by Neanderthals. Further evidence for the presence of Neanderthals in southern Siberia come from Krause et al. (2007) who extracted ancient mtDNA from a hominid specimen found at Okladnikov in the Altai Mountains, which dates to 37,750 to 43,700 yr BP. These authors found that the mtDNA sequence from the Altai hominid was similar to mtDNA from Neanderthals in Europe and the Caucasus. This finding raises the intriguing possibility that Neanderthals, who are thought to have colonized Central Asia (Hublin 1998), could have also colonized most of the Russian plains during a warm period around 125,000 yr BP, and may have migrated further east into parts of China and Mongolia (Krause et al., 2007).

The Upper Paleolithic in Siberia has been well-documented (Vasil'ev, 1993; Chlachula, 2001a, 2001b, 2001c; Vasil'ev et al., 2002) using a radio-carbon based chronology (446 ^{14}C dates for 111 sites dated older than 12,000 yr BP). These data suggest the earliest traces of modern human occupation are found in two areas of southern Siberia, the Altai Mountains and the Transbaikal, around 43,000 to 39,000 yr BP. By 13,000 yr BP (possibly later) almost all of northern Asia, including the extreme part of northeastern Siberia had been colonized by modern humans (Vasil'ev et al., 2002).

The Upper Paleolithic evidenced traditions associated with the appearance of cultural manifestations such as mobile art objects, bone technology, and personal ornaments (Goebel, 1999). Even though these items have been found, Goebel (1999)

suggests the humans who moved into these regions were relatively limited in their range, possibly being "tethered" to the local resources afforded them through the exploitation of large fauna, or even by the availability of lithic sources. Only later, around 30,000 to 20,000 yr BP after exploiting the "Mammoth-steppe" did humans begin to expand into greater territories that included trade between central-east Europe and west Eurasia as evidenced from the similarities in tools, technology, and art form. These range expansions were possibly due to the human populations' ability to adapt to extreme conditions of the subarctic biome, and ultimately colonize Siberia above 60^{o}N latitude.

In summary, it appears that Siberia was not colonized until the Middle Paleolithic (~45,000 yr BP) with the advent of the Mousterian tool tradition, generally found in the Altai region of southern Siberia. During the Late Upper Paleolithic (postdating 20,000 yr BP), modern humans began to form small groups of highly mobile hunter-gatherers and began to expand into the Sayan Mountains, the Angara River basin, the Trans-Baikal, Mongolia, and finally, into the far reaches of northeastern Siberia by at least 13,000 yr BP (Karafet et al., 2002).

Genetic Studies of Siberian Populations: Genetically, the region of Siberia is important for a number of reasons. First, Siberia is believed to be where Paleoindians migrated and eventually populated the Americas (Long and Bortolini, 2011; Dulik et al. 2012). The most ascribed model to date involves a land route through NE Siberia into Beringia and then colonizing the Americas. Second, the region of Siberia is vast, and therefore reflects a complex history of population movements and interactions by people inhabiting areas of Eurasia and central Asia. Although Siberia has been extensively documented archaeologically, questions still surround the origins, timing, and routes of founding

migrations into Siberia and the Americas. To this end, the genetic history of Siberia is replete with extensive interaction and admixture between groups from a large area of the globe.

Researchers have begun to unravel the population history and structure of Siberian groups, and how local demographic processes have shaped the genetic diversity of the region. Various authors have attempted to answer these issues using uniparentally-inherited markers, such as mtDNA (Derbeneva et al., 2002; Derenko et al., 2003, 2007a, 2007b; Pakendorf et al., 2003, 2006; Malyarchuk, 2004; Starikovskaya et al., 2005; Phillips-Krawczak et al., 2006; Volodko et al., 2008), the Y chromosome (Karafet et al., 2002; Derenko et al., 2006; Pakendorf et al., 2006), ancient DNA (Ricaut et al., 2004, 2005; Lalueza-Fox et al., 2004; Amory et al., 2006; Mooder et al., 2006; Keyser et al., 2009; Bennett and Kaestle, 2006, 2010; Crubezy et al., 2010), autosomal loci (Uinuk-Ool et al., 2003) and even strontium isotope analysis (Haverkort et al., 2008).

Today, Siberia is characterized by several ethnic groups that speak approximately 35 indigenous languages (grouped into Altaic, Uralic, or Paleosiberian). Although diversity of language still exists among these groups, they are known to share common types of economic activities, such as hunting, fishing, reindeer breeding and cattle herding. These traditional occupations are linked to nomadic or semi-nomadic lifestyles that also share common sociocultural features such as clan structure, polygamous marriages, and a high level of endogamy (Karafet et al., 2002). Studies have shown a high level of heterogeneity between Siberian groups, the existence of a co-evolution among linguistic, genetic, and geographical variation, and a clear demarcation line between eastern and western Siberian populations.

Karafet et al. (2002), studied the paternal history of 18 Siberian groups using the Y chromosome. These authors found that the majority (96.4%) of Siberian haplogroups belong to four of the major haplogroups (N, C, Q, and R) defined by the Y Chromosome Consortium (YCC 2002). The most frequent haplogroup for Siberians fall into lineages that are widely distributed throughout Central Asia and Northern Eurasia although these data do not indicate any significant founder NRY haplogroups. Starikovskaya et al. (2005), investigated the maternal history of Siberia for 9 indigenous groups and found that the majority (66%) of the mtDNA's belong to the "Asian" macrohaplogroup M, with remaining lineages belonging to the Eurasian macrohaplogroup N. Derenko et al. (2002), also described mtDNA types belonging to macrohaplogroup R, which is another founder lineage thought to have been established with initial human migration into the Eurasian continent (based on coalescent times).

Many genetic studies that focus on Siberia tend to investigate the peopling of the Americas (Zegura et al., 2004; Starikovskaya et al., 2005), whereas the problems of initial human colonization of northern Asia fell by the wayside. Derenko et al. (2002, 2003, 2006, 2007a) has extensively studied the region of Siberia using phylogeographic analysis on mtDNA and Y chromosome to investigate the timing, origins, and routes of the founding migrations to Siberia. These authors conclude that southern Siberia is genetically diverse, exhibiting maternal and paternal lineages that are heterogeneously composed of both east and west Eurasian and Central Asian haplogroups (Comas et al., 1998; Wells et al., 2001; Quintana-Murci et al., 2004).

These data suggest two migrations into the Altai-Sayan region of southern Siberia, one from eastern Europe and the other from western Asia and/or the Caucasus

(Derenko et al., 2007a). Specifically, they present evidence for a distinct branch of haplogroup X in the mtDNA that was completely sequenced in 4 southern Siberian individuals. They found that this lineage (X2e) is highly diverged and could have been present in Siberia for ~14,000 years. To date, haplogroup X, thought to originate in the Near East, is not present in northern Siberian and eastern Asian populations.

Starikovskaya et al. (2005), along with Derenko et al. (2002) indicated a small percentage of "west" Eurasian haplogroups confined to the south-west part of Siberia, notably among the Tofalars (20.7%) and Yakuts (14.5%). These western Eurasian lineages suggest either ancient remnants of an Upper Paleolithic dispersal from the Middle East/Southwestern Europe that has not been erased by subsequent migrations and gene flow, or could as easily be attributed to more recent gene flow from women of European/West Asian ancestry occurring at the time of the expanding Mongolian Empire. Derbeneva et al. (2002) and Malyarchuk (2004) also detected western Eurasian lineages (specifically haplogroup U) in Northwestern Siberians. Haplogroup U has been found throughout Europe and has been dated to exceeding 50,000 yr BP.

Particularly, the Mansi, who speak a dialect of the Finno-Ugric language of the Uralic linguistic family, have a high frequency of mtDNA subhaplogroup U4 (16.3%), which Derbeneva et al. (2002) suggested may be indicative of the remnants of Upper Paleolithic populations of Europeans that have been preserved east of the Uralic mountains. U4 is widely distributed among groups inhabiting the Volga-Ural region and actually increases in frequency among groups living east of the Ural Mountains in northwest Siberia (Malyarchuk, 2004).

Among the Mansi, phylogenetic analysis suggested the appearance of the subhaplogroup U4 was most likely caused by its divergence from Eastern Europeans in the Late Upper Paleolithic (~18,500 yr BP). In addition, subhaplogroup U7, which is rare in European populations but exhibits low frequency in the Middle East has also been found in moderate frequency in the Mansi. It is believed that the isolation of groups inhabiting the region between the Ob' and Yenisei rivers was a key factor in the presence of these unique Eurasian lineages among groups from Siberia.

Derenko et al. (2003, 2007a) conceded southern Siberia to have been shaped by complex migration processes traced to Central, Eastern Asia, and Western Eurasia that have occurred since initial colonization, however, these authors do not consider Siberia to have been colonized by a northern route *vis a vis* the Near East. This finding is based upon complete sequence mtDNA using a phylogeographic approach that does not find any evidence for ancestral lineages to major Eurasian haplogroups M, N, and R in southern Siberia. It is however apparent that groups from Eastern Europe and the Near East had an impact upon the peopling of Siberia as evidenced from the maternal Eurasian specific-lineages in Northwestern groups and archaeological finds that date to the Upper Paleolithic with associated human assemblages that resemble European morphological features. This evidence of heterogeneity, as pointed out by Derenko and colleagues, may stem from more recent significant interactions dating post-Neolithic and/or Bronze Age. In addition, low population density and/or social organization practices (patrilineal clan associations) may have significant impact upon genetic studies carried out on modern populations.

The importance of notable expansions in Eurasia have been investigated by several researchers (Zerjal et al., 2002, 2003; Xue et al., 2005; Derenko et al., 2007b; Chaix et al., 2008) and has shown to have played a significant role in shaping the genetic history of northern and Central Eurasian groups. Zerjal et al. (2002) investigated the paternal history of a large sample of males from central Asia found, similar to previous research (Wells et al., 2001) a clear pattern of an ancient east to west gradient (cline) in Y chromosomal variation, but also that this gradient has been shaped by recent population-specific events. These events included significant migrations from the West, such as the Kurgan expansion dating to ~4,000 B.C., long distance trade along the Silk Road from the 2nd century B.C., and the expansion of the Muslim world starting in the 7th century A.D. Nomadic groups also contributed to several expansions, beginning with the Xiongnu nomadic steppe empire during the 3rd century B.C. (Keyser-Tracqui et al., 2003a, 2006), followed by the Turks in the 1st millennium A.D., and lastly, the Mongol expansions during the 13th century. These Eastern nomadic groups had significant impacts on populations as far west as Iran, Anatolia, the Caucasus, and even Europe (Calafell et al., 1996; Cinnioglu et al., 2004; Nasidze et al., 2004; Berkman et al., 2008).

The Mongol Empire and expansion during the 11th to 13th centuries had far reaching genetic consequences for peoples residing from the Caspian Sea to the Pacific Ocean (Zerjal et al., 2003; Derenko et al., 2007b; Malyarchuk et al., 2010). Zerjal et al. (2003) found that a specific Y chromosomal lineage with patterns suggesting origination in Mongolia ~1,000 years ago, and found at high frequency (~8%) in men ranging across Eurasia (~0.5% of world total), is ultimately caused by selective social pressure from male-line descendents of Chinggis Khan. These authors found that this particular paternal

lineage is found in geographical association of the boundaries of the Mongol Empire at the time of Chinggis Khan's death and his subsequent male-line descendent rulers located throughout parts of Eurasia. Derenko et al. (2007b) expanded on this earlier work by including groups from north Eurasia and found that the Chinggis Khan haplogroup (known as C3) is found in highest frequency among modern Mongols (34.8%), followed by Altaian Kazakhs (8.3%), Altaians (3.4%), Buryats (2.3%), Tuvans (1.9%) and Kalmyks (1.7%). Interestingly, these authors suggest that this central haplotype is present in almost every one of four male Mongols living today.

Even later events have significantly impacted the genetic structure of modern day populations residing in Northern Eurasia. Xue et al. (2005) identified a unique Y chromosomal lineage that is highly frequent in northeastern China and Mongolia (~3.3% of males sampled from East Asia). They conclude that the most recent common ancestor for this lineage lived ~600 years ago. They suggest this lineage was spread by Qing Dynasty (1644 – 1911) nobility, who were a Manchu privileged elite sharing patrilineal descent from Giocangga, the grandfather of Manchu leader Nurhaci. This is another example of the importance of novel social selection behaviors leading to significant genetic changes in a large sample of individuals. Although these are only two examples, the rarity of these novel selection processes in the literature may be overemphasized, and may indeed, actually have been more common in the past.

Concluding Remarks

The population history and structure of greater Eurasia is characterized by significant migration and admixture events since at least the Paleolithic, if not back further in time. In order to understand the extensive phenotypic variation seen today in

Eurasia, we need to be able to characterize the genetic diversity and the demographic processes that initiated the observed variation seen in living populations today. This chapter has outlined those major demographic shifts in order to interpret the results from this dissertation.

CHAPTER 4

THE XIONGU: ARCHAEOLOGICAL, HISTORICAL & GENETIC STUDIES

It is of utmost necessity to define groups in order to perform multiple analyses – whether for archaeological material culture such as pots, or discerning evolutionary relationships based upon craniofacial diversity. Peoples of the past, including non-literate societies such as the Xiongnu addressed in this dissertation, defined themselves on the basis of complex interactions and layered kin, cultural, social, and political identities. Most often, this expression of identity has been in the opposition to other groups. Ethnicity is one approach to an ascribed designation of identity. This chapter will discuss Xiongnu ethnicity as it pertains to Chinese historical narrative and archaeological material culture, as well as discuss Xiongnu population history from a genetic and bioanthropological perspective.

Xiongnu 'Ethnicity' and Chinese Narrative

Ethnicity can be defined as a culturally constructed identity associated with particular customs and *habitus* (Bordieu, 1977), often asserting common descent among its members (Jones, 1997). Materially defined archaeological cultures do not necessarily "map the extent and boundaries of self-conscious ethnic groups in the past" (Jones 1997:120). Though group differences might exist along lines of descent or ritual, they may share similar styles or artifacts that would define them as a single group. Further, as ethnicity is a concept rather than something that exists biologically, ethnic labels should not be directly equated with biological distinctness or similarity. These are just some of the difficulties correlating exact ethnic groups in the archaeological record with the material culture produced by those groups.

As a majority of information about the Xiongnu come from historical Chinese narrative (Miller, 2009), the use of 'others' is an important concept to understand. Literate civilizations often ascribe an 'others' term to a range of peoples that may be within their circle of contact, such as the Chinese and the 'barbaric' nomads who inhabited parts of northern China and Mongolia. As these labels were applied, modern researchers have tended to use them as ethnic monikers, often glossing over the differences in the people themselves and assume kin and cultural solidarity when, in reality, only social or political ties used for smaller groups were meant to describe these peoples. Those smaller groups that the literate civilization most often come into contact with are then used to apply to all those in that direction of vicinity.

For example, the cultural designation of the Wa by the Chinese applied to all peoples of the Japanese archipelago implying complete unification among those peoples, however, the only group that appear to have negotiated with the Chinese were the Yamatai, one polity among many in the area (Farris, 1998). The general label Scythian came to stand for peoples of the north as well as nomadic peoples (Miller, 2009). The Chinese term 'Hu' became conflated with nomadic steppe peoples to the north in the late first millennium BC (Di Cosmo, 2002).

Chinese accounts of the Xiongnu are often fluid and changed depending on the political dynamic at the time. Miller (2009) notes that foreign groups of northeast Asia became increasingly complex in the first millennium AD. In northeast China, some of the 'Hu' groups became known as the Xianbei, who it appears were subsumed under the label Xiongnu during the steppe polity's reign, but regained the label Xianbei again after the collapse of the Xiongnu empire. This one example illustrates the nature of multiple

ethnicities within the Xiongnu empire as political or military circumstances changed. If indeed, the Xiongnu leader who came before a Chinese court for negotiation purposes did declare that, among the steppe peoples north of the Great Wall, "all are Xiongnu" (Shiji 110:2896, in Watson, 1961), he would have suggested numerous tribes and distinct ethnic groups that were subsumed under the single name Xiongnu and were united into a single political unit under his control (Miller, 2009:53).

Historians and archaeologists alike have come to define the Iron Age pastoralists who lived and settled large parts of Inner Asia as an ethnic group and political formation that we modern-day researchers (and the Chinese in historical narrative) call the 'Xiongnu'. In historical Chinese narrative accounts of this group, the ethnic labels, tribal names and political designations become lost with the conflation of the more general term known as Xiongnu. Miller (2009) addressed this concern archaeologists may have with using such a general term to describe people in prehistory with no evidence of the written word. He suggested that by recognizing the political nature of the name Xiongnu as it is mentioned in Chinese texts - that is the designation of a political unit against which the Chinese struggled - then we should be able to use the term to discuss a political entity known as the Xiongnu and its elite agents delineated within the archaeological record.

Archaeological Evidence of the Xiongnu

Prior to the rise of the Mongolian Empire in the 13th century, little is known about the Mongols except there were many war-like tribes occupying present day Mongolia, alternating between large-scale empires and small-scale tribal organizations (Di Cosmo, 1994; 2002; Fletcher, 1986). The Xiongnu polity is the prototypical example of regional

political organization on the northeastern steppe, defined as the territories of Mongolia, South Siberia, and Inner Mongolia (Allard and Erdenebaatar, 2004; Honeychurch, 2003; Honeychurch and Amartuvshin, 2006; Keyser-Tracqui et al., 2003a, 2003b, 2006; Wright, 2006; Wright et al., 2009; Miller, 2009; Houle, 2010; Kim et al., 2010; Ricaut et al., 2010). The Xiongnu were a nomadic group contemporary with the Qin (221 – 07 BC), the Western Han (202 BC – 8 AD), and the Eastern Han (25 – 220 AD) dynasties of China. See **Table 4.1** for a list of Chinese Dynasties. The Xiongnu are among the first of many succeeding steppe polities to dominate the large geographic expanse of Inner Asia and specifically to control the core territory of modern day Mongolia (**Table 4.2**).

TABLE 4.1. List of Chinese Dynasties used in this dissertation.

Chinese Dynasty	Time Period
Eastern Zhou	770 - 256 BC
Warring States Period	475 - 211 BC
Qin	221 - 206 BC
Han	206 BC - 220 AD
Eastern Han	25 - 220 AD
Jin-Yuan	1115 - 1368 AD

TABLE 4.2. Chronology of steppe polities of Inner Asia.

Group/Designation	Time Period
Xiongnu	3rd century BC to 2nd century AD
Turk	6th - 8th centuries AD
Uighur	8th - 9th centuries AD
Khitan	10th - 12th centuries AD
Mongol	13th -14th centuries AD
Manchu	17th - early 20th centuries AD

This steppe zone is a diverse environment and contains various vegetation, lake and river systems, mountains and deserts. It is in this ecological zone that the Xiongnu people originated, although at its height, the empire is reported to have directly or indirectly controlled territory from Manchuria to Kazakhstan, southern Siberia to Inner

Mongolia, and to the Tarim Basin of present day Xinjiang Province in western China, home of the Silk Road (Beckwith, 2009). Using historical evidence and accounts provided by Chinese sources, archaeologists have been able to define a "Xiongnu" material culture based on consistency in burial type and artifacts recovered in a mortuary context (Honeychurch and Amartuvshin, 2006; Wright, 2006).

Much of what is known archaeologically comes from mortuary research and burial data excavated in Mongolia and the Zabaikal'e region (**Fig. 4.1**), located along the Selenge River valley to the shores of Lake Baikal in southern Siberia (Allard et al., 2002; Murail et al., 2000; Crubezy et al., 2006; Wright et al., 2009). Note that this figure is for heuristic purposes only, showing where the majority of archaeological research has been conducted. Wright (2006) has suggested that over 2000 tombs of various sizes have been excavated. However, Miller (2009) explains the limitations of the archaeological data. First, the majority of documented material consists of graves of significant size or within prominent burial grounds. This observation limits our ability to interpret mostly elite members within the Xiongnu polity. Second, of the sites excavated in Mongolia and South Siberia, many have no published report, few reports of excavations, and only three have significant reports of excavation and documentation. More difficult is the provincial nature of these reports – many are only written in the national language of the excavators, such as French, Russian, or Mongolian.

FIGURE 4.1. Early Iron Age and Xiongnu Archeological Sites (Adapted from Honeychurch, 2004, pp. 67).

Although variable, Xiongnu material culture has been radiocarbon dated and a firm chronological framework established (Hall et al., 1999). This material culture includes evidence for a complex and large-scale polity of pastoral nomads. Excavation of large cemeteries and settlement sites have shown distinct Xiongnu ceramics, paleobotanical remains, metalwork, and skeletal remains of sheep, cattle and horses (Wright et al., 2009).

The Xiongnu burials reveal a hierarchy of scale and mortuary style and complexity. Large, royal Xiongnu tombs were immense constructions tens of meters

square and deep (Wright, 2006). These types have been found only at the largest cemeteries. The most common grave associated with Xiongnu material culture are stone ring burials between five and ten meters in diameter with a central shaft two or more meters deep at the center, usually containing a wooden or stone coffin, though many of these have been disturbed over the years (Wright, 2006). The majority of interments are adults, with a single individual or sometimes double burial.

Xiongnu graves are normally found in groups, ranging in size from a few burials to hundreds of graves of various sizes. Within the core area of Xiongnu control (central and northern Mongolia), three-level size hierarchies appear within the defined cemetery types (Honeychurch, 2003). The first are large cemeteries containing massive square tombs and hundreds of associated ring graves, including Khunnigol, Noyon Uul, Tsaram and Gol Mod. The second rank cemeteries include the so called 'hundred grave cemeteries' such as Borkhan Tolgoi in the Egiin Gol and Baga Gazarynn Chuluu in the Middle Gobi. Surrounding these second level cemeteries are smaller, more localized cemeteries, with spatially distinct burial locales with less than a dozen graves.

Wright et al. (2009) suggest a regional system of hierarchy and political organization as evidenced in the material remains of grave goods found in both smaller cemeteries (Borkhan Tolgoi) and lager elite cemeteries, such as Noyon Uul (Polosmak et al., 2007) or Gol Mol 2 (Allard et al., 2002). These non-local connections seem to connect inhabitants of smaller settlement sites to a larger system of external decision-making. Grave goods, such as silks, jade items, bronze mirrors and Chinese lacquer indicate a tribute system in payment by Chinese rulers to Xiongnu elite. Chinese historical sources also indicate such a relationship (Christian, 1998). However, the

archaeological evidence points to a more complex and sophisticated exchange network. At Noyon Uul, material and textual evidence suggest the Xiongnu elite also developed a system of exchange with Bactrian origins in Central Asia (Honeychurch and Amartuvshin, 2006).

Another interesting aspect to the archaeological research conducted in Mongolia concerns the distinctive mortuary and monumental transition that occurred during the Eurasian Late Bronze Age and Early Iron Age. During this period, Mongolia witnessed the emergence of three monumental forms and features that were associated with changes in social relations, technologies, and the broader socio-political setting of the time (Allard and Erdenebaatar, 2005). These mortuary forms are known as khirigsuurs, slab burials, and Xiongnu ring tombs. Khirigsuurs have been dated to the late second and early first millennium BCE, and are ubiquitous throughout Mongolia, although they are better represented in the western Altai mountains (**Fig. 4.2**). Slab burial assemblages have been stylistically dated from the terminal second to the mid-first millennium BCE and are more numerous on the eastern plains of Mongolia (**Fig. 4.3**). Xiongnu ring tombs have been dated to a range between the fourth century BCE to the third century AD and are found in both Mongolia and southern Siberia (**Fig. 4.4**). Ring tombs were used for both commoner and elite (Wright et al., 2009), and substantial differences are exhibited in grave goods, size, and depth of tombs (ranging from 1 meter to 10 meters or more).

All monumental features have supported chronologies and overlap in time and space. According to some authors (Volkov, 1967; Erdenebaatar, 2002), the construction of the khirigsuurs and slab burials were performed by differentiated cultural groups from western and eastern Mongolia, respectively. Slab burials were supposedly left by an

indigenous eastern and central group while khirigsuurs are the remnants of an intrusive group from the west with cultural ties to the central Asian kurgan building peoples (Erdenebaatar, 2002). The Xiongnu ring tombs are thought to have emerged from the slab burials or were joined within the growing Xiongnu polity (Honeychurch and Amartuvshin, 2006). If more data were available, craniofacial variability may contribute to answering these monumental transitions. However, very little skeletal data is available for the people who contributed to building of the khirigsuurs, while most of the Bronze Age material (slab grave skeletal remains) is too degraded for analysis.

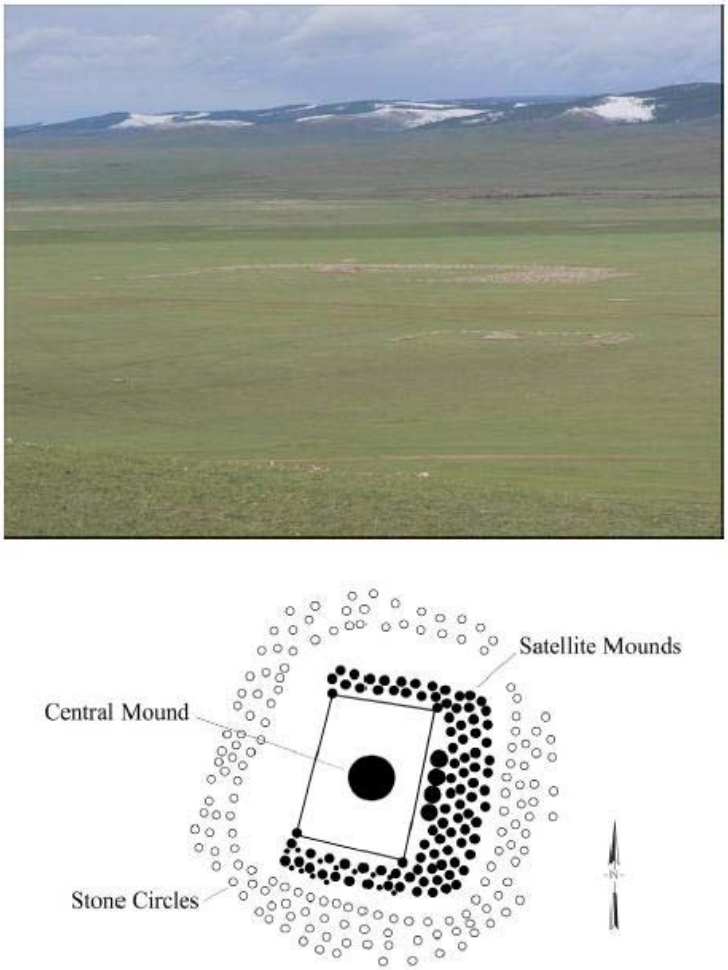

FIGURE 4.2. Photo and schematic of a *Khirigsuur* (Houle, 2010, pp. 13).

FIGURE 4.3. Photo of a slab burial.

FIGURE 4.4. Photo of Xiongnu ring tomb.

Formation of the Xiongnu Polity

A quantitative assessment of biological variation is needed to better understand the nature of human evolutionary relationships among steppe populations and could potentially explain archaeological, genetic and cultural models of steppe polity formation. For example, proposed historical and archaeological explanations for the organization of large-scale confederations originating on the steppe are known as core-periphery framework models, in that change occurred along peripheral regions on processes operating within a mature state (Beckwith, 2009). This would necessitate contact with peoples from that mature state, namely sedentary peoples on China's frontier. An alternate explanation to steppe polity formation challenges the core-periphery view and attributes change and development of steppe polities to actions taken among steppe groups themselves (Honeychurch and Amartuvshin, 2006). According to this model, although steppe polities may be diverse and self-sufficient, they are incapable of supporting large-scale confederacies without external income, in this case trade and exchange through interregional interaction.

Both of the above models have at their core a degree of external influence in maintaining and controlling a newly-formed confederacy. According to Chinese historical sources (Christian, 1998), the Xiongnu polity was built up around the dominance and conquering abilities of it's founder, Moton, who invaded the Ordos region of present day Inner Mongolia in China. In 198 BCE, the Han Emperor, Kao-tsu, negotiated the first of several treaties with the Xiongnu by agreeing to supply the nomads with regular gifts, and even a marriage proposal between Moton and a Han royal princess (Christian, 1998:186). These treaties allowed relative stability to the Chinese, and support

from China allowed the Xiongnu to control the pastoralist groups of the Mongolian steppe.

Honeychurch and Amartuvshin (2006) proposed another explanation for steppe polity consolidation that is an extension of the two traditional models of state formation. They recognize that steppe cultures were imbued with an emphasis on mobility and horse-based warfare that gave them experience in organizing activities, resources, and control of peoples over substantial spatial distances. This experience then facilitated long-distance interaction and exchange, but more importantly, the ability to effectively manage diverse peoples, languages, and cultures. These transport technologies put in place strategies for controlling the logistics and diversity of large-scale polities. Over time, these political traditions created continuity between different confederations and empires. This explanation takes into account trading and exchange with China, however, China's influence was unnecessary in maintaining the Xiongnu confederation. This is important because steppe confederation and formation continued into the early 20th century with the rise of the Manchu Empire in China.

Molecular and Bioanthropological Studies of the Xiongnu

Though restricted mainly to archaeological studies of the Xiongnu steppe polity, other researchers have begun to explore the human remains from Xiongnu settlements in an effort to define the people who composed the groups known as the Xiongnu. Many of these studies use ancient DNA taken from cemeteries to explain population history and structure; or only study a few individuals to describe some aspect of pathology. However, these efforts are mainly restricted to local evolutionary processes, and most do not make larger regional connections.

Several studies by molecular biologists have analyzed ancient remnants of DNA from teeth and bone from larger Xiongnu settlement sites and cemeteries located in Mongolia. Keyser-Tracqui et al. (2006) analyzed ancient DNA for a small (42 individuals) skeletal sample from Egiin Gol, located in northern Mongolia, and compared it to present-day Mongolian populations located in the same region (along the Selenge River, a main tributary of Lake Baikal), and a small sample of Yakuts, a pastoral people who inhabit areas of the Sakha Republic in eastern Siberia. It is widely believed that the Yakuts were the first settlers of the Altai-Baikal region of east Siberia (Amory et al., 2006).

The results for the autosomal and Y-STR loci indicate a close biological relationship among the ancient Egiin Gol sample and modern Mongolian samples, with some genetic distance between all the Mongolian samples and the Yakuts as measured by F_{ST}. Even after including Turkish data (historically linked with the Mongols and linguistically to the Yakuts), the Yakuts still formed a distinct cluster among all the samples under analysis. mtDNA analysis showed similar results, however, minimal haplotype sharing did exist among the ancient Xiongnu and modern Yakuts. Thus, these authors posit minimal genetic substructuring between different Mongolian populations reflecting the maintenance of a common genetic pool and an unclear relationship between Xiongnu and Yakuts.

Kim et al. (2010) extracted ancient DNA from three Xiongnu skeletons in the elite cemetery of Duurlig Nars. These authors were able to extract mtDNA, Y-SNPs and autosomal short tandem repeats (STR) and found for one male skeleton the presence of a distinct paternal Indo-European lineage known as haplogroup R1a1. Some authors (Zerjal

et al., 2002; Haak et al., 2008) have suggested the R1a1 haplogroup to be associated with the Kurgan expansion model, which explains the origin and eastward migration of Indo-European speaking peoples from the Volga region in modern-day central Russia. R1a1 is the most common paternal haplogroup in Europe (Malyarchuk et al., 2004) and shows decreasing frequencies from northern to southern Europe, and from central to south Asia (Wells et al., 2001; Cordaux et al., 2004). It has also been found in high frequency in India (Sharma et al., 2009). This male also had the mtDNA haplogroup U2e, which is found mostly in central Asian populations (Comas et al., 2004). These findings tentatively support the archaeological evidence for not only material exchange, but also potentially mate exchange between the Xiongnu and Central Asian groups.

Interestingly, researchers working in southern Siberia have also found evidence for the R1a1 haplogroup in a sample of ancient remains from the Krasnoyarsk area dated from between the middle of the second millennium BC to the fourth century AD (Keyser et al., 2009). Using phenotype-informative single nucleotide polymorphisms, these authors even suggest that this region in south Siberia was predominately settled by Europeans who had blue eyes, fair skin and light hair.

This finding and the evidence provided by Kim et al. (2010) should not come as a great surprise considering Indo-Europeans were most likely resident in Northwest China close to 4,000 years ago (Yao et al., 2004). According to this observation, then, it would suggest the Western Eurasian Xiongnu male in Kim et al.'s (2010) study was extant to Xiongnu homelands for approximately 2,000 years. It is also not surprising when the effect of the horse and transportation make interactions all the more common (Beckwith, 2009). Though this genetic evidence is promising, a phenotypic assessment is needed to

validate these findings. If the hypotheses outlined in this dissertation are rejected (evidence for biological discontinuity), then the alternative explanation for substantial biological exchange between the Xiongnu elite and Eurasian groups would be strengthened.

Tumen (2006), studied multiple temporal periods (Neolithic to the present) in Mongolia, and using morphological (craniometric) variation as evidence, concluded extensive heterogeneity of morphological diversity of Mongolian, and especially Xiongnu samples. However, discussion is characterized as typological, placing Mongolian population history into a few discrete categories, including "Caucasoid" and "Mongoloid". Tumen's (2006) research did suggest extensive migration from nomads on the eastern Eurasian steppe as accounting for the greater diversity during the Xiongnu period.

Lee (2007), used a large and varied dataset (China, Korea and Mongolia) of cranial and dental nonmetric traits, attempts to characterize the genetic exchange and interaction among Chinese and Mongolian samples. Lee (2007) concluded differential evolutionary history as seen in the morphological traits: dental traits show a deeper evolutionary history while cranial nonmetric traits represent a more recent history of population interaction between the central plains people of China and individuals making up the Xiongnu empire after its collapse. Lee (2007) also proposed biological continuity as reflected in both dental and craniometric data for the northern Xiongnu to modern Mongolians. This research would greatly benefit from the inclusion of more stringent models to explain morphological diversity. Specifically, the inclusion of a population genetic model could potentially strengthen, or weaken, the argument for increased gene

flow and migration between the Xiongnu and neighboring China. A population genetic approach could also strengthen the argument, and the hypothesis proposed here, for biological continuity from the Iron Age to the present.

More recently, Ricaut et al. (2010) compared non-metric trait data in the form of dental traits with genetic data for the Egiin Gol necropolis. This study was done in order to assess the usefulness of non-metric trait data to detect familial groupings in the absence of available genetic data. Therefore, the study is small-scale, and makes no attempt at reconstructing the regional history of the Xiongnu. However, interestingly, their results show that this population was highly homogenous, similar to previous studies (Keyser-Tracqui et al., 2003a, 2003b), indicting the necropolis was occupied by the same people over its continuous five centuries of use (300 BC – 200 AD). The Egiin Gol necropolis (Borkhan Tolgoi) is located in northern Mongolia.

The site has been extensively investigated by a team of French-Mongolian researchers (Crubezy et al., 1996; Keyser-Traqui et al., 2003a; Ricaut et al., 2010), containing the skeletal remains of 99 individuals and dates from the third century BC to the second century AD. The necropolis was organized into three main sections (A,B,C) that have been AMS carbon-14 dated. The oldest part of the cemetery is sector A, followed by B and then C. The development of sector C corresponds to the end of the necropolis's use and appears to reflect a Turkish influence on the Xiongnu (Keyser-Tracqui et al. 2003a). This finding is based on several Y-STRs found in present-day Turkish individuals (Henke et al., 2001). The Ricaut et al. (2010) study also found, using nonmetric traits, a distinction in sector C, indicating a possible demographic transition toward the end of the Xiongnu empire.

Several indicators suggest the cemetery of Borkhan Tolgoi represented only a subset of the Xiongnu community, who appear to have been high-status individuals. These include low-burial frequency, funerary artifacts, elaborate practices, including the use of coffins and chests, and the depth of the graves (two to five meters). The genetic analysis performed by Keyser-Tracqui et al. (2003a, 2003b) found that the majority of the Xiongnu mtDNA sequences belong to predominately Asian haplogroups, however a few (11%) belong to predominately Europeans haplogroups. This would suggest that European and Asian contacts were being made prior to the development of the Xiongnu culture, as seen in other studies of the region (Clisson et al., 2002; Keyser et al., 2009; Bennett and Kaestle, 2010; Kim et al., 2010).

The Egiin Gol valley also has been extensively investigated and researchers have found sites composed of kurgan-style graves and range in time from the Bronze Age until the period of Chinggis Khan. Crubezy et al. (1996) discuss an interesting finding in the Egiin Gol valley related to the practice of kurgan graves. Kurgans (a Russian word for tumuli) are barrows characteristic of a culture arising on the steppes of southern Russia around 5000 BC and later spread into eastern, central, and northern Europe between 4400 and 2800 BC (Keyser et al., 2009). Most of the kurgan style graves found in the Egiin Gol valley date to the Bronze Age, however Crubezy et al. (1996) described an isolated kurgan dated to around the 9th century AD, suggesting a Uighur origin.

The Uighur empire was founded by a Turkic tribe in 744 AD and fell in 840 AD after its capital in the Orhon valley of Mongolia, fell to a Turkic group of Kirghiz (Crubezy et al., 1996). This finding may be related to those individuals buried in sector C of Borkhan Tolgoi who, genetically, appear to have several paternal genetic signatures

linking them to modern Turks (Keyser-Traqui et al., 2003a, 2003b; Ricaut et al., 2011). Of importance to this dissertation, however, is the fact that, archaeologically, there is a material and cultural (and perhaps genetic) connection from Bronze Age Mongolia through the Uighur/Turk period, at least for the Egiin Gol valley, and perhaps throughout Mongolia during that period as well.

CHAPTER 5

MATERIALS

The materials used in this dissertation are from osteological collections located in museums and universities in China, Mongolia, France, Russia, Japan, and the United States. Coordinate data was collected in the form of three-dimensional landmarks. Original data were observed on a total of 1558 adult crania. Sample names, sizes, geographic coordinates, period, and institutions are shown in **Table 5.1** and **Figure 5.1**. To investigate Xiongnu population history and structure, a large comparative series of crania was assembled, reflecting various temporal periods and geographic locations. Importantly, these comparative cranial series should be sufficient to test this dissertation's hypotheses of Xiongnu interaction. To this end, samples were collected in order to test interactions among Chinese, Siberian, and Central Asian populations. Sample sizes vary depending on preservation, however, most samples used in final analyses consist of at least 10 individuals per population.

The majority of Mongolian samples come from two institutions in Mongolia: the National University of Mongolia (NUM), and the Mongolian Academy of Sciences, Institute of Archaeology (MAS), both in Ulaanbaatar, the capital of Mongolia. Total Xiongnu crania (n=68) were sampled from several locations in Mongolia. Many of the Xiongnu crania come from a location in Northern Mongolia at a site named Egiin Gol, specifically the cemetery of Borkhan Tolgoi. Various other samples were pooled from several sites around Mongolia. In addition to the Xiongnu, Mongolian Bronze Age, Medieval Period (pooled), and modern period crania (pooled) were sampled. The total Mongolian cranial series is shown in **Table 5.2**.

Geographic coordinates for samples were obtained using Google Earth™. Although site information was available for some samples, namely in China and Mongolia, most site information from museum specimens is unknown. When site location was unknown, a more general geographic coordinate was attributed to the sample. For example, the Kazakhstan sample from the Musee de l'Homme, was given general geographic coordinates (Astana, the capital city), rather than site-specific location. In China, however, most samples from Jilin University consisted of exact GPS location. In these cases, the geographic coordinate in **Table 5.1** reflects the actual site where the skeletal material was collected (provenience). Most Mongolian samples had little site information, other than temporal period and general geographic location (Western Mongolia, for example). When more specific information was included with the skull, this was used as the geographic coordinate. For example, Egiin Gol was given the coordinates 50.22045N, 100.32138E (Northern Mongolia), while the Mongol sample from the American Museum of Natural History was given the coordinates for Ulaanbaatar City, the capital of Mongolia.

Sex of crania were estimated using standard morphological criteria of the skull (Buikstra and Ubelaker, 1994). If sex could not be estimated from standard macroscopic investigation, the skull was given an unknown status. Only adult crania were recorded. Juvenile crania were avoided, as were intentionally modified crania.

Landmarks: Investigators studying geometric morphometrics normally collect data that represent biological form such as length measurements, the arrangement of morphological landmarks, or entire outlines or surfaces of the specimens (Klingenberg, 2010). The most widely used approach, and the one used in this dissertation, is to

represent organismal forms (crania) by landmarks. In biological terms, homology describes the functional correspondence between morphological features. In the field of morphometrics, landmarks, being geometric locations, must contain valid biological information about shape and form from individual to individual. Landmarks are points that can be precisely located on all biological forms, and also establish a clear one-to-one ratio between all specimens included in the study. Bookstein (1991) describes three different types of landmarks that can be collected from biological forms. Research has shown that precision of landmark location for human crania varies between these three different types (von Cramon-Taubadel et al., 2007; Ross and Williams, 2008; Sholts et al., 2011). These include Type I (sutural intersections such as bregma), Type II (geometric maxima at bony protrusions or depressions), such as jugale, and Type III landmarks (external locations with respect to other geometric entities), such as glabella.

A total of 44 landmarks were digitized for each skull (**Table 5.3, Fig. 5.2**). Homologous landmarks were chosen for their ability to reflect biological form between specimens and as a way to capture variability of craniofacial form. In addition, most of these landmarks were chosen as endpoints for commonly collected linear measurements for easy conversion from 3D to 2D analyses. Landmarks were ultimately narrowed to 24 homologous points in the final analyses due to missing data, poor preservation, or were excluded because of Type III landmarks, which have been found to contain greater coordinate measurement error (**Table 5.4**). The landmarks used in final analyses are mostly Type I and Type II.

All coordinate data was observed using a MicroScribe G2X portable digitizer (Immersion Corporation, San Jose, CA) connected to a Toshiba laptop using associated

software. Data collection protocol proceeded as follows: skull was placed into a bed of beads to stabilize and minimize movement; skull was placed into beads in a medio-lateral fashion, positioning the skull so that the occipital bone was resting flat in the beads; skull was re-positioned with basion facing up to take the remaining landmark measurements following a re-orientation of established points (nasion, prosthion, frontomalare orbitale). Landmarks were stitched together using the Immersion software. For my purposes, landmarks 1-39 were first digitized, skull was re-oriented, followed by landmarks 40-44. Following data formatting, all subsequent analyses were carried out using the analytical software package *MorphoJ* (Klingenberg, 2011).

TABLE 5.1. Comparative cranial series not including Mongolian samples.

Region	Sample Name	Sample Size			Latitude	Longitude	Period	Institution
		Male	Female	Unknown				
East Asia	Ainu	46	24		43°3'52.61	141°20'48.51	Modern (1900 CE)	UTOK, AMNH
	Japan	6	4		36° 0'0.00	138° 0'0.00	Modern	AMNH
	Jomon	22	12	12	36° 0'0.00	138° 0'0.00	14000-300 BCE	UTOK, TSM
	Taiwan	2			25° 5'27.87	121°33'35.40	Modern	AMNH
	Tibet		1		29°38'52.62	91° 7'1.22	Modern	AMNH
China	Gansu	1		2	38°38'32.35	100°46'24.57	2070-1600 BCE	JIDA
	IM Bronze Age	26	11		43°2'14'.40	118°19'12.31	1600-1046 BCE	JIDA
	IM Eastern Han	12	5		49°39'44.30	117°19'47.62	25-220 CE	JIDA
	IM Eastern Zhou	13	8	3	40°14'17.76	112°4'34.00	771-221 BCE	JIDA
	IM Warring States	40	41		40°32'43.60	111°48'45.17	475-221 BCE	JIDA
	IM Yuan	7	10	4	42°17'8'.36	116°14'58.90	1271-1368 CE	JIDA
	Jin	12	11	1	37°52'24.15	112°33'45.25	265-316 CE	JIDA
	Liaoning	25	33		41°50'7.59	123°25'45.98	1600-1700 CE	JIDA
	Neolithic China	5	3	1	34°54'49.42	113°32'26.49	5800-5400 BCE	JIDA
	Qinghai	38	42		36°67'48.44	101°75'18.61	206 BCE-316CE	JIDA
	South China	23	5		23° 7'44.99	113°15'51.97	Modern	AMNH
	Tientsin	8	2		39° 5'2.97	117°12'3.54	Modern	AMNH
	Tungku	5	1		39° 5'2.97	117°12'3.54	Modern	AMNH
	Xinjiang Bronze	14	16		42°84'16.51	93°50'21.21	2000-1500 BCE	JIDA
	Xinjiang Han	28	31		41°33'81.78	86°26'36.27	206 BCE - 8 CE	JIDA
	Xinjiang Modern	9	4	1	43°66'57.23	90°12'55.91	300 BCE	JIDA
Siberia	Buryat	10	12		53°59'55.35	112°53'30.71	Modern	MSU
	Chuckchi	4	5		69°41'50.07	170°22'33.19	Modern	AMNH
	Evenks	7	11		56°54'26.21	91°51'37.90	Modern	MSU
	Iron Siberia	22	20		56°27'50.36	84°57'45.17	700-200 BCE	RASN
	Iron Tuva	8	13		51°53'14.16	95°37'33.66	700-200 BCE	RASN
	Kalmyk	17	20		46°34'3.66	45°46'23.38	Modern	MSU
	Orochi	8	9		48°28'36.19	135° 5'38.65	Modern	MSU
	Pazyryk	33	26		50°37'5.49	86°13'11.75	600-300 BCE	RASN
	Siberia Bronze	8	9		56°27'50.36	84°57'45.17	4500-700 BCE	RASN
	Tagar	12	15		53° 2'42.82	90°23'53.57	700-300 BCE	RASN
	Tuva	21	21		51°53'14.16	95°37'33.66	Modern	MSU
	Ulchi	5	12		51°56'41.67	140°24'51.67	Modern	MSU
	Volga Region	24	12		51°31'59.77	46° 2'4.38	Modern	MSU
	West Siberia	30	25		56°27'50.36	84°57'45.17	Modern	RASN
	Yakut	16	19		66°45'40.84	124° 7'25.51	Modern	MUSE
Central Asia	Baluchistan	1	1		28°29'26.64	65° 5'44.80	Modern	AMNH
	Chuvash	10	9		55°29'19.97	46°57'50.54	Modern	MSU
	Kazakh	7	8		48° 1'10.46	66°55'25.26	Modern	MUSE
	Kyrgyz	15	15		41°12'15.77	74°45'57.95	Modern	MSU
	Turkmen	14	6		38°58'10.99	59°33'22.60	Modern	MSU, MUSE
	Uighur	11	10		43°47'34.90	87°37'40.12	Modern	MSU
	Uzbek	13	9		41°22'38.97	64°35'6.94	Modern	MSU, MUSE
South Asia	India	9	4		22°59'12.33	87°51'17.91	Modern	AMNH
	Iran Bronze			13	32°25'40.47	53°41'16.97	4000-1000 BCE	UTOK
	Singapore	13	7		1°21'7.50	103°49'11.41	Modern	AMNH
	Tamil	2	1		7°52'22.99	80°46'18.47	Modern	AMNH
	Thailand	12	12		13°43'24.31	100°28'34.44	Modern	AMNH
Europe	Austria	2	4		47°30'58.43	14°33'0.26	Modern	AMNH
	Czech	7	8		49°49'2.97	15°28'22.66	Modern	AMNH
	Faroe Islands	3	1		61°53'33.49	6°54'42.50	Modern	MUSE
	Norway	4	0		60°28'19.29	8°28'8.21	Modern	MUSE
	Sweden	6	5		60° 7'41.38	18°38'36.60	Modern	MUSE
Africa	Bushmen	2	5	1	22°19'42.51	24°41'5.52	Modern	AMNH
	Zulu	6	1	1	22°19'42.51	24°41'5.52	Modern	AMNH
Totals:		**704**	**599**	**39**				

AMNH: American Museum of Natural History, New York, USA
JIDA: Research Center for Chinese Frontier Archaeology, Jilin University, Changchun, China
MSU: Moscow State University, Moscow, Russian Federation
MUSE: Musee d l'Homme, Paris, France
RASN: Institute of Archaeology and Ethnography, Russian Academy of Sciences, Siberian Branch, Novosibirsk, Russian Federation
TSM: National Museum of Nature and Science, Tokyo, Japan
UTOK: University Museum, University of Tokyo, Tokyo, Japan

FIGURE 5.1. Comparative cranial series locations used in this dissertation (African samples not shown).

TABLE 5.2. Mongolian Cranial Series.

Sample Name	*Sample Size*			*Location*	*Period*	*Institution*
	Male	**Female**	**Unknown**			
Chandman	14	19	2	Central, Western Mongolia	700-400 BCE	MAS, NUM
Pooled Mongol Modern	30	38	3	Mongolia	Modern (1900 CE)	NUM, MASUB, AMNH, MSU, MUSE
Pooled Mongol Period	15	15	3	Eastern, Central, Northern	1100-1500 CE	MAS, NUM
Mongol Turk			9	Central Mongolia Eastern, Central, Northern	1300-1050 CE	MAS
Pooled Xiongnu Period	27	12		Western	209 BCE - 93 CE	NUM
Egiin Gol Xiongnu			29	Northern Mongolia	209 BCE - 93 CE	MAS
Totals:	**86**	**84**	**46**			

MAS: Mongolian Academy of Sciences, Institute of Archaeology, Ulaanbaatar, Mongolia
NUM: National University of Mongolia, Ulaanbaatar, Mongolia

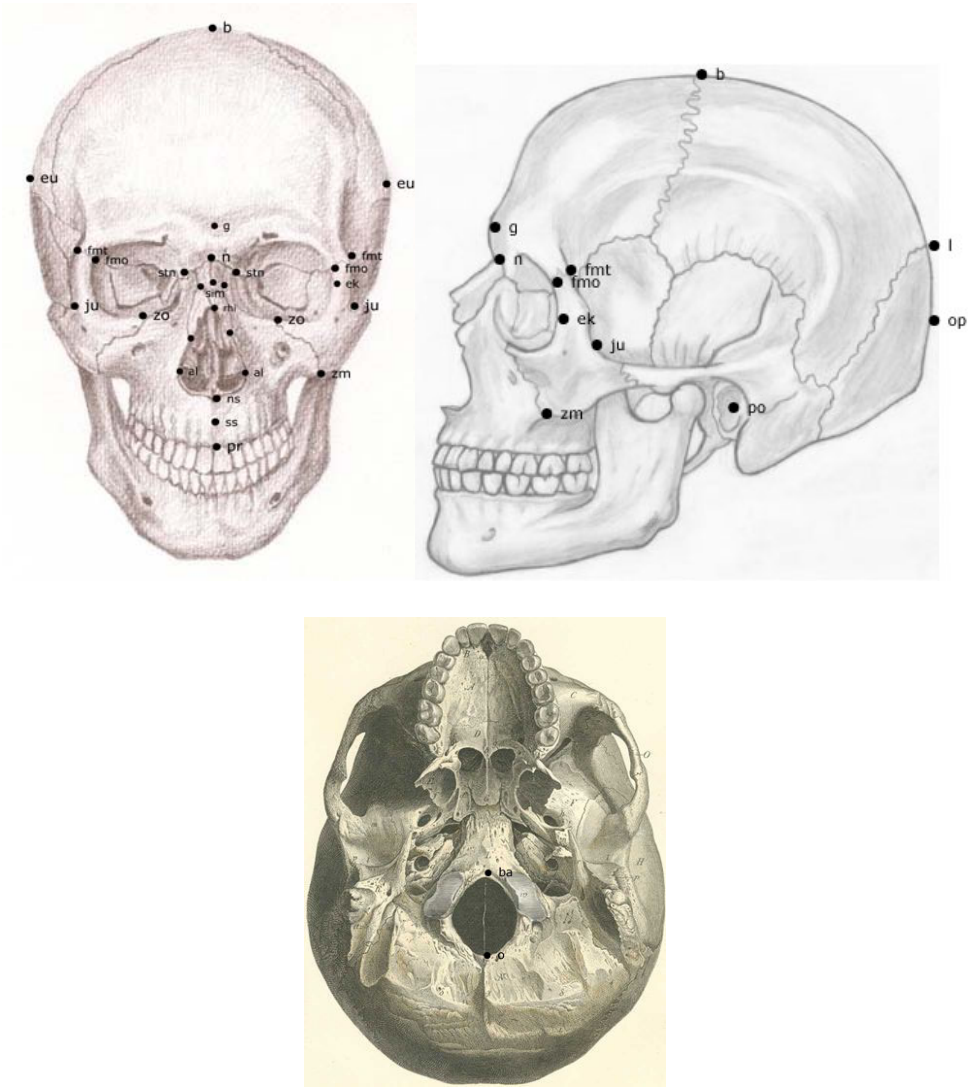

FIGURE 5.2. Landmark abbreviations in anterior, lateral and inferior views.

TABLE 5.3. Description of Craniofacial Landmarks.

Landmark	Abbreviation	Description*
Porion (L)	PO	Slight different from porion. Measurement was taken centered on the external auditory meatus.
Porion (R)	PO	
Nasion	N	Intersection of the nasofrontal suture with the midsagittal plane.
Ectoconchion	EK	Known as 'EC' in Bass (1995). The point where the orbital length line, parallel to the upper border, meets the oute
Zygoorbitale	ZO	Point of articulation between the zygomaxillary suture and the lateral and inferior margin of the orbit.
Subspinale	SS	Point of the nasal aperature below the nasal spine.
Zygomaxillare	ZM	Most inferior point of the zygomaticomaxillary suture.
Prosthion	PR	The most anterior point in the midline on the upper alveolar process.
Bregma	B	The intersection of the coronal and sagittal sutures, in the midline.
Nasal Sill (L)		Bottom of nasal aperture. Measured at most inferior point of anterior nasal aperture.
Nasal Sill (R)		
Superior Terminate at Nasomaxillary Suture (left)		Taken at the most superior point along the nasomaxillary suture.
Superior Terminate at Nasomaxillary Suture (right)		
Jugale (L)	JU	The union of the fronal and temporal processes of the zygomatic bone.
Jugale (R)	JU	
Nasospinale	NS	The point where a line drawn between the lower margins of the right and left nasal apertures is intersected by the
Simotic left		Simotic measurements taken at the simotic chord, the minimum horizontal breadth of the two nasal bones, taken a
Simotic middle		
Simotic right		
Alare (L)	AL	Instrumentally determined most lateral point on the nasal aperture taken perpendicular to nasal height.
Alare (R)	AL	
Frontomalare Temporale (L)	FMT	The most laterally positioned point on the frontomalar suture.
Frontomalare Posterior (L)	FMO	The most posterior point on the frontomalar suture that is not in the temporal fossa.
Frontomalare Temporale (R)	FMT	
Frontomalare Posterior (R)	FMO	
Orbitale (L)	OR	The lowest point in the margin of the orbit.
Orbitale (R)	OR	
Glabella	G	The most forward projecting point in the midline of the forehead at the level of the supra-orbital ridges and above
Euryon (L)	EU	Point determined instrumentally at the most widely separated points on the two sides of the skull.
Euryon (R)	EU	
Basion	BA	The midpoint of the anterior margin of the foramen magnum most distant from bregma.
Opisthocranion	OP	The most posterior point on the skull not on the external occipital protuberance.
Lambda	L	The intersection of the sagittal and lambdoidal sutures in the midline.
Inion	I	The point at the base of the external occipital protuberance.
Opisthion	O	The midpoint of the posterior margin of the foramen magnum.

*Descriptions from Bass (1995); Martin and Saller (1957); Brace and Hunt (1990)

TABLE 5.4. Landmarks used in final analyses.

Landmark
Nasion
ek(a) (left)
ZO (left)
Zm (a) (left)
Pr
bregma
Nasal sill (left)
superior terminate at Nasomaxillary suture (left)
superior terminate at Nasomaxillary suture (right)
ju (left)
simotic left
simotic middle
simotic right
nasal breadth left
nasal breadth right
fmt (left)
fmo (left)
fmo (right)
fmt (right)
midorbital width Zo (left)
glabella
basion
OP
l (crosspoint of lambodidal and sagittal suture)

CHAPTER 6

ANALYTICAL METHODS

This chapter outlines this dissertation's analytical methodologies employed to better understand the origins of the Xiongnu nomadic population. To this end, Xiongnu population history, structure and origins were investigated using three-dimensional craniometric data analyzed in a quantitative genetic context. The investigation of population history and structure of the Xiongnu was performed using a suite of geometric morphometric methods, quantitative genetics, and traditional multivariate statistical analysis. This research will first test the population structure of the Xiongnu followed by testing biological interactions of the Xiongnu with various other regional populations through time and space. This includes an analysis for comparison with the global series, followed by separate analyses with only the Chinese series, then the Central Asian series, and finally, with the Siberian series. This work is accomplished though several layers of analytical procedures.

General Description of Analyses

The following procedures were performed for all levels of analyses (Within-group Mongolian variation; Mongolian series vs. Global series; Mongolian series vs. Chinese series; Mongolian series vs. Central Asian series; Mongolian series vs. Siberian series) and are described in greater detail below. All individuals (crania) were first subjected to Procrustes superimposition (*described below*) to scale, translate, and rotate for further statistical analysis using the software program *MorphoJ* (Klingenberg, 2011). An outlying procedure was then used in *MorphoJ* to exclude individuals whose landmarks deviated strongly from the consensus after Procrustes superimposition. Only a few

individuals were excluded in this process. This was followed by generating a total covariance matrix. A principal components analysis was then performed on the total covariance matrix to generate residuals (coefficients) for further microevolutionary testing. Principal component scores that accounted for 95 percent of the variation (normally around 30 PCs) within the samples were then used for further input to interpret shape variation and test various hypotheses in order to ascertain evolutionary parameters, such as biological divergence (F_{ST}), biological distance, migration, and genetic drift (Relethford, 1986; Relethford and Blangero, 1990). Principal component scores (coefficients) for each individual generated from the total covariance matrix were used as input in the program RMET (Relethford, na). In RMET, heritability was set to 0.55 or 1.0 and small sample sizes were corrected for according to the method of Relethford et al. (1997). All groups in the RMET analyses were weighted equally. I also generated biological distance matrices (R-matrix and Mahalanobis) using RMET.

Multidimensional analysis was then performed to attain consensus among the various statistical methods and look for overall patterns of morphological variation among groups. Canonical variate (CVA) and principal component (PCA) scores were generated in *MorphoJ* and then aggregated into group means using SPSS 19.0. Bi-plots were then displayed using the first three dimensions and modified in SPSS 19.0. Principal coordinate plots (PCO) were generated from group mean PCs that accounted for 95 percent of the variation. I used Mahalanobis distances to generate all PCO plots in this study. Minimum spanning trees (MST) were overlayed on the morphospace of the PCO and PCA to infer group relationships in a nearest-neighbor sense. Clustering analysis was performed on either the R-matrix or Mahalanobis distances. I used two clustering

procedures: Ward's and neighbor-joining (NJ). Clustering, PCA, and PCO plots were generated using PAST 2.0. NJ trees were further modified using SplitsTrees (Huson and Bryant, 2006).

I also used a model-based clustering approach to assess for sub-populations within the Xiongnu samples and as an exploratory approach to test for the number of clusters in the global cranial series. The program *MCLUST*, written in R, was used to assess multiple groups within the data. Briefly, *MCLUST* implements a finite mixture model using a Bayesian informative prior in order to choose the best among a number of clustering algorithms. This was done in order to pool Xiongnu samples in a effort to increase sample size. Since one of the goals of this dissertation is to assess Xiongnu population structure, it was important to assess within-group variation without having *a priori* grouping.

Mean group principal component scores were assessed through a multiple regression analysis to determine possible geographic or temporal significance associated with a particular PC score. This was done using the RT program written by Manly (1997). The biological distance matrix generated from the RMET analyses was used in conjunction with spatial (geographic) and temporal distance matrices to perform Mantel tests as a method of testing correspondence between the distance matrices. The Mantel test is assessed to better understand the effects of spatial and temporal distance on population structure among groups in the analysis. Pairwise geographic distance matrices were generated from coordinate points for each sample using geodesic distance. Temporal distances were generated by using mean sample time with modern samples having a value of 0. Then, Euclidean temporal distances were generated between pairs of

samples. All Mantel and partial Mantel tests were performed using the software program PASSaGE 2. Significance in PASSaGE was assessed using a two-tail distribution after 10,000 permutations.

In order to test for various microevolutionary process, such as genetic drift or gene flow, or to test indirect relationships among samples, comparisons of Mongolian samples were made with regional samples using four different analyses. I first tested all of the groups together in a global analysis that included samples that could be included as outliers, such as groups from Europe and Africa. As observed versus expected phenotypic variance differs depending on the samples used in the Relethford-Blangero model, I tested the Mongolian samples to three main regions among which they may have interacted. The first was to compare Mongolian samples to Chinese samples. Historically and archaeologically, there is evidence the Xiongnu interacted to a greater extent with sedentary groups residing in China. The second analysis included only Central Asian samples. Few studies have compared the Xiongnu to groups in Central Asia, therefore in order to characterize Mongolian population history, a separate analysis was conducted to understand potential biological interaction with Central Asian samples included in this dissertation. Lastly, the Xiongnu and Mongolian samples were compared with various temporal and spatial Siberian samples. Several studies have suggested the Xiongnu are biologically similar to several Siberian groups. Therefore, I tested this assumption using only Siberian populations.

***Analytical Procedure 1: Within-Group Population Structure of the Xiongnu*:** In order to detect sub-populations within the Mongolian samples, and specifically to look for population structure within the Xiongnu, the Mongolian samples were first subjected to

the General Procrustes Analysis (GPA). Covariation matrices were then generated in order to perform PCA. Using the PC scores, the *MCLUST* procedure was implemented to detect sub-clusters. This was followed by separating the Egiin Gol Xiongnu crania into their own group and testing within-group variation through Principal components analysis, Ward's hierarchical clustering, Mahalanobis distance, R-matrix analyses, and Discriminant function analysis using a leave-one-out classification table.

***Analytical Procedure 2: Mongolian series tested against Global Cranial Series*:** Mongolian crania were then compared to the global series. Crania were first pooled by region to assess population history. All crania included in the analysis were first subjected to GPA and a covariance matrix was generated. This was followed by performing PCA and CVA in *MorphoJ*. A PCO plot was also generated using RMET. The pooled series were then analyzed using Mahalanobis distances. Using the PC scores as input for new shape variables, a Relethford-Blangero (using two different heritability estimates) and R-matrix analysis were assessed. Groups were then analyzed on their own (not pooled) and subjected to Principal components analysis and clustering analysis. Using results from the R-matrix, pairwise F_{ST} values were calculated between groups. Lastly, *MCLUST* was used to assess for sub-populations using all available crania. To clarify the phenetic relationships among samples in the Discussion chapter, PC plots with MST are included for easy interpretation without needing to refer to the *Results* section.

***Analytical Procedure 3: Mongolian series tested against Chinese Cranial Series*:** Mongolian crania were then compared to the Chinese cranial series. All crania included in the analysis were first subjected to GPA and a covariance matrix was generated. This was followed by performing PCA and CVA in *MorphoJ*. A PCO plot was also generated

using RMET. The series were then analyzed using Mahalanobis distances. Using the PC scores as input for new shape variables, a Relethford-Blangero and R-matrix analysis were assessed using a heritability of 1.0. Groups were then subjected to clustering analysis. Included in the Discussion chapter are PCA plots with MST to illustrate and clarify sample phenetic relationships.

***Analytical Procedure 4: Mongolian series tested against Central Asian Cranial Series*:** Mongolian crania were then compared to the Central Asian cranial series. All crania included in the analysis were first subjected to GPA and a covariance matrix was generated. This was followed by performing PCA and CVA in *MorphoJ*. A PCO plot was also generated using RMET. The series were then analyzed using Mahalanobis distances. Using the PC scores as input for new shape variables, a Relethford-Blangero and R-matrix analysis were assessed using a heritability of 1.0. Groups were then subjected to clustering analysis. Included in the Discussion chapter are PCA plots with MST to illustrate and clarify sample phenetic relationships.

***Analytical Procedure 5: Mongolian series tested against Siberian Cranial Series*:** Mongolian crania were then compared to the Siberian cranial series. All crania included in the analysis were first subjected to GPA and a covariance matrix was generated. This was followed by performing PCA and CVA in *MorphoJ*. A PCO plot was also generated using RMET. The series were then analyzed using Mahalanobis distances. Using the PC scores as input for new shape variables, a Relethford-Blangero and R-matrix analysis were assessed using a heritability of 1.0. Groups were then subjected to clustering analysis. Included in the Discussion chapter are PCA plots with MST to illustrate and clarify sample phenetic relationships.

Analytical Procedure 6: Manly and Mantel Testing: A multiple regression test was assessed to look for temporal and geographic patterns within the data. Using several independent variables (latitude, longitude, time) and mean group PC scores and dependent variables, the RT program written by Manly (1997) was used to look for correlation and significance. This analysis was conducted separately according to sample comparison. For example, mean group PC scores were assessed among all samples in the global analysis. The test was then repeated for the three separate analyses. Once significant PC scores were found to be correlated with any of the independent variables, they were then left out of the Mantel matrix testing. Mantel testing was conducted separately according to the analysis, i.e. first the Mongolian samples were tested against the global series, then the Chinese, then Central Asia, and lastly the Siberian series. All biological distance matrices used in the Mantel testing were generated from the Mahalanobis distances obtained from the separate analyses conducted in RMET.

Craniofacial Variation as an Analytical Methodology

The analysis of ancient populations through the investigation of skeletal features (such as craniofacial diversity) is an effective and informative way to understand modern population structure, infer relationships in the past, and assess potential selection or neutral processes on cranial traits (Relethford and Harpending, 1994; Hanihara, 1996; Brace et al., 2001; Jantz and Owsley, 2001; Brace et al., 2006; Nystrom, 2006; Ross et al., 2002; Hemphill and Mallory, 2004; Ross, 2004; Manica et al., 2007; Hanihara et al., 2008; von Cramon-Taubadel and Lycett, 2008; Relethford, 2010). Craniometrics and the theoretical extension of craniometric data (such as distance analysis, quantitative population genetic models, and multivariate statistical analysis) can answer specific

research questions and hypotheses. Quantitative trait variability in Mongolian groups was evaluated through the use of craniometrics, which are simply measurements designed to quantify craniofacial morphology. The foundation for the use of craniometrics as a tool to quantify phenotypic variance lies in its ability to effectively measure genetic, or biological distance and diversity among prehistoric and modern human populations; or to make inferences to the evolutionary history of particular groups (McKeown and Jantz, 2005).

Anthropometry on living populations has a long history in physical anthropology (Boas, 1912; Stinson et al., 2000), and has been used extensively in osteological research to answer questions of among and within population variability, growth and development, demographic changes, stature estimation and patterns of sexual dimorphism, among other interests (Larsen, 1997; Katzenberg and Saunders, 2000; Buikstra and Beck, 2006). Craniofacial morphology has been used extensively in the literature to assess patterns of human variation (Hrdlicka, 1924; Hooton, 1930; Martin, 1957; Howells, 1973, 1989; Relethford, 1994), and test hypotheses for the emergence of modern human origins (Relethford and Harpending, 1994; Relethford, 1994, 1995; Hanihara, 1996; Lahr and Foley, 1998). Craniometrics has the potential to reveal evolutionary relationships among groups, known as biodistance studies (Buikstra et al., 1990). Biodistance analysis is a well-developed analytical and methodological technique that researchers have employed to better understand microevolutionary processes, such as gene flow and genetic drift within and among geographic populations (Jantz, 1973; Relethford et al., 1997; Powell and Neves, 1999; Steadman, 2001; Stojanowski and Schillaci, 2006; Perez et al., 2007).

One of the major goals in human biological variation is to determine the overall genetic similarity between populations. Two of the most basic questions to determine genetic relatedness are *1)* Which populations are more similar to each other genetically? and *2)* Why? What are the reasons for genetic similarity or dissimilarity? Are groups more closely related because of shared gene flow or a common historical origin? Are populations less genetically similar because of some isolation factor, which could influence population size and thus increase genetic drift? What do other variables, such as geography, demography, or cultural variation have on the relative similarity among populations? One method of answering these questions is to use geometric morphometrics, a suite of applications in the analysis of shape to accurately describe morphological variation. Coupled with quantitative genetic theory, the use of geometric morphometrics was employed in an attempt to answer some of these important anthropological questions.

Geometric Morphometrics as an Analytical Methodology

Most morphological traits can be quantified effectively by single measurements of the size of a part, such as the length of primate limb elements (Young et al., 2010). Other traits are more complex, and cannot be characterized by size alone. For these traits, such as craniofacial traits, information about shape, which concerns the proportions and relative positions of parts, is important (Klingenberg, 2010). Historically, studies assessing craniofacial variation have used "traditional morphometrics", which can be defined as the field of multivariate statistical analysis concerned with the methods necessary to answer questions in biological research concerned with shape (Marcus, 1990; Slice, 2005, 2007). Past studies have relied on the analysis of distances, angles,

chords, or ratios to answer questions of biological variation (Martin, 1957; Howells, 1973; Bass, 1987). Recent theoretical and computational advances have shifted the focus of morphometric procedure from linear measurements to Cartesian coordinates of anatomical points (Bookstein, 1991; Slice, 2007).

This latest and relatively new approach to shape analysis in physical anthropology (although see Benfer, 1975 and Cheverud et al., 1983) is called geometric morphometrics (Kendall, 1981, 1984; Bookstein, 1989, 1991, 1996; Rohlf and Slice, 1990; Rohlf and Marcus, 1993; Rohlf, 2000; Klingenberg and Leamy, 2001; Klingenberg and Monteiro, 2005; Slice, 2005; Klingenberg, 2008a, 2009, 2010). Geometric morphometrics (GM) is the suite of methods for the acquisition, processing, and analysis of shape variables that retain *all* of the geometric information contained within the data (Slice, 2005:5). GM methods also allow for the separation of shape difference from absolute size difference (Yaroch, 1996).

Of central importance for GM is its ability for complete retention of geometric information throughout the research process, which linear measurements fail to capture (Slice, 2007). Also, of primary concern is the emphasis in shape analysis, which does not include size as a factor in defining the variance. GM also has the added advantage of visualization techniques for archival purposes (in the case of repatriation), lower intra- and inter-observer error rates, greater data efficiency, greater speed of collection, and easy conversion to linear measurements (Ousley and McKeown, 2001). The collection of coordinate data is relatively simple with digitizing equipment that downloads data directly into a computerized format, eliminating the need for multiple calipers and manual recording (McKeown and Jantz, 2005).

Often, shape is the central concern for both craniometric and coordinate data analysis in the context of biological variation. The shape of an object can be defined as a property that encompasses all of its geometric properties except its size, orientation, and position (Dryden and Mardia, 1998). Though this definition is somewhat abstract, we intuitively use this when viewing objects in a picture. Consider the Eiffel Tower. We can easily recognize this iconic landmark on a small picture, even if we are far from Paris when looking at it, or we are holding the picture upside down. In the case of linear data, the confounding effect of size must be removed. To circumvent the effects of size differences, traditional size and shape variables can be computed according to the method described by Darroch and Mosimann (1985) using raw measurements. In this case, size is removed and redefined as the *geometric mean* of all variables. The size variable is calculated as follows:

$$Size = \left(\prod_{i=1} X_i \right)^{1/n}$$

Each raw measurement is then divided by the *Size* variable to create new shape variables, which are simple ratios of the geometric mean and are scale-free or dimensionless (Ross, 2004).

The most widely developed methodological approaches used for GM have been the Procrustes methods, which are based on the least-squares estimation of translation, rotation and scaling parameters that optimally align sets of landmark coordinates for pairs of specimens (Slice, 2005, 2007). Because landmark coordinates are recorded with respect to arbitrary digitized axes, GM methods must be mapped into a common

coordinate system so that they can be used in traditional statistical analysis, or indirect applications of biological variation.

It may be helpful to understand how shapes are compared in GM and the properties of their shape spaces. The data for a shape consists of a *k x p* matrix of coordinates, where *p* is the number of landmark points, and *k* is the dimensionality of the physical space in which the objects are digitized (Rohlf, 1999). Therefore, since I collected three dimensional coordinates for 35 landmarks, the figure space for the cranial configurations has (35 x 3 = 105) dimensions. A basic approach to shape comparison is to superimpose them and note any differences in the positions of landmark points. The shapes are first superimposed by centering them on their origin and scaling them by what is known as the "centroid size", a unit computed as the square root of the sum of their squared coordinates (Bookstein, 1991). In geometric morphometric literature, the centroid size is conceptually similar to the geometric mean. I will continue to use centroid size throughout this dissertation. The coordinates of the shape are treated as a single unit length vector. One shape is then rotated to align it with another so that the Procrustes distance (the square root of the sum of squared differences between corresponding points), sometimes referred to as *d*, is as small as possible.

Another fundamental operation in the comparison of shape is to compute an average shape for all specimens in a sample. This is generally done by what is known as generalized least-squares Procrustes superimposition method, or GLS (Rohlf and Slice, 1990). The landmark coordinates (3D) for a set of objects are transformed into points (2D) in the shape space of Kendall (1984) through scaling and alignment procedures known as generalized least-squares Procrustes analysis (GPA), which addresses the issue

of location and orientation with respect to the digitizing axes by estimating the parameters for location and orientation (Gower, 1975; Rohlf and Slice, 1990). This superimposition method goes through a series of analytical procedures. First, all specimens are scaled to the same size in two steps. Initially, centroid size is calculated (the average configuration), and then, the *x*, *y*, and *z* value of every coordinate are divided by centroid size (Rohlf, 1990). This step results in *x*, *y*, and *z* values for which size has mostly been factored out, unless there are some inherent allometric effects in the data. Then, translational and rotational differences are removed in order to remove variation in position, in which all configurations are translated so that their centers of gravity are at the origin of the coordinate system. The centroid for each object them becomes superimposed onto the centroid of the first object, and a series of least squares fitting calculations is undertaken until the distances between shapes are minimized (Rohlf, 1990). The difference between each corresponding landmark is the Procrustes distance (H. Smith, 2009). See **Figure 6.1** for a visual description.

The coordinates of landmarks on each specimen are then usable as shape variables projected into a linear space tangent to Kendall's shape space (1984), which can be used to investigate shape differences; or can be subjected to the usual kinds of multivariate analysis to quantify covariance structure around the mean and group differences (Slice, 2005, 2007). Kendall's shape space is non-Euclidean in nature, being visualized as the surface of sphere (Rohlf, 1996). As dimensionality increases, this space becomes increasingly more complex. As this space is non-Euclidean, traditional statistics cannot be preformed. However, for each configuration that exists in Kendall's shape space (or the Procrustes hemisphere, Slice, 2001), a projection can be made that is tangent to the

shape space. This “tangent” space has properties of Euclidean geometry and intersects with the shape space that also coincides with the Procrustes consensus configuration (Rohlf, 1996, 1999).

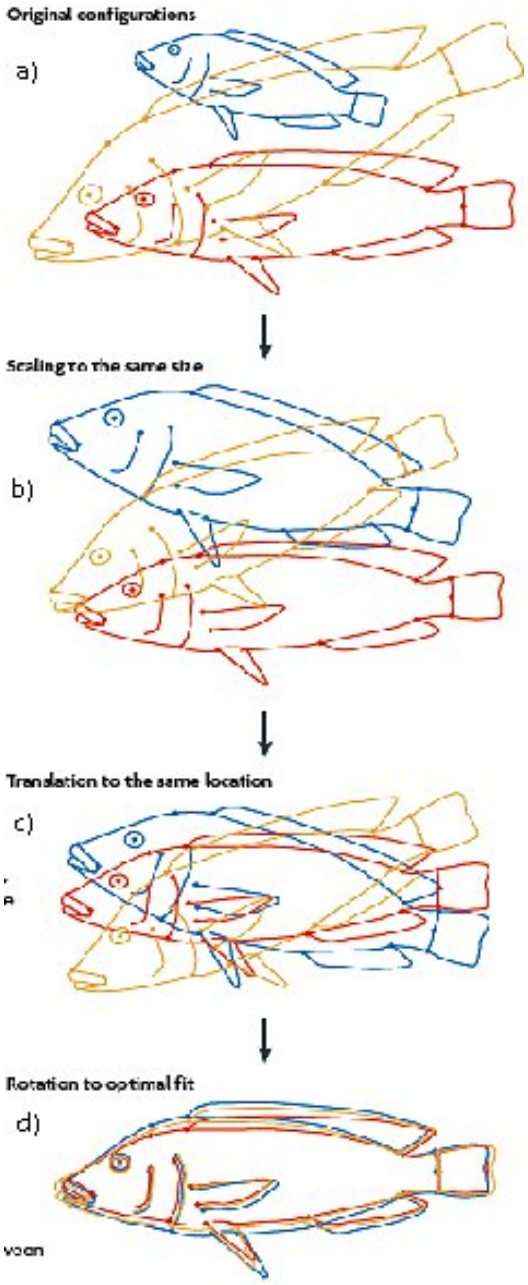

Figure 6.1. Procrustes Superimposition. The process starts with a) the configurations of landmark coordinates as they were measured; b) scaling figures to the same size; c) moving them to a standard position; and d) rotating specimens around a center of gravity to bring all specimens into an optimal orientation in which the sum of all squared deviations between corresponding landmarks is minimal. **Adopted from Klingenberg, *Nature Reviews Genetics*, 2010, pp. 626.**

Multivariate Approaches to the Analysis of Shape

Shape variation is inherently multidimensional. As even simple shapes vary in quite different ways, analyses should utilize a suite of multivariate methods that simultaneously consider the covariation of all landmark coordinates (Klingenberg and Monteiro, 2005). There are a number of different multivariate approaches to shape variation, most of them utilizing the ability to find new variables, corresponding in direction to shape space, which optimizes criteria related to the question of interest (Klingenberg 2010). For example, principal components analysis can be used for examining primary patterns of variation (Drake and Klingenberg, 2010), canonical variate analysis provides the best separation of known groups (Pretorius et al., 2006), multivariate regression, whereby one set of variables is explained by other variables, can be used to assess allometry or evolutionary change in shape over time (McKeown, 2000; Drake and Klingenberg, 2008), or partial least squares analysis, which attempts to find optimal variables for showing patterns of covariation of shapes (Bruner et al., 2010).

The samples used in this dissertation were subjected to a full Procrustes fit (GPA). The projected data becomes orthogonal to the tangent space (Dryden and Mardia, 1998). Specimens were aligned in *MorphoJ* by principal axes. After alignment, an outlier identification procedure was performed in order to exclude potential specimens that may have been measured incorrectly. *MorphoJ* automatically identifies specimens with missing landmarks and excludes them from further analysis. An optional procedure involves manually excluding individuals that strongly deviate from the consensus shape. This is done by visually inspecting a diagram showing the cumulative distribution of distances of individual specimens from the average shape of the entire sample. One curve

shows the expected curve for a multivariate normal distribution fitted to the data, whereas another curve shows the distribution of distances in the dataset. High dimensionality of the data and a large number of individuals uses Mahalanobis squared distance as the measure of how unusual an individual is relative to others in the sample. Most morphometric datasets do not conform to a multivariate normal distribution. Therefore, using a curve that indicates only a few individuals deviating is what was most commonly used in further analyses. After Procrustes superimposition was performed (GPA), a covariance matrix was generated. It is from this covariance matrix that further multidimensional analyses were performed.

Multidimensional Analysis

Principal components analysis: The output of GPA can be used as an exploratory method for additional parametric statistical analyses such as *principal components analysis* (PCA), a technique that reduces a large set of variables into a smaller, more meaningful interpretation of the data through the examination of coefficients. PCA attempts to maximize the within sample variance of a linear combination of variables. This results in axes in which the observations are maximally dispersed (Kachigan, 1991). The overall pattern of variation around each landmark can be summarized by plotting the first and second principal component axes, which are the eigenvectors of the variance-covariance matrix of object landmark locations expressed as deviations from the reference landmark (Rohlf and Slice, 1990). The component scores are typically organized whereby the component accounting for the majority of the variation is first, the second, orthogonal to the first, accounts for the next greatest amount of within-group variance, and so on. This is preferable to using raw data, which tend to be correlated due to similar size and shape

space. PCA, due to orthogonal vectors, are not correlated to one another, thus representing an independent aspect of size or shape. The number of components is influenced by the number of input variables. Each succeeding component accounts for less and less of the variance. The scatter of residuals would then indicate the direction of variability.

For this research, the principal components scores themselves were utilized as new shape variables for further analytical testing (constructing craniometric affinity matrices). Using the approach of Roseman and Weaver (2004) and von-Cramon Taubadel (2009a, 2011), for each individual analyzed, the number of PCs required to explain at least 95% of the overall morphometric variance were employed. The large configuration of coordinate landmarks yields a large number of PCs. Using principal components scores derived from the covariance matrix of the Procrustes residuals accomplish several things. First, visual interpretation can be made about shape variation from the projected axes. This information can be used to assess population relationships. Second, PCA reduces dimensionality in such as way as to make large, complex datasets, manageable. This allows the researcher to interpret shape variation present in the overall sample. Lastly, and most importantly, as employed by Roseman and Weaver (2004) and von-Cramon Taubadel (2011), PC scores can be used as input shape variables for further statistical and model-bound analyses to detect significant within-group variation that may be contributing to the detection of gene flow, admixture, or genetic drift. The approach can also be used to construct morphological matrices to test for spatial or temporal trends in the data.

Principal coordinate analysis and D^2: Principal components analyses applied to shape variables represent a fraction of the total variance that can then be subjected to *principal coordinate analysis*, which is derived from the squared Mahalanobis distance matrix (D^2). Mahalanobis distance is a distance measure introduced by P. C. Mahalanobis (1936). It is based on correlations between variables by which different patterns can be identified and analyzed. It differs from Euclidean distance in that it takes into account the correlations between variables in the data set and is scale-invariant, i.e. not dependent on the scale of measurements. Mahalanobis distances are good indicators of group similarity or dissimilarity.

Principal coordinate analysis (PCO) derives linear combinations of variables that best reflect the variation between groups under investigation (Gower, 1966). Principal coordinate analysis is exactly similar to metric multidimensional scaling, where dimensionality is reduced to one, two, or three dimensional space that depicts the greatest amount of variation within the data. The amount of variance is also given for each eigenvector. PCO is sometimes preferred over PCA because the user can choose the particular distance measure of interest, Mahalanobis distance, for example. All PCO analyses were performed using PAST v. 2.12 (Hammer et al., 2001).

Geometric Morphometrics in Anthropological Studies: A Review

Geometric morphometric methods have been developed for use in physical anthropology, and its utility has been demonstrated in answering some fundamental biological shape questions for both humans and primates alike. For example, Fleagle et al. (2010) studied primate cranial morphology to better understand primate evolution and relatedness among extant primate taxa. Other studies include regional variability of

modern human craniofacial form (Hennessy and Stringer, 2002; Bruner and Manzi, 2007). In that study the authors found GM methods to be in common agreement with classical studies of regional craniofacial diversity (Howells, 1973, 1989). Harvati et al. (2010) studied the phylogenetic relationships during the European Pleistocene fossil record using GM and find that a clear pattern emerges for craniofacial variability between Neanderthal and archaic hominin groups. Harvati (2009) also explored Eurasian hominid evolution by re-examining the Upper Cave (Zhoukoudian) fossil material from China. She found that these specimens, which have been controversial and have not classified into any recent modern human population, are closely related to Upper Paleolithic European samples, and have most likely retained archaic features as explained in the Single Origin hypothesis of modern human origins.

Sexual dimorphism has also been studied in a number of contexts (Rosas and Bastir, 2002; Oettle et al., 2005; Pretorius et al., 2006). For example, Pretorius et al. (2006) found the use of coordinate data as a better indicator of sexual dimorphism for classical dimorphic features such as the greater sciatic notch and mandibular ramus. In addition, the orbit exhibited greater than normal sexual dimorphism under the scrutiny of thin-plate splines and canonical variate analysis. That is, using GM, the authors were able to identify dimorphic orbit variation that was previously not found using only macroscopic techniques.

Studies of population history and structure have also been undertaken using geometric morphometrics. Phenotypic evolution of human craniofacial morphology in South African crania (Franklin et al., 2007) and South American populations after genetic admixture (Martinez-Abadias et al., 2006; Perez et al., 2007) have also been studied at

the local level. McKeown and Jantz (2005) conducted a comparison study for coordinate and craniometric data in biological distance studies among prehistoric Native American groups and found geometric morphometrics to elucidate a higher resolution of population variation by indicating vault asymmetry in some of the groups tested. Bernal (2007) found that using morphometrics in the size and shape analysis of human molars on three archaeological samples from Argentina, when compared with traditional measurements, greatly enhances information about molar contour and captures morphological features with low levels of variation not previously reported. More recently, Gonzalez-Jose et al. (2008) and de Azevedo et al. (2011) attempted to model the settlement of the New World using GM methods. These authors found that rather than a "single wave" or "two waves/components" model to explain the modern and archaic craniometric diversity of the New World, a "recurrent gene flow" model has more explanatory power, whereby Native American groups emerged from a single migration into the New World, followed by local, within-continent evolution with continued and persistent contact among Circum-Arctic groups.

Recently, geometric morphometrics have become applicable in the study of evolutionary-developmental biology of organisms, especially in the area of morphological integration and modularity. Organisms are integrated to function as a whole, but integration in organisms is not uniform throughout. Traits of organisms do not vary independently, but are integrated with each other to reflect coordination in development, function, and evolution. Integration is rarely homogenous and there are complexes of more tightly integrated traits called modules that are relatively independent of one another (Klingenberg, 2010). This tension between coordination and independence

is captured in the concepts of integration and modularity. Modularity is a general property of many types of networks. Biological modularity has concerned itself with a wide range of levels of organization, including molecular interactions in gene expression, metabolic networks, and networks of ecological interaction. In order for the concept of modularity to be useful to biological problems, a specific context that defines the nature of the interactions or imposes limitations on the parts of the module are necessary. It is helpful to think of the concept of a module as requiring an adjective, such as developmental, genetic, or evolutionary that describes the particular context. Integration is the cohesion among traits that results from the biological processes producing a variety of phenotypic structures under study, such as craniofacial traits (Klingenberg, 2008). Essentially, modularity is about differences in the degree of integration of parts within and between sets of traits.

Morphological integration tends to derive from data on the covariation of multiple traits as morphological data lacks information on network interactions among measured traits. Emphasis is placed on the extent to which different traits are linked to one another, and on the patterns of covariation, which are focused on the specific changes of traits that occur together. The human skull has been hypothesized to be strongly integrated throughout, however main parts of the skull (modules) are thought to be relatively independent of one another. Although there is a general consensus that cranial morphology can provide a strong signal of phylogenetic efficacy (tracking hominin evolutionary relationships) and to reconstruct human population history, there are differences regarding the relative neutrality of different cranial regions and how these regions are influenced by processes of integration among traits or plastic response to the

environment (Harvati and Weaver, 2006; Smith, 2009; von Cramon-Taubadel, 2009a,b). In humans, facial form, particularly the nasal region, and the size and shape of the neurocranium have been thought to be related to climatic adaptations while the shape of the basicranium, particular the basal aspect of the temporal bone, has been shown to be the most genetically determined and evolutionarily conservative aspect of the cranium with minimal environmental influence.

During the course of hominid evolution, morphological alterations of the skull have occurred due to the transition to bipedal posture, while the modern human skull has seen the development of a globular and expanded cranial vault, retraction of the face, and strong cranial base flexion (Aiello and Dean, 1990; Lieberman, 2011a). These changes have been debated as the consequence of adaptation to transitions in locomotion, diet, language, and cognitive abilities. Conversely, others see a few basic developmental changes related to the size and shape of the brain and face to have triggered a whole suite of integrated cranial features common among modern humans (Bastir et al., 2010). This debate as to the significance of the evolution of the human skull can be understood and is related to the question of whether variation in the skull is morphologically integrated (single change that jointly affected a suite of integrated cranial features), or modular (localized adaptive changes).

If the human skull is strongly integrated throughout, and the main parts of the skull (modules) are not as weakly integrated, then there is the possibility that selection has not acted strongly on cranial diversity, and the skull can be used to infer selectively neutral process, such as demographic histories of populations. Although there is a general consensus that the human skull can provide a strong signal of phylogenetic efficacy of the

entire cranium, there are differences regarding the relative neutrality of different regions (Roseman, 2004; Harvati and Weaver, 2006; von Cramon-Taubadel, 2009a,b; H. Smith, 2009; Hollo et al., 2010). There is compelling evidence that selective forces have not acted to shape the human skull (Lieberman, 2011a), though some authors suggest some regions more than others are more susceptible (Betti et al., 2009). That is, are particular regions of the skull better indicators of past population history?

The use of geometric morphometrics has the ability to answer this question. Lockwood et al. (2004) have shown that the shape of the temporal bone can distinguish among species of extant great apes. These authors demonstrated, using temporal bone shape coordinate data from modern humans, chimpanzees, orangutans, gorillas, and bonobos, that the resultant phylogenetic tree of these taxa was identical to the molecular phylogeny of these species, As such, several studies (Harvati and Weaver, 2006; Smith et al., 2007; H. Smith, 2009) have suggested that human temporal bone shape is highly correlated with neutral molecular distances (loci not influenced by selective processes), while temporal bone size is a reflection of environmental differences related to climate and latitude. Harvati and Weaver (2006) suggested that the temporal bone's phylogenetic signal was tracking much older evolutionary events in human history (i.e., its ability to separate sub-Saharan African from non sub-Saharan African groups), while cranial vault (neurocranium, facial shape) changes were related to more recent events, such as adaptation to extreme climates.

von Cramon-Taubadel (2009a) studied the rationale for temporal bone shape in reflecting greater phylogenetic signal based on either the functional complexity of the bone itself or the overall morphological contribution of the temporal bone to the

basicranium. She found the temporal bone's unique status for tracking evolutionary events may be due to it being the only bone to be individually compared against other cranial regions, or equivalent cranial units, such as the frontal, parietal, or occipital bones. Although the temporal bone is a reliable indicator of past population history, she found it cannot be distinguished statistically from the frontal, parietal, or sphenoid bones in terms of its congruence with neutral molecular data.

von Cramon-Taubadel (2011) then tested the efficacy of functional and developmental cranial modules for reconstructing human population history. She tests two hypotheses for logically using developmental or functional criteria in delineating suitable cranial units related to congruence with neutral molecular data. The first hypothesis predicts that the basicranial region of endochondral ossification is more reliable to reconstruct population history than the intramembranously ossifying regions of the human cranium. This hypothesis is based on the assumption that the early ossification of the basicranium and its distinct functional constraints are relatively immune to non-neutral evolutionary forces. The second hypothesis tests the theory that cranial regions associated with a single sensory function are less reliable indicators of neutral genetic history. This is based on the idea that multifunctional cranial regions are less likely to exhibit homoplasy, and therefore, provide a more accurate morphological proxy for genetic relationships. She finds little support for the "basicranium hypothesis" as intramembranously ossifying regions (modules) of the cranium showed just as much genetic congruence. She also finds less support for defining cranial modules on the basis of anatomical or functional complexity as this did not provide a consistent means in predicting phylogenetic relationships or population history. Overall, she suggests

researchers should be focused on areas that are particularly unreliable (such as the zygomatic and occipital bones) and removing these from the analysis, rather than identifying informative regions for congruence with neutral molecular data.

Martinez-Abadias et al. (2012) studied the particular question of morphological integration and modularity of the human skull by applying geometric morphometrics and quantitative genetic theory to the study of the Hallstatt, Austria ossuary. Their results are encouraging as they find that the face, cranial base, and cranial vault should not be seen as independent modules, but are strongly integrated structures. The methodology applied by Martinez-Abadias et al. (2011) has a significant advantage over previous research (Ackermann and Cheverud, 2004; Roseman, 2004; Weaver et al., 2007; von Cramon-Taubadel, 2009; Betti et al., 2010) that used phenotypic covariance structure as a proxy for genetic data. Instead, they estimated a genetic covariance matrix directly from the traits as the Hallstatt population provides a large number of crania with associated genealogical information (Sjovold, 1984). They find strong integration for cranial shape throughout the skull as genetic variation is concentrated in only a few dimensions. When a suite of hypothetical selection scenarios were applied, the authors found global responses to localized selection, thus indicating a strong genetic component and integration to overall cranial shape change. As a result of the concentration of overall cranial shape variation, a change in response to selection will strongly depend on the direction of selection, and likewise, an evolutionary response to drift will tend to be in directions with large amounts of genetic variation (Lande, 1979). This means that, overall, the skull behaves as a composite, and changes in one region will produce correlated phenotypic changes in other regions, similar to studies in mouse and newt

skulls (Halgrimmson et al., 2009; Ivanovic and Kalezic, 2010), and previous studies of the human skull (Bookstein et al., 2003; Mitteroecker and Bookstein, 2008; Bastir et al., 2010).

Relethford (2004b) analyzed traditional immigrant data originally collected by Boas (1912) to understand the interaction between environmental plasticity (natural selection) and craniometric variation. His results indicate that craniometric variation is affected by both natural selection and genetic influences; however, the relative *patterns* of craniometric variation are not obscured or erased by these environmental influences. Importantly, this observation would suggest that craniometric data can be used to study developmental plasticity, long-term environmental adaptation, and models of population structure and history that reflect an underlying neutral model (Powell and Neves, 1999; Relethford, 2001, 2004a, 2004b).

A neutral, or nearly neutral model for craniofacial morphology assumes that the traits under analysis are selectively neutral and are not significantly being affected by environmental conditions and plasticity. That is, the patterns of biological relationships inferred from craniometric trait variability will not be significantly obscured by selective forces (Relethford, 2004a, 2004b).The craniofacial dataset used in this study has been recently tested for the effects of climate and other variables that may affect interpretation of population history, with results similar to other recent studies (Relethford, 2004a, Roseman, 2004; Harvati and Weaver, 2006; Weaver et al., 2008; von Cramon-Taubadel, 2009; Betti et al., 2009, 2010; Relethford, 2010).

Betti et al. (2010), using a large and varied craniometric dataset (Hanihara and Ishida, 2001) tested the effects of climate on the size and shape of the overall cranium for

between-population diversity and found that only a moderate link exists between climate and population structure. The expected differences can be accounted for through an isolation-by-distance (IBD) model, whereby geography plays a much stronger role in determining phenotypic differentiation. Additionally, they observed that once IBD is accounted for, climate plays an even weaker role in shaping human population history. Betti et al. (2009), using the same craniometric dataset, tested for within-population diversity and found that climate plays no role in shaping within-group phenotypic diversity. They attribute a relative role for climate in between-group diversity as the nature of selective forces. Unless the effect of directional selection is particularly strong, a trait's mean value could shift without necessarily affecting within-population diversity, and only be reflected in between-group diversity.

Other researchers (Roseman, 2004) have suggested populations residing in extremely cold environments have been more affected by climate. Betti et al. (2010) confirmed this observation. However, when the sample populations living in cold climates (i.e. Inuit) were excluded from the analysis, the correlation for minimum temperature was erased and maximum temperature significantly reduced. Others (Harvati and Weaver, 2006) have suggested climatic adaptation for certain areas of the skull, particularly the facial region, however, they concede the correlations may be confined to only arctic populations, and conclude that craniofacial shape and size retains a population history signal. Betti et al. (2010) also tested their large dataset to single trait correlations and found that weak, but significant, correlations exist for measurements of facial breadth and the dimensions of the orbits and nasal aperture. These authors stress, however, that after correcting for IBD and removing extreme climate samples, the correlations are

either eliminated or reduced. Relethford (2010), using geography as a proxy for neutral variation, found that certain groups deviate from the expected pattern of neutrality as a result of selection to past environments. He concludes that though selection may by affecting craniometric variation, it does not exclusively determine variation.

These studies still leave open the question of what particular regions of the skull might be more susceptible to adaptive processes. Harvati and Weaver (2006) have shown that several populations are distinct due to the extreme climate in which they live (Inuit, for example). One area of the skull that has been researched extensively is the nasal cavity. As humans inhabit a wide range of environments associated with extreme respiratory function, it has long been hypothesized that the nasal cavity plays an important role in climatic adaptation.

Noback et al. (2011) studied the relationship between modern human variation in the morphology of the nasal cavity and climatic factors such as temperature and vapor pressure, and test the hypothesis that within extreme environments (cold, dry, hot, humid), nasal cavities will exhibit features that enhance turbulence and air-wall contact to improve conditioning of the air. Noback et al. (2011) sampled 10 modern human populations residing in extreme climates and use GM to analyze the shape of the bony nasal cavity using 21 nasal cavity landmarks. The authors of this study found a high degree of correlation between nasal cavity morphology and climatic variables. They concluded that nasal cavity morphology appears mostly related to temperature, whereas morphology of the nasopharynx is associated with humidity. Similar to previous studies, they found that the shape of the nasal aperture is higher and narrower in cold climates compared to hot-humid climates. These shape changes in cold-dry climates appear to be

functionally consistent with an increase in contact with air and mucosal tissue through greater turbulence during respiration and a higher surface-to-volume ratio in the upper nasal cavity. However, the authors found significant overlap between populations and only modest shape differences, suggesting a possible functional compromise morphology of the nasal cavity and/or absence of extreme adaptations that reduce the versatility of humans as generalists.

Quantitative Genetic Approaches

R-matrix Approach: Among the direct applications in quantitative genetics to calculate genetic similarity is the use of the R-matrix, originally developed by Harpending and colleagues for the use of allele/ haplotype data (Harpending and Jenkins, 1973). For a given allele, the genetic similarity between population *i* and population *j* is defined as:

$$r_{ij} = \frac{(p_i - \overline{p})(p_j - \overline{p})}{\overline{p}(1 - \overline{p})}$$

where p_i and p_j are the frequencies of the allele in populations *i* and *j* respectively, and $\overline{p}$ is the mean allele frequency over all populations in the analysis, ideally a weighted mean where weighting is by population size (Harpending and Jenkins, 1973; Relethford, 2007:195). The R-matrix provides for an estimate of genetic similarity within and among populations relative to the contemporary means of allele frequencies in a region. The weighted mean of all the R-matrix elements is 0, while the weighted mean of the diagonal elements is a reduced variance estimate, known as r_0, of the overall level of genetic differentiation (Relethford, 1991). A positive r_{ij} value (residual) indicates a pair of populations more similar to each other than average, and a negative r_{ij} indicates a pair of populations that are less similar to each other than average. The method was extended to

include quantitative traits as developed by Williams-Blangero and Blangero (1989) and Relethford and Blangero (1990). When computed for phenotypic data, the diagonal of the matrix contains the minimum estimates of the 'true' R-matrix derived under the assumption that heritabilities are equal to 1 (Williams-Blangero and Blangero, 1989).

Assessing the degree of differentiation is often accomplished through genetic (or biological) distance analysis. Genetic distance studies are widespread and usually employ either estimates derived from the R-matrix or another measure of similarity, such as Mahalanobis (D^2) distance. The R-matrix is transformed into an unbiased R-matrix by adjusting the diagonal elements (r_{ii}) for sample size effects using the method of Workman et al. (1973). As shown by Harpending and Jenkins (1973), the transformed genetic distance between populations *i* and *j* can be computed from the scaled, unbiased R-matrix as

$$d^2_{rj} = r_{ii} + r_{jj} - 2r_{ij}$$

These distances are roughly proportional to the Mahalanobis distances used in studies of quantitative variation. The biological distances are then displayed graphically by plotting the first two eigenvectors obtained from the scaled, unbiased distance matrix (Relethford and Blangero, 1990).

This dissertation will also make use of F_{ST}, a measure of differentiation among populations that for neutral traits reflects a balance between gene flow, genetic drift and mutation (Wright, 1951). Estimation of F_{ST} is given as the average weighted diagonal of the R-matrix,

$$F_{ST} = \sum_{i=1}^{g} w_i r_{ii}$$

where w_i is the relative population size of population *i*, and *g* is the number of populations, and r_{ii} is the genetic distance of subpopulation *i* to the centroid (Relethford and Blangero 1990). Assuming heritabilities are equal to 1 and the phenotypic and genotypic covariances are equal, F_{ST} represents the *minimum* genetic differentiation among regional populations (Williams-Blangero and Blangero, 1989; Relethford, 1994). The higher value of F_{ST}, the greater the variation around the *contemporary* allele frequencies, indicating greater differentiation. Note that this F_{ST} value (as in studies of genetic variation) is not a hypothetical array of ancestral allele frequencies, which are never known (Relethford, 1994).

Relethford and Blangero (1990) tested several alternate estimates of heritability and found that the underlying pattern of differentiation as measured by F_{ST} does not change. As mentioned above, this proposal will use an average value of 0.55 for craniometric heritability and a value of 1.0 to assess minimum genetic distance among groups. F_{ST} values among Eurasian nomadic steppe peoples using craniofacial variation should reflect values similar to previous research (Relethford, 1994). F_{ST} values should be relatively low for an analysis of samples from within Mongolia, but higher for an analysis of samples from the wider geographical area**.**

The use of the R-matrix necessitates the estimation of sample sizes. Estimates of effective census size have not been widely studied in prehistoric Inner Asia. Steadman (2001) discusses the problem of differential population sizes in her study of Woodland and Mississippian groups in North America. This dissertation will make similar use of a scaled, unbiased R-matrix to account for genetic drift in small populations with the elements

$$g(\sqrt{w_i})(\sqrt{w_j})(r_{ij})$$

where *g* is the number of populations and *w* is the relative weight of populations *i* and *j*. Powell and Neves (1999), using a similar model on Holocene hunter-gather groups, estimate effective size from modern studies conducted by Steele et al. (1998). They use an upper range carrying capacity of 0.023 persons/km^2 and estimate small effective population sizes in North America of 36 to 100 persons per subpopulation. Using a relative weight for all groups (1.0) and a scaled weight for Paleoindian groups (0.30), they found their results to be improved.

Although site and cemetery sizes are known for some Xiongnu settlements (Wright et al., 2009), an accurate estimate of effective population size is not known for the prehistoric Xiongnu or other nomadic groups at the time. Archaeological research conducted by A. Weber et al. (2002), van Geel et al. (2004), and Dirksen et al. (2007) in south-central Siberia suggested an increase in population density around 850 BC using calibrated radiocarbon dates. These authors point to an abrupt climatic shift with increased humidity leading to a higher biomass production and carrying capacity for nomadic groups and accompanying herbivores. This evidence would suggest that nomadic populations could be weighted proportionately, with increasing weight for nomadic groups during this period. Though any estimate of census for prehistoric nomadic groups is arguable, a proportional weighting of group size differences has been used to good effect in previous studies (Relethford and Harpending, 1994; Steadman, 2001). This dissertation experiments with a similar scaled weight in the R-matrix analysis in the *Results* section.

Relethford-Blangero Model: Genetic distance studies can help understand the *pattern* of differentiation; however, the interpretation of genetic distance is often confounding. An alternative approach is to use a model-bound method developed by Harpending and Ward (1982) for genetic markers and its extension to quantitative traits by Relethford and Blangero (1990). This method compares two different measures of variation within populations: the observed and expected levels of heterozygosity. Variation in a population can be assessed by computing the average level of heterozygosity within each population from allele or haplotype frequencies. The average per locus heterozygosity in population *i* is

$$H_i = 1 - \frac{\sum p^2 k}{l}$$

where p_k is the frequency of allele *k* in population *i*, *l* is the number of loci, and the summation is over all loci and alleles. This quantity is the *observed* heterozygosity. In general, the level of heterozygosity increases with mutation and gene flow, and decreases with genetic drift (Relethford 2007:198). Harpending and Ward (1982) have shown that the *expected* level of heterozygosity in a population could be derived from the total population heterozygosity (allele frequencies from all populations pooled together), H_T, and the genetic distance, r_{ii}, of population *i* to the set of mean allele frequencies. This genetic distance is the diagonal element of the R-matrix. Given these values, Harpending and Ward (1982) showed that the expected level of heterozygosity in population *i* is

$$E[H_i] = H_T(1 - r_{ii})$$

Relethford and Blangero (1990) have shown that there is a proportional relationship between expected heterozygosity and phenotypic variation, and as such extended the Harpending and Ward (1982) model to quantitative traits as

$$E[V_i] = \frac{V_w(1 - r_{ii})}{1 - F_{ST}}$$

where V_i is the average phenotypic variance over all traits in population *i* (after conversion to standardized scores), V_w is the average phenotypic variance averaged over all groups, and r_{ii} and F_{ST} are estimated from quantitative traits (Relethford, 2007:198).

A comparison is then made between the *observed* and *expected* values of heterozygosity, which can indicate something about the level of *external* gene flow into populations. An assumption is that the observed and expected levels of heterozygosity will be the same across all populations in the analysis. If the observed is *greater than* the expected, then greater than average external gene flow is likely the cause of the excess heterozygosity. If the observed is *less than* the expected, then that population would appear to be more isolated and has received less gene flow. This measure should be highly informative for inferring the biological diversity of groups that have maintained extensive contact through time but have had diverging histories, possibly implementing a level of isolation, and hence genetic drift, among some of the groups under analysis.

The R-matrix method, the calculation of distances for each population, and levels of heterozygosity (as calculated using the Relethford-Blangero model), were generated using the program RMET 5.0 (Relethford, na). Absolute distances were used to compare populations under analysis and were used for further multivariate testing.

Cluster Analysis

Neighbor-joining: The computed distances from the R-matrix will be displayed in a Neighbor-Joining (NJ) procedure to visualize population structure among samples (Saitou and Nei, 1987). The NJ method expresses the structure of groupings visually in a phylogenetic, unrooted tree, or dendrogram, and also evaluates how often a particular

connection between groups has occurred among trees by repeated samples generated from bootstrapping. The NJ procedure is appropriate and can be used even for populations that have not always evolved in a hierarchical manner, such as humans who often conform to a model of isolation by distance (Kalinowski, 2009). Kalinowski (2009) has also shown the NJ procedure to have an accurate fit to the original genetic distance matrix, although he cautioned that the trees often impose a hierarchical relationship among populations and gene flow among populations may have a genetic structure that cannot be represented with a tree.

Ward's Method: Ward's (1963) amalgamation rule uses Euclidian distance to construct a hierarchical tree. This approach is somewhat different to other linkage methods, in that it uses an analysis of variance (ANOVA) to evaluate the distances between clusters. In short, this method attempts to minimize the sum of squares for any two hypothetical clusters that could possibly be formed at each step in the analysis. Both NJ trees and Ward's clustering trees were first constructed in PAST version 2.13 (Hammer et al., 2001) and then modified using SplitsTree4 (Huson and Bryant, 2006).

MCLUST: To further test the number of possible clusters within the data, I used a model-based procedure implemented in the statistical program R using the package *MCLUST* (Fraley and Raftery 1999, 2002, 2006). This method does not assign *a priori* group names to individual crania. Model-based clustering is based on the idea that the observed data come from several subpopulations. The subpopulations are modeled separately and the overall population is viewed as a mixture of these subpopulations, using finite mixture models. The general form of the finite mixture model with *G* groups is

$$f(x) = \sum_{g=1}^{G} \pi_g f_g(x),$$

where π_g is the proportion of the population in the *g*th group and $f_g(\bullet)$ is the probability density function for the *g*th group. Often the subpopulations are modeled by members of the same parametric density family, in which case the finite mixture model can be written as

$$f(x) = \sum_{g=1}^{G} \pi_g f(x \mid \phi_g),$$

where ϕ_g is the parameter vector for the *g*th group.

The mixture model is then used to partition the data using some criteria, such as Bayes factors (Kass and Raftery, 1995). The Bayes rules then classify some observation **x** into some *g* cluster if the posterior probability that it belongs to group *g* is greater than the posterior probabilities that it belongs to any other group (Fraley and Raftery, 2002). Bayes factors are used to compare various models based on the data. Models are compared in *MCLUST* using the Bayes Information Criterion (BIC). This is defined by

BIC = 2 x log(maximized likelihood) – (no. of parameters) x log(n) ,

where n is the number of observations. The model with the highest BIC score is then selected (includes parameterizations of the covariance matrix and number of components, or clusters within the data), and helps decide which among two or more partitions most closely matches the data for a given model (Fraley and Raftery, 1998). The partitions are determined through the Expectation-Maximization (EM) algorithm (Dempster et al., 1977) for maximum likelihood. The EM can also provide a measure of uncertainty about the resulting classification.

MCLUST uses a model-based framework for clustering by parametrizing the covariance matrix in terms of its eigenvalue decomposition in the form of

$$\sum_k = \lambda_k D_k A_k D_k^T \ ,$$

where D_k is the orthogonal matrix of eigenvectors, A_k is a diagonal matrix whose elements are proportional to the eigenvalues of $\sum_k$ and λ_k is a scalar. The orientation of the principal components of $\sum_k$ is determined by D_k, while A_k determines the shape of the density contours (Banfield and Raftery, 1993). Geometric characteristics of distributions, such as orientation, volume, and shape, are estimated from the data. This approach takes into account parameters normally obtained from Gaussian mixture models and chooses the best classification method through the BIC.

Regression Analysis

Morphological variation between groups may be the result of variation in time and/or geography. In order to account for this potential variation, a multiple regression test was assessed to identify possible correlation among the dependent variables (principal components) and the independent variables (latitude, longitude, median time period). Group mean PC scores were regressed onto the independent variables using the multiple regression program RT written by Manly (1997). Correlations generated by this multiple regression analysis are tested for significance by randomizing the independent variable and then computing a *t*-statistic that determines the percentage of the randomized coefficients that exceed the observed coefficient. Depending on the level of analysis (Mongol vs. China, Central Asia, Siberia), each PC score was regressed onto each of the independent variables – time, latitude, longitude. The independent variables were randomized for 1000 runs and significance was assessed via a *t*-statistic at the 0.05 level. Those PC scores significantly correlated with time and/or geography are interpreted in the *Discussion* chapter.

Mantel Tests: Distance Matrix Correlation

In addition to biological distance matrices (both D^2 distance and R-matrix distance) generated from the R-matrix analysis, geographic and temporal distances were constructed to assess information about the correlation correspondence in overall variation for different sources of variability. Geographic distance matrices were generated using PASSaGE 2, an integrated software package for performing spatial analysis and statistics on biological data (Rosenberg and Anderson, 2011). Spatial analysis and patterns is of interest for two reasons. First, spatial patterns can be inherently interesting, as it is often indicative of other underlying patterns. Second, and more important for my purposes, is that the presence of spatial patterns in the data violates assumptions about independent observations that underlie many statistical tests. In this case, the spatial pattern is a nuisance parameter that needs to be dealt with in order to account for other patterns in the data.

Temporal distance matrices were using median time periods for the samples under analysis. For example, the Chandman sample (Mongol Bronze Age) was from approximately 700-400 BCE. In setting up the temporal distance matrix, a value of 550 BCE was used in comparison to other periods. The *n x n* matrix was constructed to be the same size as the biological and geographic distance matrices.

Geographic distances were calculated in PASSaGE using latitude and longitude coordinates for each sample. As mentioned in the previous chapter, some samples are well known with high resolution (such as in China), whereas in others (samples from museums), the site location is unknown, and thus a relative site location was chosen based on the identification of the sample. For instance, the Mongolian sample obtained

from the American Museum of Natural History in New York did not have site information about where the skeletal material came from. In this case, the latitude and longitude of Ulaanbaatar City was used as a proxy. Geographic distance matrices were calculated as two-dimensional spherical distances from coordinate data. This distance is used as it is more accurate for points spaced around the globe (such as skeletal samples), and is calculated as the great circle distance along the surface of the Earth. If x_i and y_i are the longitude and latitude of point *i,* the spherical distance between points *i* and *j* is calculated as:

$$\alpha = \sin(y_i)\sin(y_j) + \cos(y_i)\cos(y_j)\cos\left|x_i - x_j\right|$$

$$d_{ij} = R_E \cos^{-1}\alpha$$

where R_E is the radius of the Earth (R_E = 6379.336847km). Spherical distances are measured in kilometers.

All distance matrices were then tested for correlation using the Mantel test (Mantel, 1967). The Mantel test is extremely versatile, has been generalized by Manly (1995) and Smouse et al. (1986), and has many uses, making it appealing to test the correspondence between two or more distance matrices. The Mantel test compares two square matrices, usually distance matrices, **X** and **Y**. The values within each matrix (*Xij* or *Yij*) represent a relationship between points *i* and *j*. The basic Mantel statistic is simply the sum of the products of the corresponding elements of the matrices

$$Z = \sum_{ij} X_{ij} Y_{ij},$$

where $\sum_{ij}$ is the double sum over all i and all j where $i \neq j$. The Mantel coefficient is usually normalized, calculated as the correlation between pairwise elements of **X** and **Y**, ranging in value from -1 to 1.

Due to the non-independent observations within the matrices, significance is tested through a randomization procedure by permuting the order of the elements in one matrix. More specifically, significance is tested through the number of comparisons involving a randomly rearranged matrix to produce a correlation value as large as or larger than the observed correlation (Smouse and Long, 1992). When the number of points is large ($n > 40$), it is possible to transform the Mantel statistic into a *t*-test statistic, where the significance of *t* is obtained from an asymptotic approximation of the *t*-test (Dutilleul, et al. 2000).

Partial Mantel tests were also calculated. This is an extension of the Mantel test and allows a third (or more) matrix to be held constant while the relationship between the other two is determined (Smouse et al., 1986). For use in biological variation, this test is important as we want to know how the biological matrix is related to the temporal and geographic matrices while holding one or the other constant. This is done using a multiple regression relationship whereby the elements of **X** and **Y** are regressed onto an additional matrix, and using the residuals from the regression as input for the standard Mantel test (Dow and Cheverud, 1985). Estimating significance of the partial Mantel test is accomplished by permuting one of the original matrices through regression prior to the multiple regression, whereby the regression for that matrix is repeated and the partial Mantel correlation determined (Legendre, 2000). Mantel tests are performed using PASSaGE 2.

Konigsberg (1990) explicates and formally develops the theoretical underpinning of space-time variation for chronologically defined archaeological skeletal samples. The classical approach (described in chapter 2) to model spatial correlation between populations is the isolation-by-distance model (IBD). However, this model does not consider the temporal patterns of biological variation. Konigsberg (1990) uses several population genetic models that incorporate spatial and temporal structure (infinite island, unidimensional stepping stone, and migration matrix) that allow the prediction of biological distances between groups separated by a given spatial and temporal lag, such as the skeletal lineages used in this dissertation (Bronze Age, Iron Age, Modern period).

Using the results from the migration matrix and the stepping stone models as a basis for analyzing regional variation across time and space in prehistoric samples, the isolation by distance model predicts that if groups conform to such a model (IBD), then genetic and spatial distance should be positively correlated (when controlling for temporal distance), while genetic and temporal distance will be negatively correlated (when controlling for spatial distance). Konigsberg's (1990) theoretical assumptions will be used for interpreting the results from the Mantel matrix analyses used in this dissertation.

CHAPTER 7

RESULTS

The goal of this dissertation is to ascertain the possible origin of the Xiongnu nomads, who inhabited large swaths of Inner Asia during the Iron Age, and to examine their potential biological relationships with groups in the region. 3D geometric morphometric craniofacial variability was examined and analyzed in this effort. Both indirect (principal components analysis) and direct (Relethford-Blangero model) methods were used to assess Xiongnu population history and structure. As the origins of the Xiongnu are unknown, it was essential to first explore intra-group heterogeneity in order to understand within-group population structure. Xiongnu and various other temporal Mongolian groups were then compared with regional populations to examine broader population history. A hierarchical approach was taken, in that Mongolian (and Xiongnu) were first compared to a large sample of populations that spanned the globe. This was done in order to place the Mongolian groups into a larger regional context for comparison with 'local' groups. Local comparison was then made separately with groups from China, Central Asia, and Siberia, as these groups were within the geographic expanse of the Xiongnu nomadic steppe empire. This chapter will outline the results from these analyses.

Within-Group Xiongnu Population Structure

The term Xiongnu was constructed on the basis of historical Chinese narrative and material culture re-constructed by archaeologists over the years. To better understand possible within-group population structure, a series of analyses were constructed in an

effort to partition possible multiple biological groups within the moniker known as 'Xiongnu'.

The results for the model-based clustering approach for the Mongolian samples are shown in **Figures 7.1 - 7.3**. Using individual PC scores as input (which restricts the covariance matrix making the results obtained here somewhat untenable), *MCLUST* obtained a classification model with the following parameters (*EEI*): diagonal distribution with equal volume and shape (covariance), coordinate axes orientation with one component, or cluster (**Fig. 7.1**). The other models (key in **Fig. 7.1**) have lower BIC scores, and are therefore disregarded. However, it should be noted that two components, or clusters, has almost as large a BIC score as only one component in the *EEI* model. **Figure 7.2** shows the plot of the first two PC scores. The triangles are individuals. The superimposed ellipses correspond to the covariance of the components, or in this case, the one component. **Figure 7.3** plots any uncertainty in the classification. If any individuals were classified with a degree of uncertainty, the dot size would increase and would turn black (explained in more detail below). In this case all of the dots are small and grey with low uncertainty classification, as there is only one cluster. These results would indicate the Mongolian samples are not dissimilar and that multiple subpopulations were not detected.

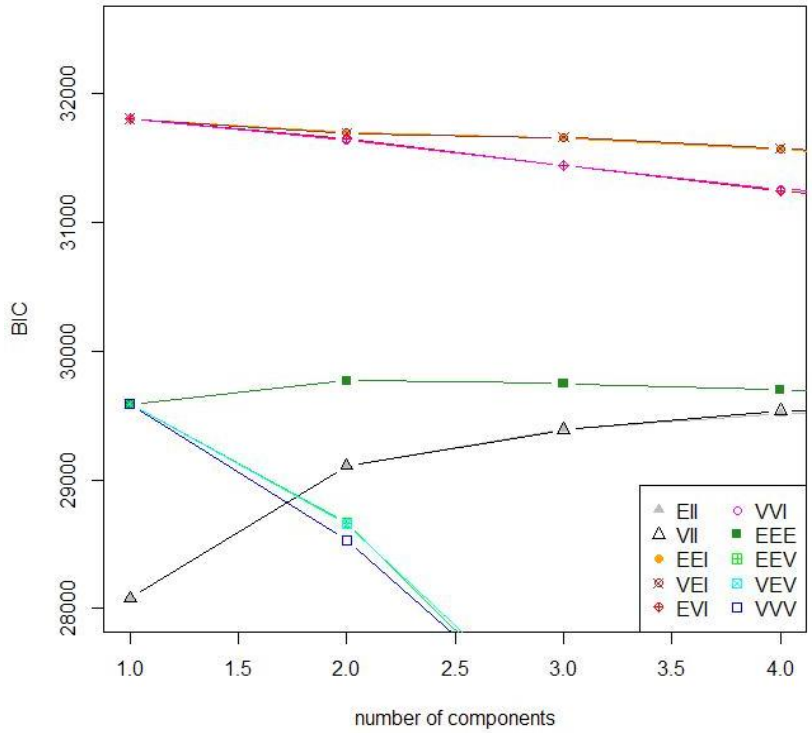

FIGURE 7.1. Number of components (clusters) within the Mongol samples using MCLUST.

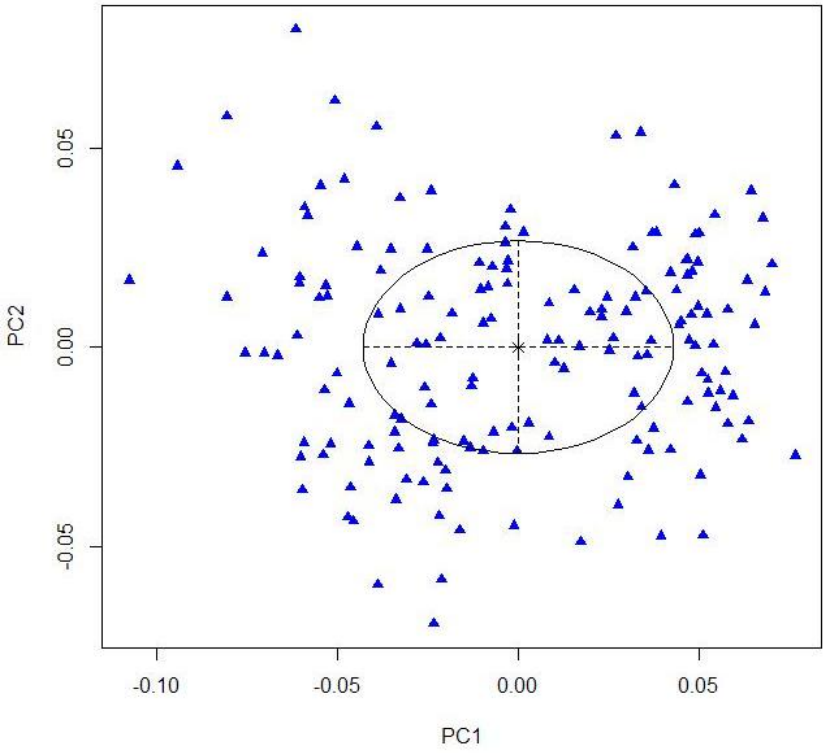

FIGURE 7.2. Classification of the component(s) for the Mongol data.

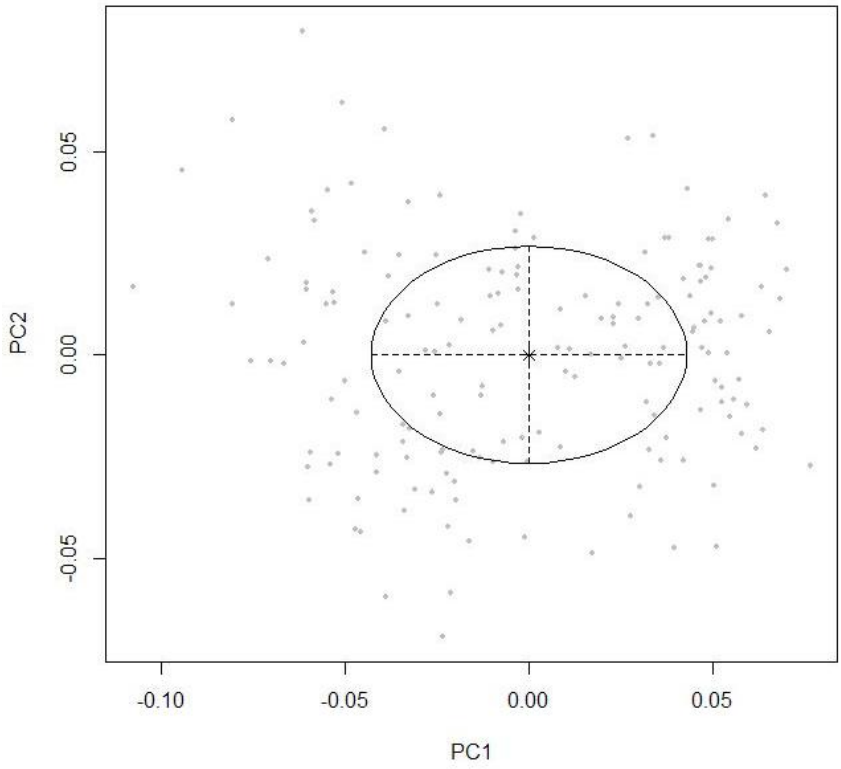

FIGURE 7.3. Uncertainty plot in the MCLUST classification model for Mongol data.

To understand if the Egiin Gol sample could be its own group separate from other Xiongnu individuals, it was analyzed using Mahalanobis distance, principal coordinate plots, Ward's clustering, and a Discriminant function test. **Figure 7.4** shows the results from a principal components test on all Mongolian individuals included in this dissertation (first two PCs account for 41.7% of the sample variance). Though difficult to interpret at first, there does appear to be a general distinction for those individuals labeled as "Egiin Gol" (EG), a sample ascribed to the Xiongnu culture and is located in northern Mongolia. These individuals are clustered together with most of the Bronze Age sample (Chandman), which comes from western Mongolia, and Mongol Turk period individuals along the right side of the first principal component (X-axis). This result is similar to

what was obtained in the Ward's clustering analysis obtained from using individual principal component scores (**Fig. 7.5**). Ward's clearly identifies two separate clusters within the Mongolian data. These results indicate a potential trend for multiple biological groups within the data.

Table 7.1 shows the results for the Mahalanobis distances between the various Mongolian samples. As can be seen, the Egiin Gol sample, which has been ascribed to Xiongnu material culture, is actually more similar to the Mongol Turk period, and then the Mongolian Bronze Age sample. **Figure 7.6** shows a similar result from the discriminant function analysis. The discriminant plot shows the first two functions which account for 91.6% of the variation. There is a strong separation along function 1, with the Mongol Turk, Chandman, and Egiin Gol samples clustering close together, while the modern Mongolians, Mongol Period (Medieval period) and the aggregated Xiongnu sample forming a separate cluster. If the Egiin Gol sample were similar in biological terms, then they should cluster closer to other known Xiongnu samples. However, this is not the case. The groups were also subject to a classification procedure. The results for this are shown in **Table 7.2**. To see if geography is driving some of the variation, the samples were subjected to a Mantel test. These results can be seen in **Table 7.3** and are interpreted as showing a strong isolation by geographic distance model between samples as indicated in the strong correlation between geographic and biological distance based on Mahalanobis distances between groups.

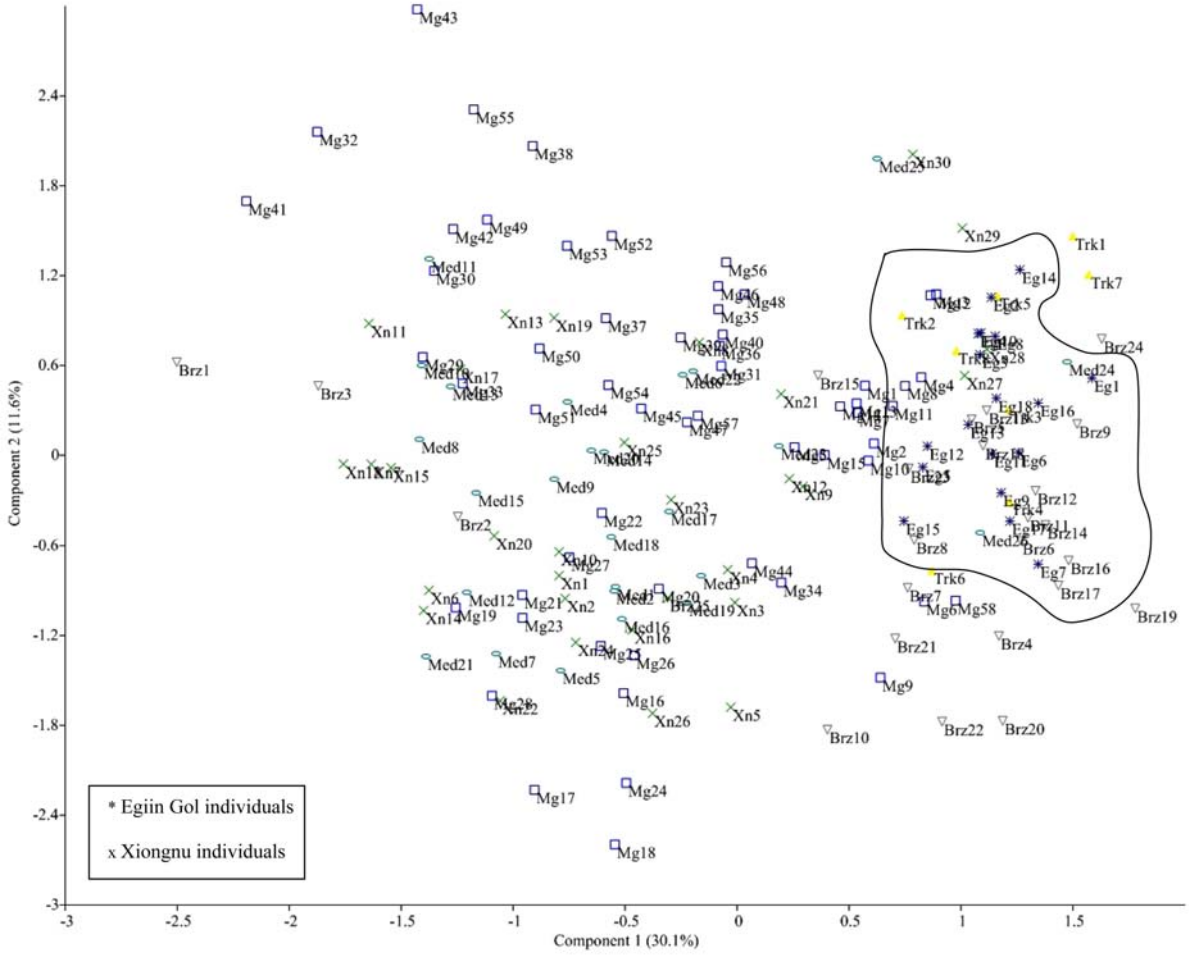

FIGURE 7.4. Principal component plot of individual Mongolian crania. Area circled is Egiin Gol cluster.

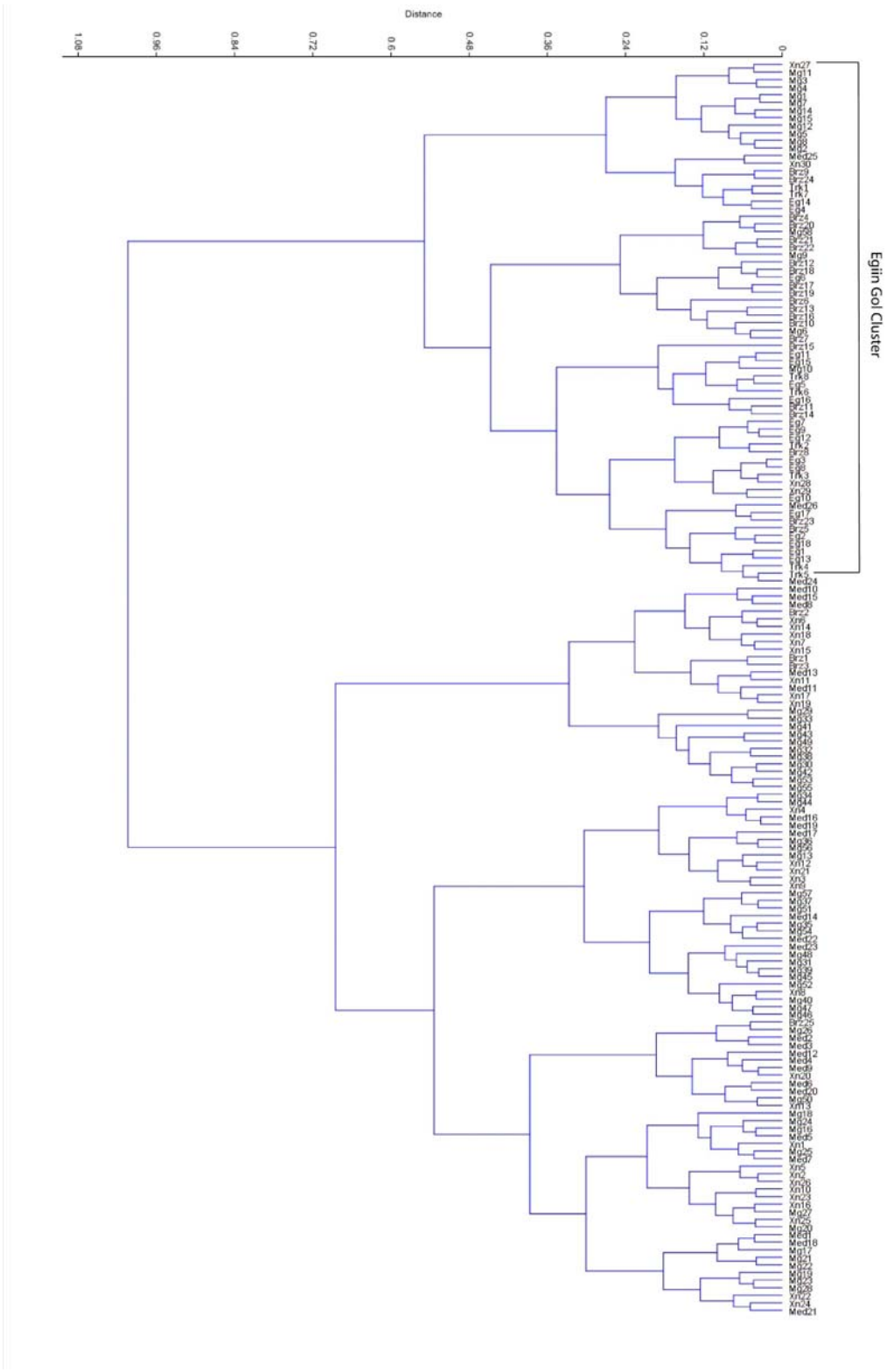

FIGURE 7.5. Ward's Hierarchical Clustering of individual Mongolian crania.

TABLE 7.1. D^2 results for within-group Mongolian variation.

Population	MongolTurk	Chandman	Mongolia	MongolPeriod	Xiongnu	EgiinGol
MongolTurk	0					
Chandman	0.257271	0				
Mongolia	0.249929	0.347111	0			
MongolPeriod	0.235064	0.322821	0.104472	0		
Xiongnu	0.232449	0.207035	0.066059	0.05514	0	
EgiinGol	**0.05896**	**0.133009**	**0.22113**	**0.190915**	**0.146567**	0

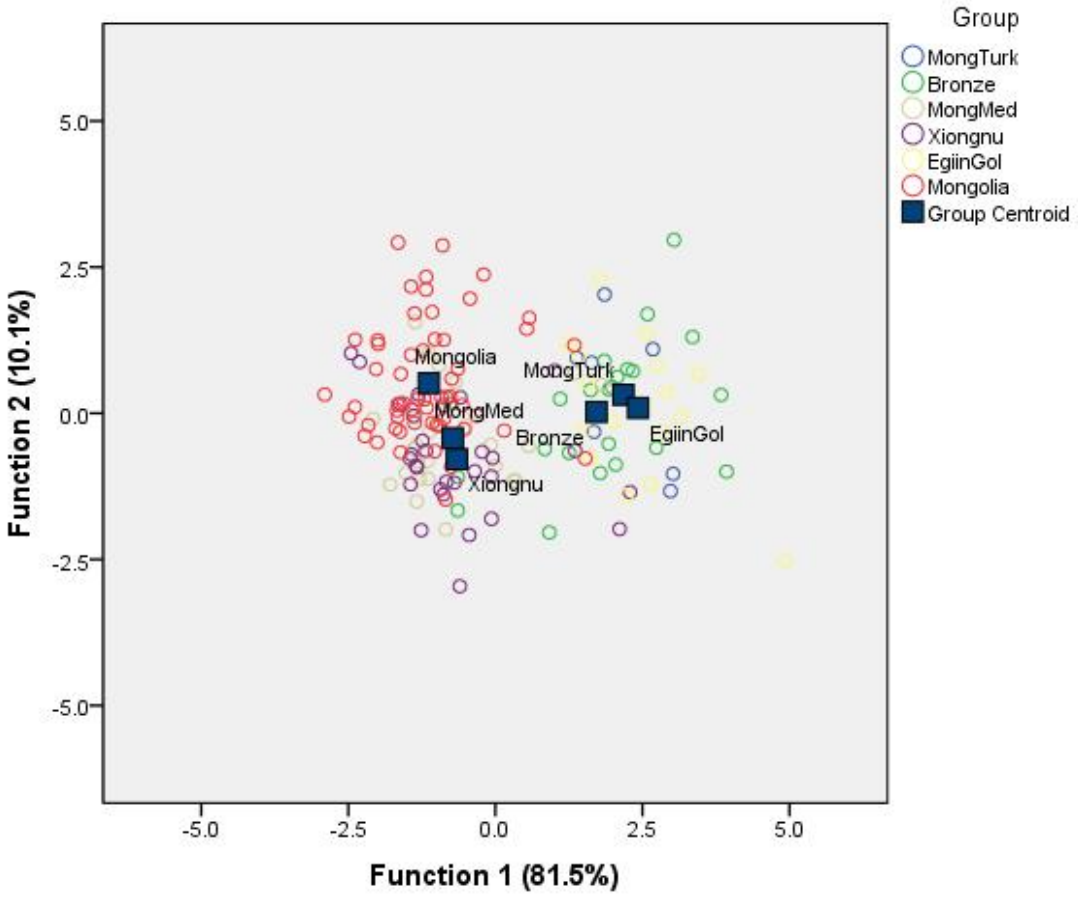

FIGURE 7.6. Discriminant function plot for Mongolian samples.

TABLE 7.2. Cross-validation classification results for Mongolian samples.

			Predicted Group Membership						
		Group	MongTurk	Bronze	MongMed	Xiongnu	EgiinGol	Mongolia	Total
Original	Count	MongTurk	0	2	0	0	6	0	8
		Bronze	0	17	2	0	4	2	25
		MongMed	0	1	8	7	1	9	26
		Xiongnu	0	2	8	10	2	8	30
		EgiinGol	0	4	0	0	14	0	18
		Mongolia	0	2	1	4	0	51	58
	%	MongTurk	.0	25.0	.0	.0	75.0	.0	100.0
		Bronze	.0	68.0	8.0	.0	16.0	8.0	100.0
		MongMed	.0	3.8	30.8	26.9	3.8	34.6	100.0
		Xiongnu	.0	6.7	26.7	33.3	6.7	26.7	100.0
		EgiinGol	.0	22.2	.0	.0	77.8	.0	100.0
		Mongolia	.0	3.4	1.7	6.9	.0	87.9	100.0
Cross-validated	Count	MongTurk	0	2	0	0	6	0	8
		Bronze	0	15	2	1	5	2	25
		MongMed	0	2	4	8	1	11	26
		Xiongnu	0	2	8	7	2	11	30
		EgiinGol	1	5	0	0	12	0	18
		Mongolia	0	2	1	4	0	51	58
	%	MongTurk	.0	25.0	.0	.0	75.0	.0	100.0
		Bronze	.0	60.0	8.0	4.0	20.0	8.0	100.0
		MongMed	.0	7.7	15.4	30.8	3.8	42.3	100.0
		Xiongnu	.0	6.7	26.7	23.3	6.7	36.7	100.0
		EgiinGol	5.6	27.8	.0	.0	66.7	.0	100.0
		Mongolia	.0	3.4	1.7	6.9	.0	87.9	100.0

TABLE 7.3. Partial Mantel Test for Mongolian samples.

Mongolian Series	Partial	r
BIO x GEO	Temporal	0.65407 (0.01840)

As shown in the classification table, zero of the Egiin Gol sample classifies with other known Xiongnu groups. Based on these results, the Egiin Gol sample was not pooled with the larger sample of Xiongnu crania.

Xiongnu Population History

Global Comparative Series: The Xiongnu, Egiin Gol and Mongolian samples were then compared to crania from around the globe, including Africa and Europe. **Table 7.4** shows sample sizes for the global comparative analysis. This analyses was performed in order to place the Xiongnu into a larger regional context.

TABLE 7.4. Regional samples (pooled) used in global cranial comparative analysis.

Population	N
CentralAsia	103
CentralChina	67
EastAsia	11
Europe	20
Mongolia	76
NorthEurope	18
NEChina	31
NESiberia	86
SubSahAfrica	12
SouthAsia	12
SouthChina	28
SSiberia	181
SEAsia	43
WestChina	201
WestSiberia	77
Xiongnu	32
Total	998

The results from the principal coordinate analysis and canonical variate and are shown in **Figures 7.7** and **7.8**, respectively. The CV plots and the PCO plot shows the location of the pooled Xiongnu (not including the Egiin Gol sample) and Mongolian samples (modern and Mongol Period) compared with aggregated regional samples. In **Figure 7.7**, the first two eigenvectors account for 76.8% of the variation, and in **Figure 7.8**, the first two canonical variates account for 63% of the total variation. Both plots show similar results. CV 1 is separating groups from SE and East Asia, Africa and Europe from Northern and Western Chinese, Siberian, and Mongolian groups. CV 2 seems to be separating the Mongolian and Chinese (except the Southern Chinese) from the other groups in the analysis.

In **Figure 7.9**, canonical variate 1 is plotted against canonical variate 3, accounting for 56.1% of the variation. In this plot, it appears that CV 3 is separating out samples from Europe and West China relative to the other groups in the analysis. Though somewhat isolated, the Mongolian and Xiongnu samples fall into the region with other

Chinese samples in the canonical and eigenvector plots, while in **Figure 7.9** the Xiongnu plot close to NE Siberia.

These results suggest the Mongolian groups, including the Xiongnu, have a closer shared population history with groups from China and Siberia than from Central Asia. **Table 7.5** shows the Mahalanobis distances for the pooled regional sample global cranial series. Xiongnu are closest to the aggregated Mongolian sample (0.020569), followed by NE Siberia (0.095383), NE China (0.098229), Southern Siberia (0.101278), and Central China (0.133998).

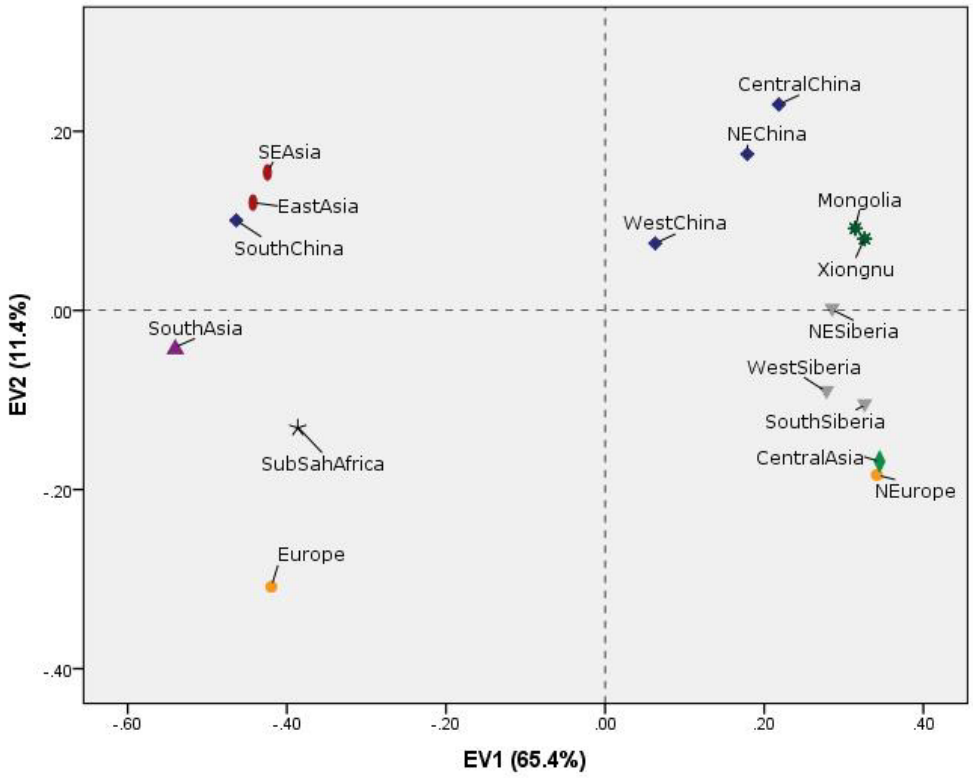

FIGURE 7.7. Principal coordinate plot, pooled global cranial series.

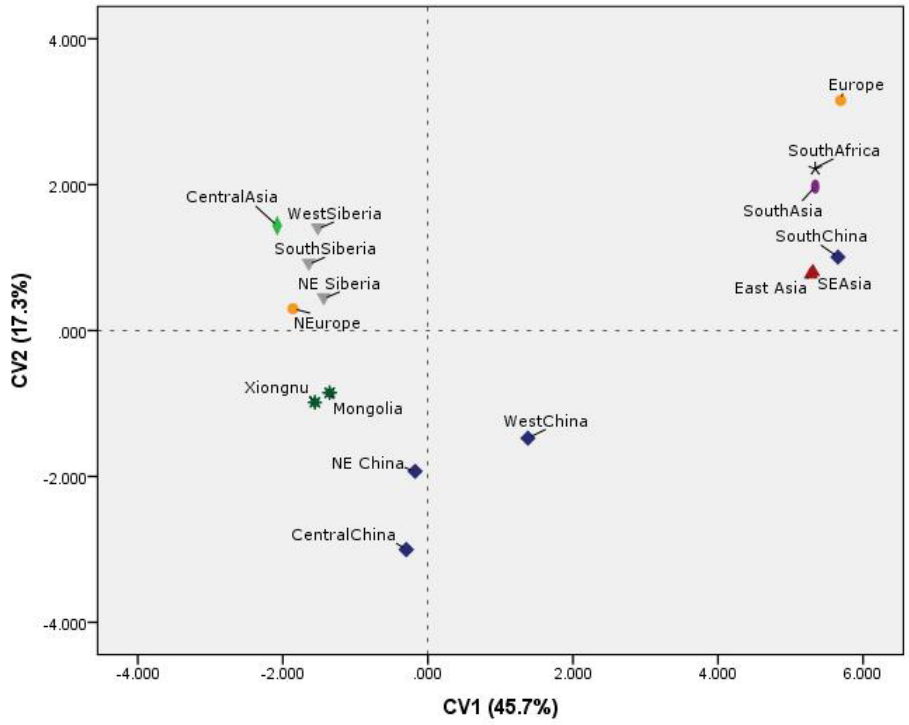

FIGURE 7.8. CV1 against CV2, pooled global cranial series.

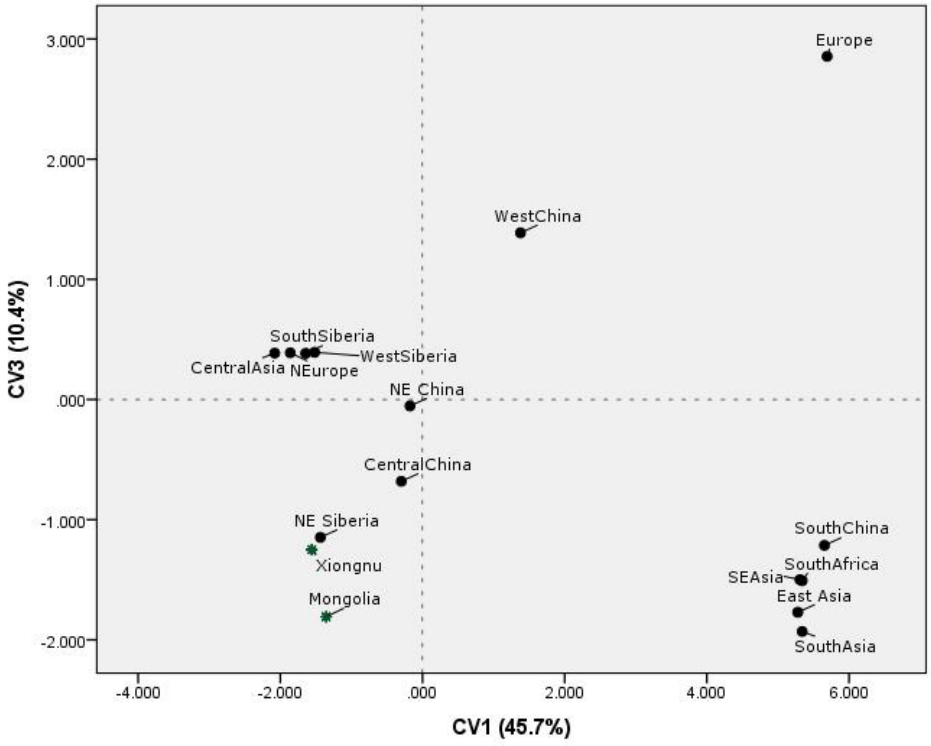

FIGURE 7.9. CV1 plotted against CV3, pooled global cranial series.

TABLE 7.5. Mahalanobis distances for global comparative series.

Population	CenAsia	CenChina	EastAsia	Europe	Mongolia	NEurope	NEChina	NESiberia	SSAfrica	SAsia	SChina	SSiberia	SEAsia	WChina	WSiberia	Xiongnu
CentralAsia	*0.00*															
CentralChina	0.271326	*0.00*														
EastAsia	0.730504	0.533423	*0.00*													
Europe	0.705484	0.782352	0.253608	*0.00*												
Mongolia	0.167213	0.151531	0.594940	0.788653	*0.00*											
NEurope	0.172801	0.225436	0.826980	0.709312	0.188946	*0.00*										
NEChina	0.176771	0.044509	0.404283	0.636091	0.111940	0.243982	*0.00*									
NESiberia	0.062812	0.182891	0.570875	0.744980	0.091935	0.223843	0.111284	*0.00*								
SubSahAfrica	0.699539	0.660684	0.157172	0.301706	0.662902	0.720475	0.554305	0.583042	*0.00*							
SAsia	0.902255	0.724182	0.159611	0.280476	0.909861	0.904924	0.673423	0.801338	0.261036	*0.00*						
SChina	0.754086	0.569324	0.007503	0.227883	0.658331	0.857836	0.433094	0.619080	0.194821	0.147671	*0.00*					
SSiberia	0.017581	0.218003	0.652700	0.682763	0.115667	0.175351	0.125919	0.052144	0.649438	0.865498	0.686124	*0.00*				
SEAsia	0.734372	0.524897	0.026433	0.276648	0.621750	0.833866	0.407080	0.602277	0.276709	0.196255	0.055607	0.665133	*0.00*			
WChina	0.225987	0.111596	0.338066	0.425597	0.210991	0.266244	0.040272	0.193294	0.455305	0.540092	0.336540	0.173011	0.331482	*0.00*		
WSiberia	0.050042	0.253509	0.599423	0.660754	0.148645	0.225344	0.118151	0.088929	0.611070	0.823061	0.629109	0.019744	0.605519	0.164009	*0.00*	
Xiongnu	0.156891	0.133998	0.637412	0.800283	0.020569	0.157054	0.098229	0.095383	0.667854	0.877016	0.665710	0.101278	0.639887	0.192521	0.141384	*0.00*

Tables 7.6 and **7.7** show the results from the Relethford-Blangero analysis for heritabilities 0.55 and 1.0. As the tables make clear, Xiongnu residual variance changes very little between the two heritabilities, showing a positive residual, indicating greater than expected extralocal gene flow compared to the other groups in the analysis. The remainder of the Relethford-Blangero regults use a heritability of 1.0.

TABLE 7.6. Relethford-Blangero Results using h^2 =1.

		Within-Group Phenotypic Variance			
Population	r_{ii}	Observed	Expected	Residual	SE
WChina	0.06114	0.816	0.958	-0.142	0.00384
NEChina	0.07033	0.768	0.948	-0.181	0.01141
NESiberia	0.12442	0.945	0.893	0.052	0.00841
WSiberia	0.13149	0.746	0.886	-0.139	0.00915
SSiberia	0.13581	0.790	0.881	-0.092	0.00598
Xiongnu	0.14577	0.949	0.871	0.077	0.01535
CentralChina	0.14693	0.705	0.870	-0.165	0.01037
Mongolia	0.15055	1.064	0.866	0.198	0.00983
CentralAsia	0.17476	0.898	0.842	0.056	0.00902
EastAsia	0.21126	0.801	0.804	-0.003	0.03301
NEurope	0.22844	0.770	0.787	-0.017	0.02578
SEAsia	0.23455	0.786	0.781	0.005	0.01635
SChina	0.23657	0.820	0.779	0.041	0.02060
SubSahAfrica	0.27193	0.727	0.743	-0.015	0.03493
Europe	0.32530	0.821	0.688	0.133	0.02860
SAsia	0.37260	0.831	0.640	0.191	0.04015

F_{ST} = 0.188865
V_{GW} = 0.827
h^2 = 1.0

TABLE 7.7. Relethford-Blangero Results using h^2 =0.55.

		Within-Group Phenotypic Variance			
Population	r_{ii}	Observed	Expected	Residual	SE
WChina	0.09649	0.816	1.073	-0.257	0.00444
NEChina	0.11836	0.768	1.047	-0.280	0.01316
NESiberia	0.19677	0.945	0.954	-0.009	0.00970
WSiberia	0.20815	0.746	0.941	-0.194	0.01055
SSiberia	0.21280	0.790	0.935	-0.146	0.00690
CentralChina	0.23271	0.705	0.911	-0.206	0.01197
Xiongnu	0.23545	0.949	0.908	0.041	0.01770
Mongolia	0.23786	1.064	0.905	0.159	0.01133
CentralAsia	0.27455	0.898	0.862	0.036	0.01041
EastAsia	0.35389	0.801	0.767	0.034	0.03808
NEurope	0.37079	0.770	0.747	0.023	0.02974
SEAsia	0.37133	0.786	0.747	0.039	0.01886
SChina	0.37793	0.820	0.739	0.081	0.02376
SubSahAfrica	0.44616	0.727	0.658	0.070	0.04030
Europe	0.51992	0.821	0.570	0.251	0.03299
SAsia	0.60275	0.831	0.472	0.359	0.04632

F_{ST} = 0.303493
VGW = 0.827
h^2 = 0.55

TABLE 7.8. R matrix distances for global comparative series.

Population	CenAsia	CenChina	EastAsia	Europe	Mongolia	NEurope	NEChina	NESiberia	SSAfrica	SAsia	SChina	SSiberia	SEAsia	WChina	WSiberia	Xiongnu
CentralAsia	0.174757															
CentralChina	0.025181	0.146931														
EastAsia	-0.172243	-0.087615	0.211261													
Europe	-0.102715	-0.155062	0.141475	0.325297												
Mongolia	0.079049	0.072977	-0.116562	-0.156401	0.150554											
NEurope	0.115197	0.074966	-0.193641	-0.077789	0.095023	0.228437										
NEChina	0.034157	0.086374	-0.061347	-0.120234	0.054471	0.027391	0.070327									
NESiberia	0.118180	0.044227	-0.117600	-0.147634	0.091517	0.064505	0.041729	0.124415								
SubSahAfrica	-0.126426	-0.120912	0.163010	0.147760	-0.120209	-0.110054	-0.106024	-0.093348	0.271930							
SAsia	-0.177451	-0.102328	0.212123	0.208708	-0.193355	-0.151946	-0.115250	-0.152164	0.191745	0.372595						
SChina	-0.171378	-0.092910	0.220165	0.166993	-0.135602	-0.196413	-0.063097	-0.129046	0.156841	0.230748	0.236573					
SSiberia	0.146494	0.032370	-0.152814	-0.110827	0.085349	0.094449	0.040110	0.104041	-0.120848	-0.178546	-0.156870	0.135812				
SEAsia	-0.162532	-0.071707	0.209690	0.141600	-0.118322	-0.185439	-0.051101	-0.121656	0.114886	0.205446	0.207758	-0.147385	0.234551			
WChina	0.004953	0.048236	-0.032834	-0.019582	0.000350	0.011665	0.045596	-0.003871	-0.061119	-0.053180	-0.019415	0.011969	-0.017897	0.061137		
WSiberia	0.128104	0.012457	-0.128335	-0.101983	0.066701	0.067292	0.041834	0.083488	-0.103824	-0.159487	-0.130523	0.123780	-0.119738	0.014309	0.131492	
Xiongnu	0.081820	0.079353	-0.140189	-0.164606	0.137880	0.108579	0.058936	0.087403	-0.125075	-0.179324	-0.141682	0.090154	-0.129781	0.007195	0.067941	0.145774

Table 7.8 shows the R-matrix distances. Here, positive values indicate a closer relationship than on average, while negative values indicate further biological distance than on the average. Xiongnu are closest to Mongolians, followed by Northern Europe, Southern Siberia, NE Siberia, and Central Asia. Interestingly, these results could be indicating a component of Western Eurasian admixture not shown in the Mahalanobis distance results. **Figure 7.10** shows the results from the cluster analysis using Ward's (1963) clustering method with corresponding bootstrap values after 1000 replicates using the R-matrix distances. **Figure 7.11** shows the neighbor-joining tree (Saitou and Nei, 1987) produced from the R-matrix distances using 1000 bootstrap replicates. The samples are clustered into relatively large geographic regions. In the case of the cluster containing the European, African, SE Asian, and Indian samples, this forms one cluster as opposed to several simply due to the lack of samples from these regions.

Results using minimum pairwise F_{ST} distances for the Mongolian samples are shown in **Tables 7.9-11**. The lowest F_{ST} values for the pooled Xiongnu sample are similar to the results obtained from another study (Bennett and Kaestle, 2006) for the overall Egiin Gol series based on mtDNA. The Egiin Gol sample used in this dissertation shows very different results (**Table 7.9**). Whereas the pooled Xiongnu seem to be more similar to other Northeastern Asian groups, the Egiin Gol sample is more related to groups from South Asia (India), Europe (Czech, Austria), Africa (Bushmen), and the archaic populations of East Asia (Jomon, Ainu). It is interesting to note that those groups most similar to the Egiin Gol in the Bennett and Kaestle (2006) study, some are from Central and Southwestern Asia, such as the Lombadi and Lobana, groups sampled from northern India, similar to those Indian individuals included in the craniofacial analysis in

this study (sample comes from Northern India). Egiin Gol also shows a close relationship to the Chandman and Mongol Turk samples, similar to PCA. The Chandman and Mongol period results are discussed in more detail in the Discussion chapter.

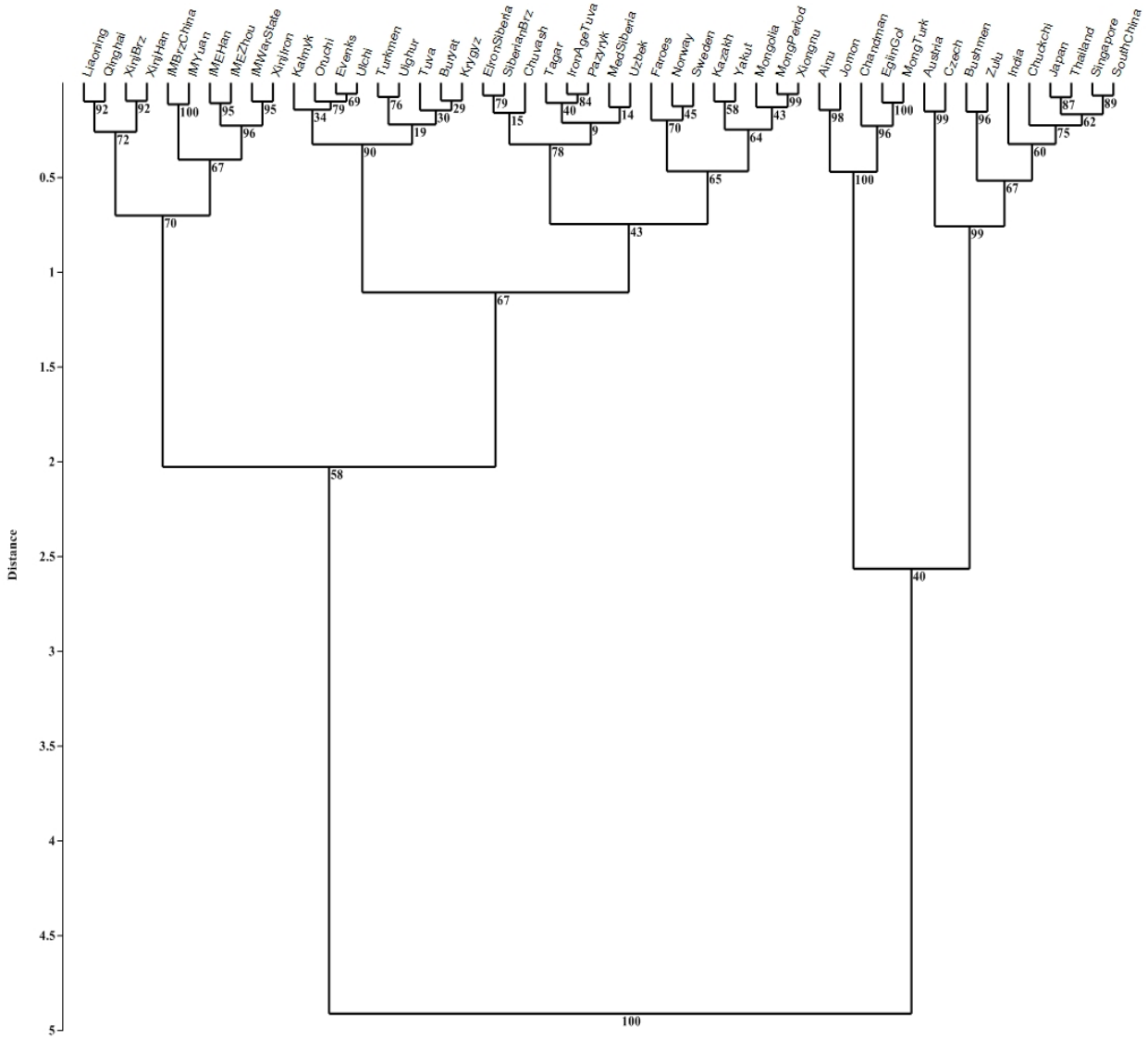

FIGURE 7.10. Ward's hierarchical clustering, global comparative series with bootstrap values after 1000 replicates. R-matrix distances used to construct tree.

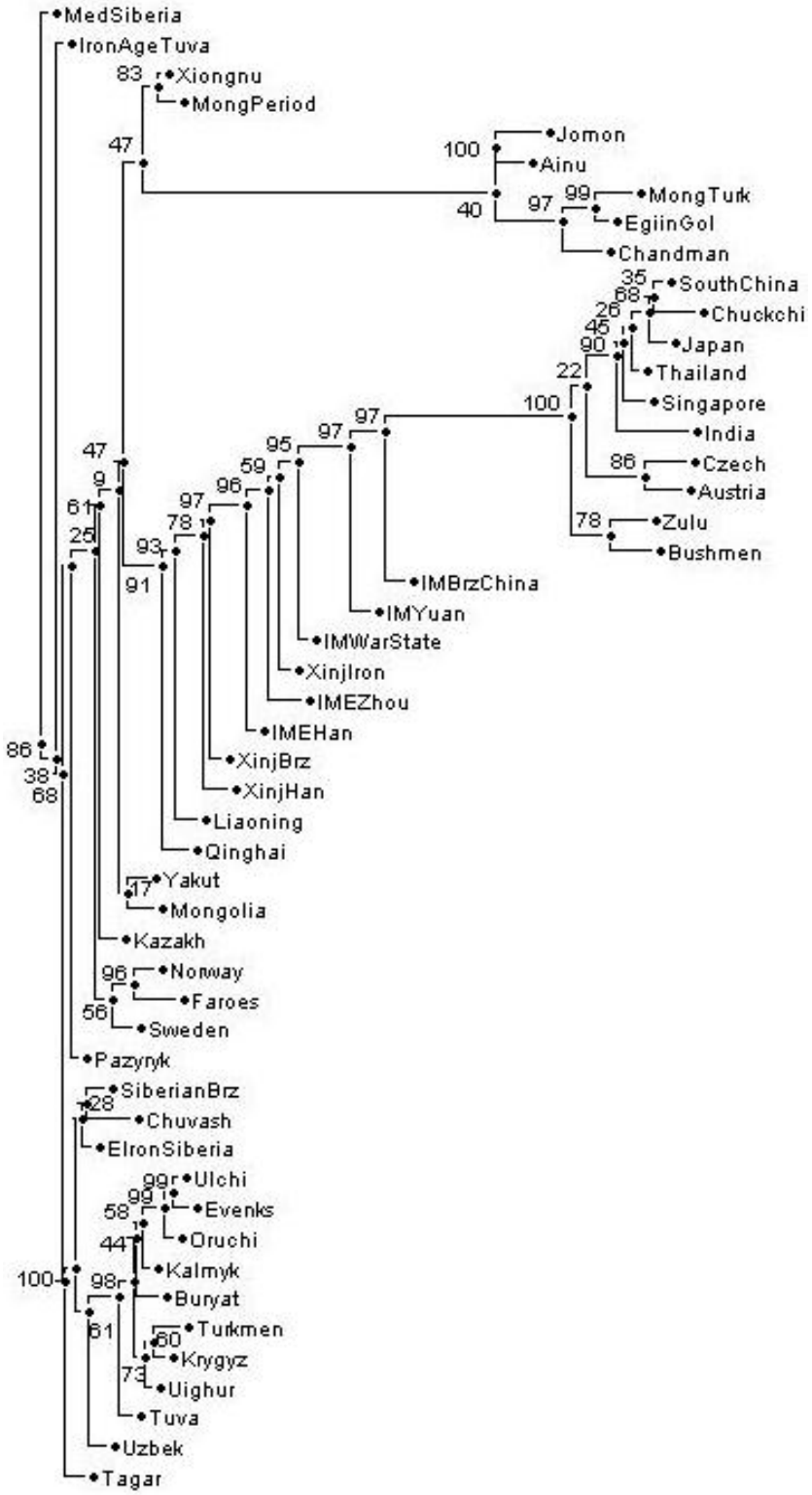

FIGURE 7.11. Neighbor-Joining Tree with corresponding bootstrap values, global comparative series using R-matrix distances.

TABLE 7.9. Pairwise F_{ST} comparisons for Egiin Gol and Xiongnu Samples.

Egiin Gol		Egiin Gol		Xiongnu		Xiongnu	
Group	F_{ST}	Group	F_{ST}	Group	F_{ST}	Group	F_{ST}
Jomon	0.00973	Yakut	0.52287	XinjBronze	0.00031	Krygyz	0.11720
India	0.04683	Norway	0.52312	IMEHan	0.00239	Buryat	0.11963
Czech	0.07543	SiberianBronze	0.52912	XinjHan	0.00514	Sweden	0.12025
Chandman	0.07603	Uighur	0.53196	Tagar	0.00529	Oruchi	0.13303
Bushmen	0.10144	Kalmyk	0.53334	Kazakh	0.00621	Ulchi	0.13707
Austria	0.10181	Tuva	0.53397	MedSiberia	0.01457	Turkmen	0.14306
Chuckchi	0.12862	Qinghai	0.53641	Mongolia	0.01803	Chuvash	0.14856
Ainu	0.13972	IMBronzeChina	0.56268	XinjIron	0.02006	Evenks	0.17907
MongTurk	0.14393	MongPeriod	0.56335	IMWarState	0.02149	Faroes	0.22298
Zulu	0.19921	Uzbek	0.56583	Liaoning	0.02708	Thailand	0.29266
Japan	0.26849	EIronSiberia	0.57367	Pazyryk	0.02766	SouthChina	0.32805
Singapore	0.27425	IMWarState	0.58241	EIronSiberia	0.03024	Singapore	0.32965
SouthChina	0.27586	Mongolia	0.58588	Uzbek	0.03807	Japan	0.33541
Thailand	0.31125	Kazakh	0.59770	MongPeriod	0.04056	Zulu	0.42476
Faroes	0.38093	XinjHan	0.59877	IMBronzeChina	0.04123	Ainu	0.46419
Evenks	0.42484	IMEHan	0.60151	IronAgeTuva	0.04918	Chuckchi	0.47528
Chuvash	0.45534	Xiongnu	0.60390	Qinghai	0.06750	Austria	0.50209
Turkmen	0.46084	XinjBronze	0.60422	Tuva	0.06994	Bushmen	0.50246
Ulchi	0.46684	Tagar	0.60920	Kalmyk	0.07056	India	0.55708
Oruchi	0.47088	MedSiberia	0.61847	Uighur	0.07195	EgiinGol	0.60390
Sweden	0.48366	XinjIron	0.62397	SiberianBronze	0.07479	Jomon	0.61364
Buryat	0.48428	Liaoning	0.63099	Norway	0.08078	Czech	0.67934
Krygyz	0.48671	Pazyryk	0.63157	IMYuan	0.08239	Chandman	0.67994
IMEZhou	0.50857	IronAgeTuva	0.65309	IMEZhou	0.09534	MongTurk	0.74784
IMYuan	0.52152			Yakut	0.10110		

TABLE 7.10. Pairwise F_{ST} comparisons for Chandman Sample.

Chandman		Chandman	
Group	F_{ST}	Group	F_{ST}
Czech	0.00060	Yakut	0.59891
Jomon	0.06630	Norway	0.59916
MongTurk	0.06790	SiberianBronze	0.60515
EgiinGol	0.07603	Uighur	0.60799
India	0.12286	Kalmyk	0.60938
Bushmen	0.17748	Tuva	0.61000
Austria	0.17785	Qinghai	0.61244
Chuckchi	0.20466	IMBronzeChina	0.63871
Ainu	0.21575	MongPeriod	0.63938
Zulu	0.27524	Uzbek	0.64187
Japan	0.34453	EIronSiberia	0.64970
Singapore	0.35029	IMWarState	0.65845
SouthChina	0.35189	Mongolia	0.66191
Thailand	0.38728	Kazakh	0.67373
Faroes	0.45696	XinjHan	0.67480
Evenks	0.50087	IMEHan	0.67755
Chuvash	0.53138	Xiongnu	0.67994
Turkmen	0.53688	XinjBronze	0.68025
Ulchi	0.54287	Tagar	0.68523
Oruchi	0.54691	MedSiberia	0.69451
Sweden	0.55969	XinjIron	0.70000
Buryat	0.56031	Liaoning	0.70702
Krygyz	0.56274	Pazyryk	0.70760
IMEZhou	0.58460	IronAgeTuva	0.72912
IMYuan	0.59755		

TABLE 7.11. Pairwise F_{ST} comparisons for Mongol Period and modern Mongol Samples.

Mongol Period		Mongol Period		Modern Mongol		Modern Mongol	
Group	F_{ST}	Group	F_{ST}	Group	F_{ST}	Group	F_{ST}
IMBronzeChina	0.00067	Buryat	0.07907	IMWarState	0.00346	Krygyz	0.09917
Uzbek	0.00249	Sweden	0.07969	Kazakh	0.01182	Buryat	0.10160
EIronSiberia	0.01032	IronAgeTuva	0.08974	EIronSiberia	0.01221	Sweden	0.10222
IMWarState	0.01907	Oruchi	0.09247	XinjHan	0.01289	Oruchi	0.11500
Mongolia	0.02253	Ulchi	0.09651	IMEHan	0.01563	Ulchi	0.11904
Qinghai	0.02694	Turkmen	0.10250	Xiongnu	0.01803	Turkmen	0.12504
Tuva	0.02938	Chuvash	0.10800	XinjBronze	0.01834	Chuvash	0.13053
Kalmyk	0.03000	Evenks	0.13851	Uzbek	0.02004	Evenks	0.16104
Uighur	0.03139	Faroes	0.18242	MongPeriod	0.02253	Faroes	0.20495
SiberianBronze	0.03423	Thailand	0.25210	IMBronzeChina	0.02320	Thailand	0.27463
Kazakh	0.03435	SouthChina	0.28749	Tagar	0.02332	SouthChina	0.31002
XinjHan	0.03542	Singapore	0.28909	MedSiberia	0.03260	Singapore	0.31162
IMEHan	0.03817	Japan	0.29485	XinjIron	0.03809	Japan	0.31738
Norway	0.04022	Zulu	0.36414	Liaoning	0.04511	Zulu	0.38667
Yakut	0.04047	Ainu	0.42363	Pazyryk	0.04569	Ainu	0.44616
Xiongnu	0.04056	Chuckchi	0.43472	Qinghai	0.04947	Chuckchi	0.45726
XinjBronze	0.04087	Austria	0.46153	Tuva	0.05191	Austria	0.48406
IMYuan	0.04183	Bushmen	0.46190	Kalmyk	0.05253	Bushmen	0.48443
Tagar	0.04585	India	0.51652	Uighur	0.05392	India	0.53905
IMEZhou	0.05478	EgiinGol	0.56335	SiberianBronze	0.05676	EgiinGol	0.58588
MedSiberia	0.05513	Jomon	0.57308	Norway	0.06275	Jomon	0.59561
XinjIron	0.06062	Czech	0.63878	Yakut	0.06301	Czech	0.66131
Liaoning	0.06764	Chandman	0.63938	IMYuan	0.06436	Chandman	0.66191
Pazyryk	0.06822	MongTurk	0.70728	IronAgeTuva	0.06721	MongTurk	0.72981
Krygyz	0.07664			IMEZhou	0.07731		

I tested for significant number of clusters using all the samples included in the global cranial analysis via the model-based procedure in *MCLUST*. These results, using individual PC scores, are presented in **Figures 7.12 – 7.16**. Based on the BIC scores (**Fig. 7.12**), the global dataset has 4 (BIC = 554548.1) or 5 (BIC = 554500.6) components using the **VEI** model (diagonal distribution, variable volume with equal covariance shape). **Figure 7.13** shows the classification for all 50 groups using individual crania. Cluster 1 consists of 225 crania, cluster 2 consists of 10 crania, cluster 3 consists of 850 crania, and cluster 4 consists of 47 crania. The ellipses correspond to the covariances of the components. **Figure 7.14** plots uncertainty within the covariance components. The large filled symbols (grey and black) indicate these individuals are within the 95% quantile of uncertainty, and the smaller filled dots, the first three quarters of uncertainty. Quantile plots for PCs 1 and 2 are shown in **Figures 7.15** and **7.16**. The amount of

uncertainty within the data is based on overlapping clusters of variance. As has been pointed out by many previous studies (Lewontin, 1972; Relethford, 1994), the amount of within-group variance is higher for humans than between-group variance. These results support this observation.

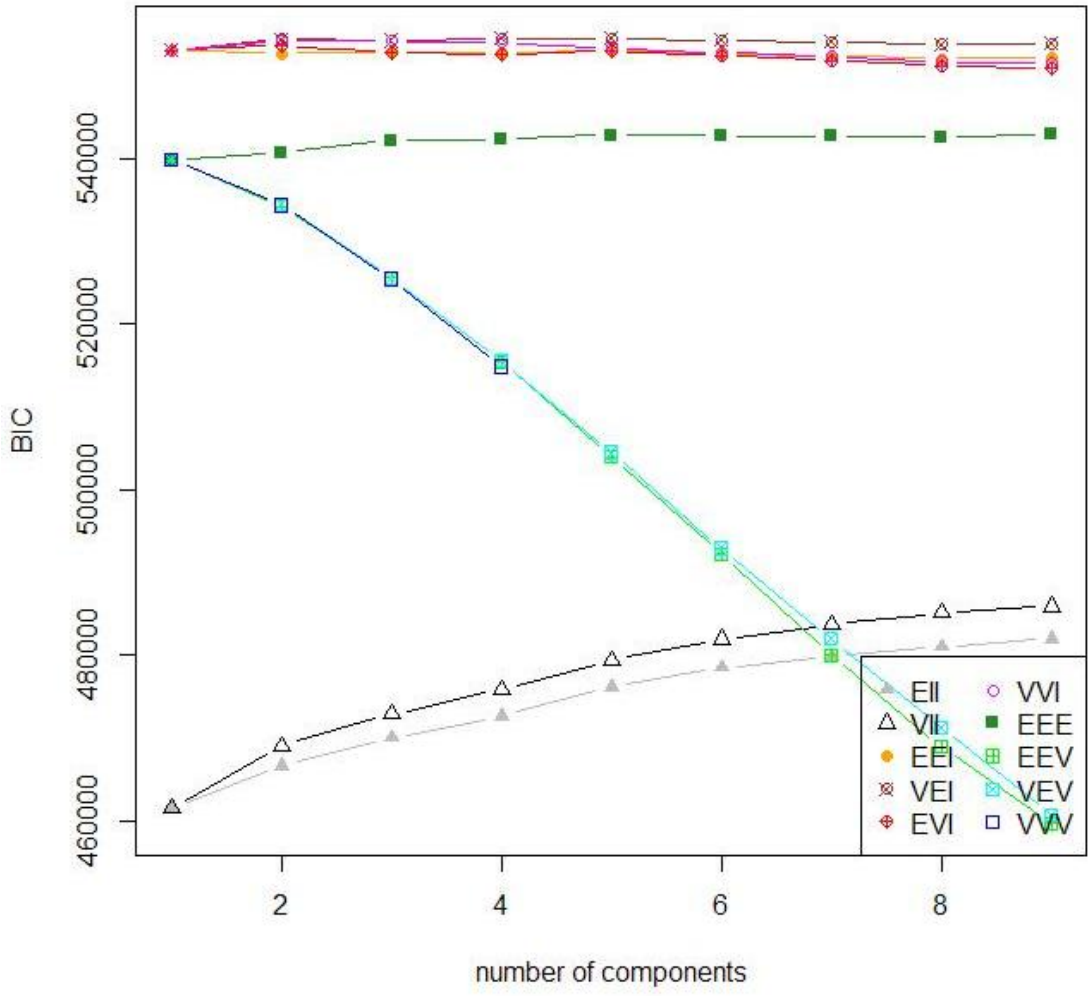

FIGURE 7.12. Number of components (clusters) within the Global dataset using MCLUST. Four or five clusters are detected.

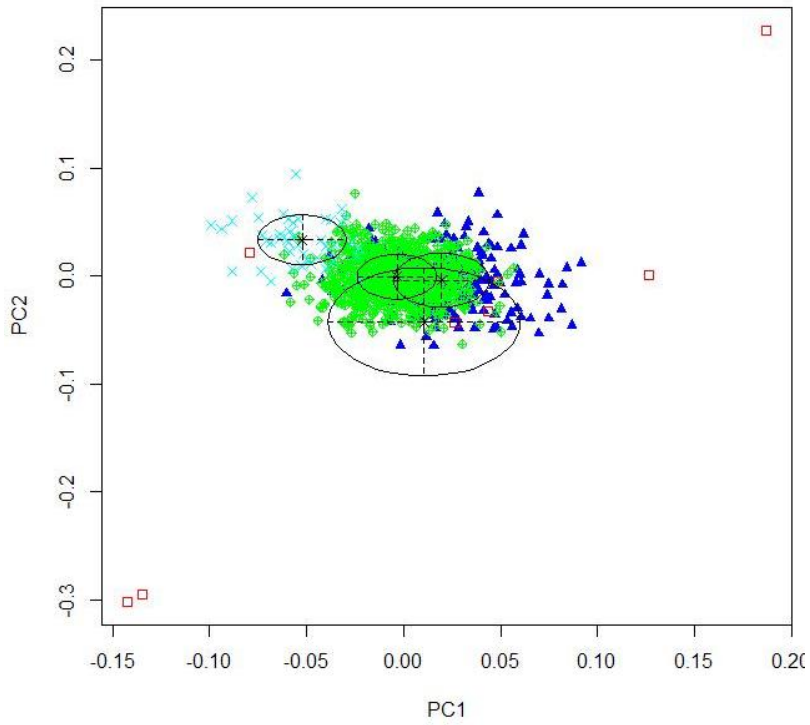

FIGURE 7.13. Classification Plot based on model-based clustering for global dataset. Symbols correspond to clusters for individuals within the data.

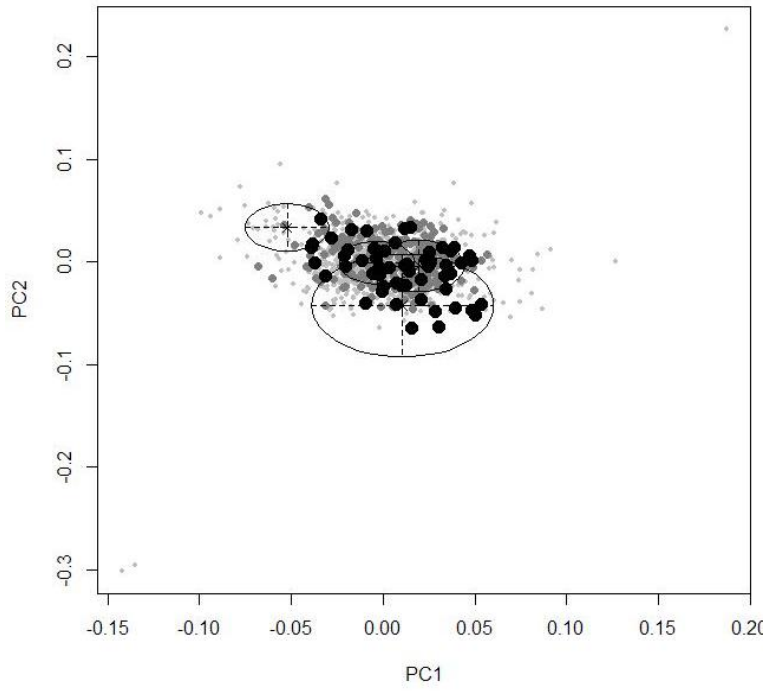

FIGURE 7.14. Uncertainty plot within the components for the global dataset. Large, filled circles are individuals with classification uncertainty.

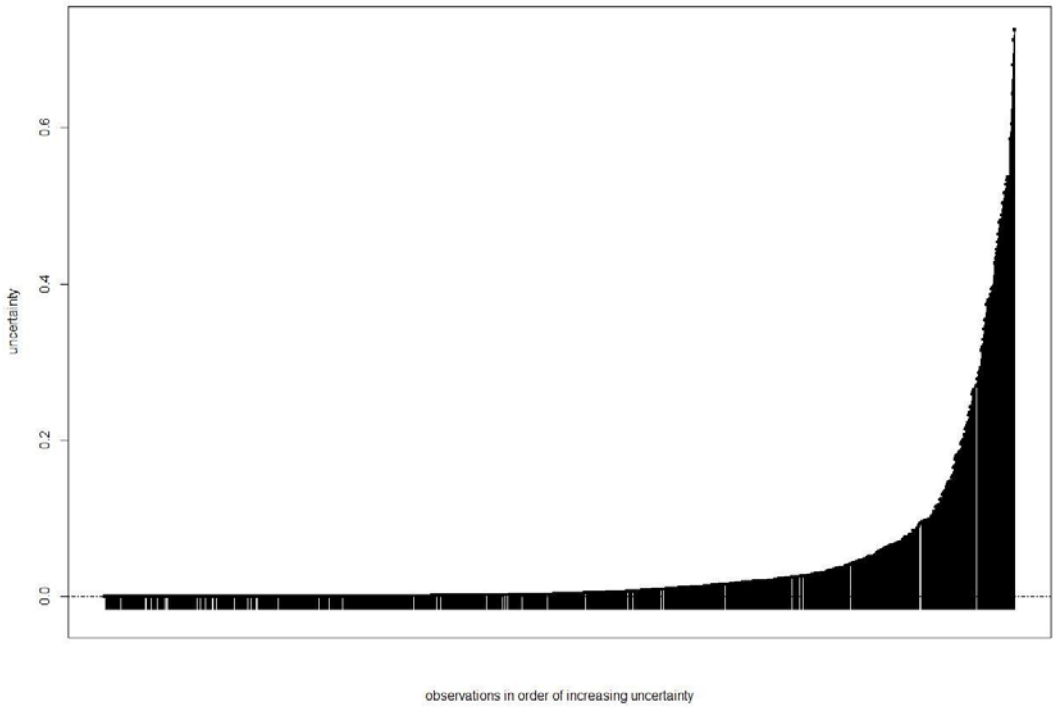

FIGURE 7.15. Uncertainty plot for the 4-cluster mixture model fit of the global dataset via EM based on unconstrained Gaussian mixtures. The vertical lines indicate misclassified observations for PC 1.

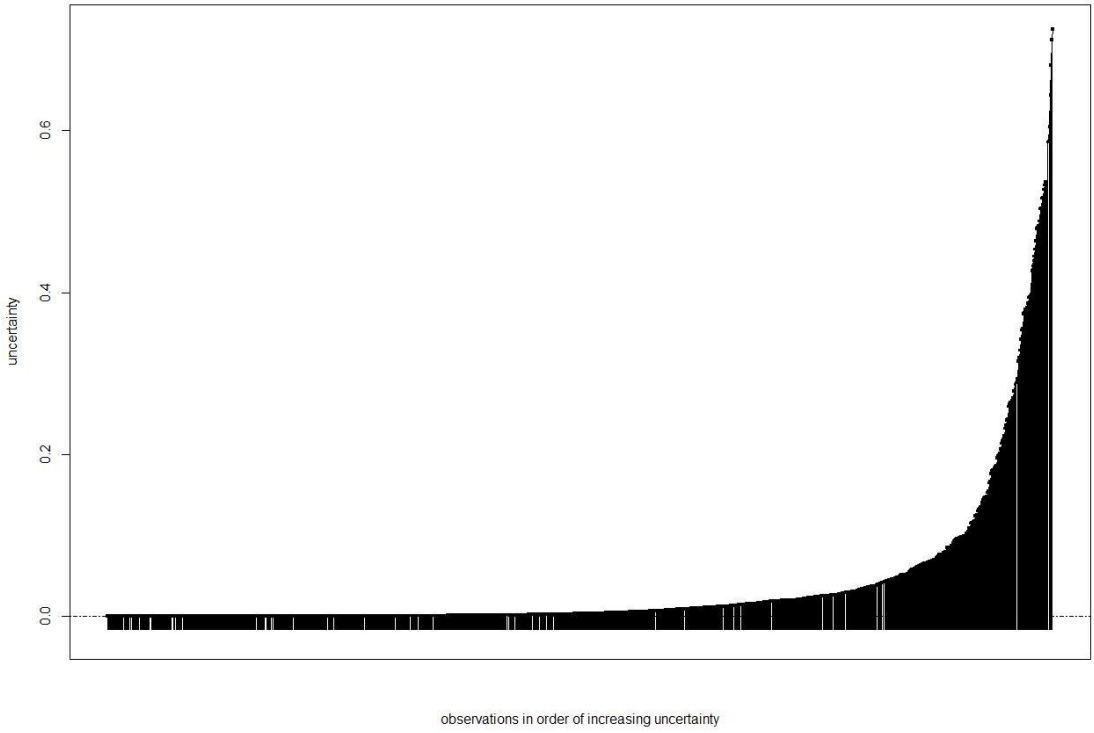

FIGURE 7.16. Uncertainty plot for the 4-cluster mixture model fit of the global dataset via EM based on unconstrained Gaussian mixtures. The vertical lines indicate misclassified observations for PC 2.

Chinese Comparative Series: Mongolian samples were then analyzed against the Chinese series in order to better construct group relationships on a local, rather than a regional or global scale. **Table 7.12** shows the groups and sample sizes used in the Chinese comparative series.

TABLE 7.12. Samples included in Chinese comparative analysis.

Population	N
Chandman	26
InnerMongBronze	22
InnerMongEHan	15
InnerMongEZhou	15
InnerMongWarState	44
Yuan	14
Liaoning	32
ModernMongol	38
MongPeriod	28
MongolTurk	8
Qinghai	67
SouthChina	28
Xinjiang Bronze	21
Xinjiang Han	56
Xinjiang Iron	14
Xiongnu	33
EgiinGol	19
Total	480

The principal coordinate plot is shown in **Figure 7.17**. The first two eigenvectors account for 76.6 percent of the variance. Results indicate a separation of the Egiin Gol, Mongol Turk, and Chandman samples, while the Xiongnu, modern Mongols, and Medieval Period Mongols are plotted along the bottom of EV2. Although the Mongolian samples are isolated, they cluster closest to Qinghai (Central China) and Liaoning (NE China). EV2 is clearly separating the Southern Chinese from the rest of the Chinese samples, while the Xinjiang province samples are dispersed throughout samples from Inner Mongolia, indicating a close relationship among Chinese samples in North and West China.

Results from the Principal components analysis (**Fig. 7.18**) shows similar results to the principal coordinate analysis. However, South China is less separated in the PC plot. PC1 versus PC3 (**Fig. 7.19**) shows a similar result for the Mongolian samples, with the Xiongnu isolated from the Chinese groups. The Chandman remain separated from the other groups along PC1, however, the Egiin Gol samples is closely related to the Xinjiang Iron Age and Xinjiang Han sample along PC3. When PC2 is plotted against PC3 (**Fig. 7.20**), the Mongolian sample and the Xiongnu sample cluster in between the Inner Mongolian Eastern Zhou and the Inner Mongolian Eastern Han. This could be indicative of a closer biological relationship.

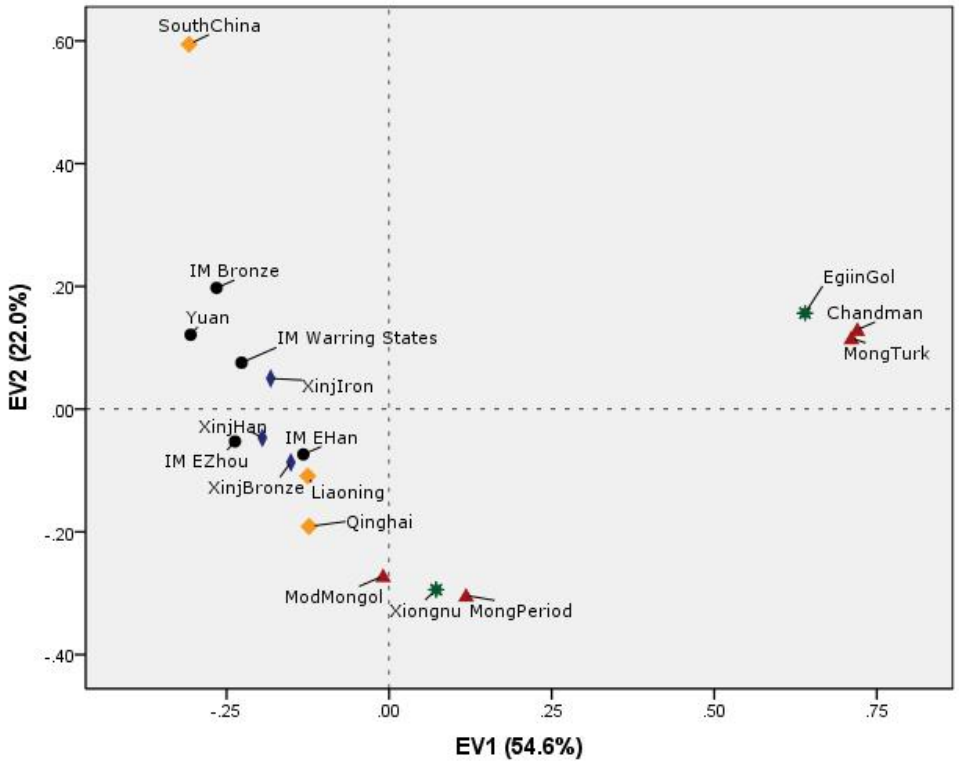

FIGURE 7.17. Principal Coordinate Plot showing comparative Chinese samples.

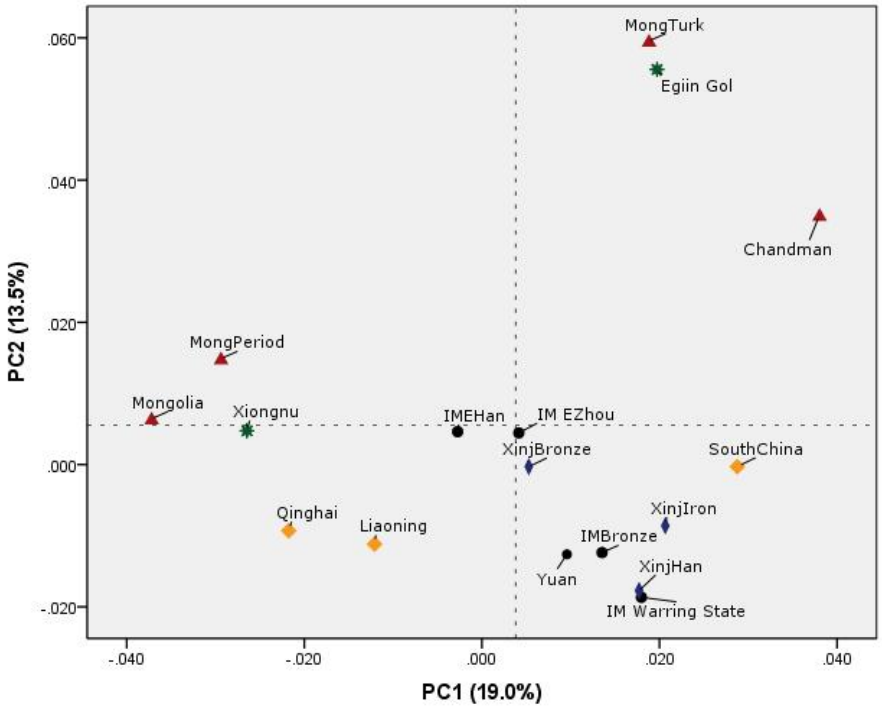

FIGURE 7.18. PC1 plotted against PC2 for comparative Chinese series.

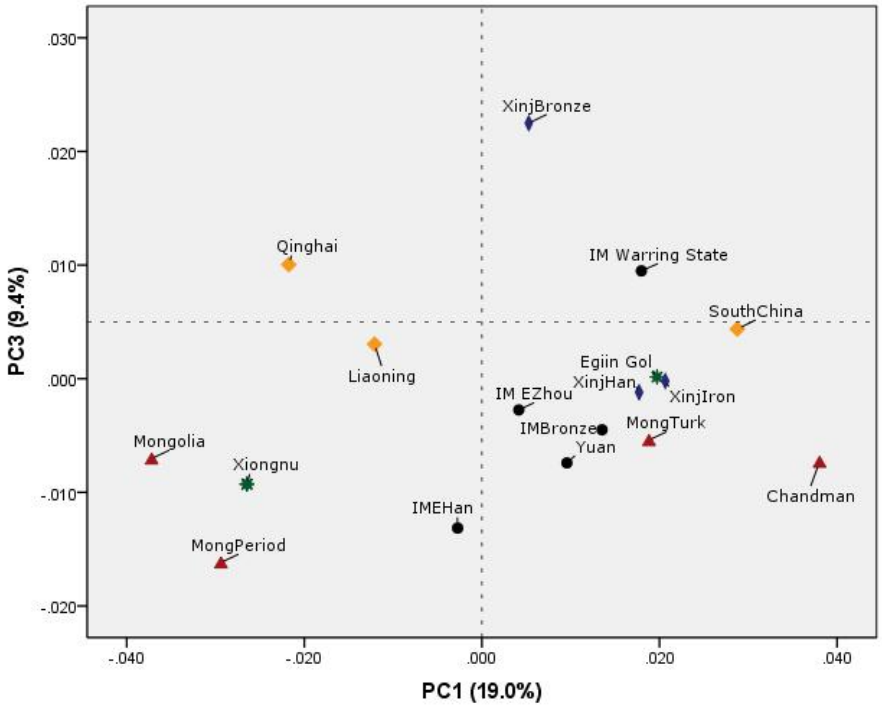

FIGURE 7.19. PC1 plotted against PC3 for comparative Chinese series.

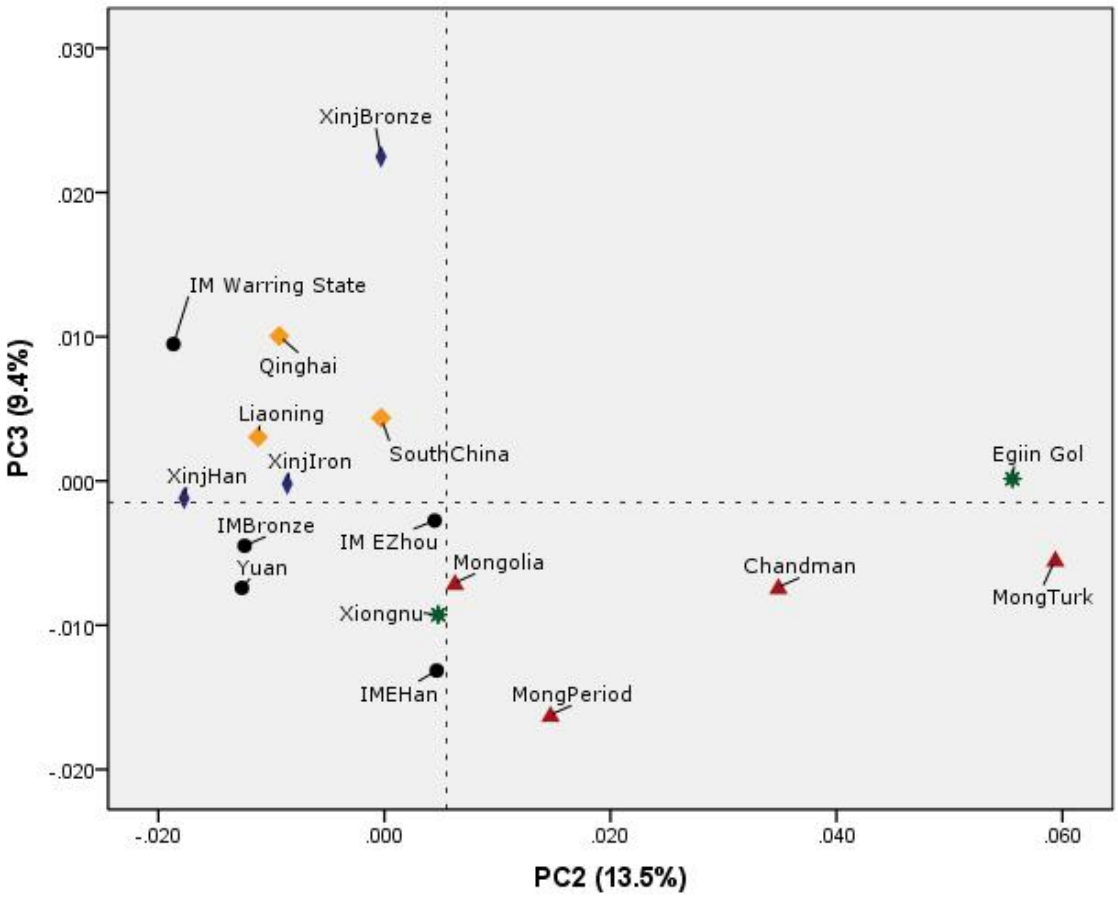

FIGURE 7.20. PC2 plotted against PC3 for comparative Chinese series.

TABLE 7.13. Mahalanobis distances for Chinese comparative series.

Population	Chandman	IMBronze	IMEHan	IMEZhou	IMWarring	Yuan	Liaoning	Mong	MongPer	MongTurk	Qinghai	SChina	XinjBrz	XinjHan	XinjIron	Xiongnu	EgiinGol
Chandman	*0.00*																
IMBronze	1.033691	*0.00*															
IMEhan	0.901208	0.126942	*0.00*														
IMEzhou	1.141619	0.178127	0.102105	*0.00*													
IMWar	0.994835	0.089006	0.171347	0.250260	*0.00*												
Yuan	1.178509	0.041076	0.176946	0.129195	0.183685	*0.00*											
Liaoning	0.844008	0.115926	0.083123	0.147569	0.097344	0.135270	*0.00*										
Mongolia	0.854177	0.353392	0.134301	0.241528	0.343211	0.301345	0.095403	*0.00*									
MongPeriod	0.652939	0.439227	0.134816	0.359164	0.443286	0.436627	0.166225	0.049691	*0.00*								
MongolTurk	0.167952	1.000796	0.803106	1.027555	0.974477	1.094814	0.777875	0.707961	0.616984	*0.00*							
Qinghai	0.943001	0.224005	0.146092	0.255393	0.158966	0.257151	0.052196	0.128861	0.199566	0.849498	*0.00*						
SChina	1.431875	0.239044	0.539883	0.617574	0.467991	0.337135	0.614535	0.864218	1.014292	1.361521	0.765104	*0.00*					
XinjBronze	0.884153	0.160957	0.107573	0.182860	0.101787	0.225583	0.075425	0.204975	0.266356	0.868030	0.087705	0.647965	*0.00*				
XinjHan	0.950013	0.187343	0.188050	0.284990	0.077326	0.211619	0.114980	0.275733	0.355661	1.004212	0.215859	0.661267	0.118714	*0.00*			
XinjIron	0.864586	0.084860	0.102040	0.195372	0.081557	0.234919	0.124270	0.315525	0.353767	0.929474	0.180002	0.489253	0.045707	0.099601	*0.00*		
Xiongnu	0.664530	0.386509	0.128610	0.284459	0.326530	0.393936	0.108493	0.045383	0.023452	0.654770	0.129938	0.959203	0.189707	0.276832	0.291638	*0.00*	
Egiin Gol	0.096825	0.822531	0.663032	0.884516	0.819785	0.944205	0.670729	0.651930	0.538969	0.000000	0.759864	1.153652	0.738945	0.853089	0.768589	0.541684	*0.00*

Table 7.13 shows the results for the Mahalanobis distances between samples in the Chinese comparative series. Similar to the PC plot, the results show the Xiongnu as being closest to modern Mongolians and Medieval period Mongolians, followed by Qinghai and Liaoning. **Table 7.14** shows the results from the Relethford-Blangero analysis (results from $h^2 = 0.55$ not shown). Here r_{ii} (distance from the centroid) indicates the Xiongnu are between the Inner Mongolian Bronze Age and Qinghai (central China) samples. The residual variance for the Xiongnu is negative, indicating some possible drift or isolation compared with other samples in the analysis. The Egiin Gol sample falls between Inner Mongolia Eastern Zhou and Southern Chinese with a positive residual variance, indicating possible gene flow. The F_{ST} (biological divergence) is 0.21, indicating the majority of the variance is within-group, rather than between-groups.

TABLE 7.14. Relethford-Blangero results for Chinese comparative series.

		Within-Group Phenotypic Variance			
Population	r_{ii}	Observed	Expected	Residual	SE
Liaoning	0.04003	1.021	1.084	-0.063	0.009448
InnerMongEHan	0.05476	0.821	1.067	-0.246	0.017362
Xinjiang Bronze	0.07925	0.676	1.040	-0.363	0.015871
Xinjiang Iron	0.09283	0.650	1.024	-0.374	0.021709
Qinghai	0.10745	0.736	1.008	-0.272	0.009382
Xiongnu	0.10963	0.981	1.005	-0.024	0.013931
InnerMongBronze	0.11331	0.963	1.001	-0.039	0.017815
ModernMongol	0.11939	0.954	0.994	-0.040	0.013380
InnerMongWarState	0.12041	0.705	0.993	-0.288	0.026702
Xinjiang Han	0.13799	0.730	0.973	-0.244	0.011604
MongPeriod	0.14727	1.026	0.963	0.063	0.017398
Yuan	0.15876	0.988	0.950	0.038	0.026702
InnerMongEZhou	0.15906	0.726	0.949	-0.223	0.025658
EgiinGol	0.43195	0.990	0.641	0.348	0.035185
SouthChina	0.50689	0.909	0.557	0.352	0.031015
MongolTurk	0.54126	0.911	0.518	0.393	0.062239
Chandman	0.59140	1.442	0.461	0.981	0.034720

$F_{ST} = 0.206567$
$h^2 = 1.0$
$V_{GW} = 0.896$

T**able 7.15** shows the temporal and geographic distance matrices for the Chinese series.

TABLE 7.15. Temporal and geographic distances for Chinese comparative series.

Population	Chandman	IMBronze	IMEHan	IMEZhou	IMWarring	Yuan	Liaoning	Mong	MongPer	MongTurk	Qinghai	SChina	XinjBrz	XinjHan	XinjIron	Xiongnu	EgiinGol
Chandman	0	1835.00	1808.20	1903.38	1864.04	853.03	2572.82	1109.40	1109.40	852.52	1441.04	3515.62	746.99	821.49	669.33	1138.27	591.91
IMBronze	773	0	1078.45	73.49	33.08	1001.23	988.34	876.12	876.12	1001.55	963.56	1975.40	1497.52	1833.43	1794.34	769.63	1367.43
IMEhan	673	1446	0	1127.10	1102.87	1103.38	991.02	787.98	787.98	1104.16	1796.43	2975.06	1919.29	2247.21	2139.93	858.87	1217.49
IMEzhou	54	827	619	0	40.85	1072.62	969.01	949.47	949.47	1072.93	987.11	1907.91	1550.05	1883.26	1848.41	843.06	1440.13
IMWar	202	975	471	148	0	1032.15	982.94	909.15	909.15	1032.46	969.67	1943.61	1517.97	1852.56	1815.62	802.29	1399.32
Yuan	1870	2643	1197	1816	1668	0	1734.24	316.23	316.23	0.78	967.25	2836.81	820.51	1143.88	1038.65	293.70	383.50
Liaoning	2200	2973	1527	2146	1998	330	0	1464.86	1464.86	1734.87	1951.86	2286.70	2417.52	2762.08	2698.66	1441.07	2003.71
Mongolia	2450	3223	1777	2396	2248	580	250	0	0.00	317.01	1145.38	2817.06	1132.14	1459.41	1354.79	137.21	543.86
MongPeriod	1850	2623	1177	1796	1648	20	350	600	0	317.01	1145.38	2817.06	1132.14	1459.41	1354.79	137.21	543.86
MongolTurk	1725	2498	1052	1671	1523	145	475	725	125	0	966.82	2836.80	819.73	1143.11	1037.88	294.35	383.44
Qinghai	605	1378	68	551	403	1265	1595	1845	1245	1120	0	2094.49	787.03	1043.59	1070.58	1016.63	1289.41
SChina	2450	3223	1777	2396	2248	580	250	0	600	725	1845	0	2876.72	3085.74	3143.97	2690.89	3218.98
XinjBronze	200	573	873	254	402	2070	2400	2650	2050	1925	805	2650	0	345.19	301.89	1069.29	904.55
XinjHan	451	1224	222	397	249	1419	1419	1999	1399	1274	154	1999	651	0	152.76	1405.85	1158.73
XinjIron	250	1023	423	196	48	1620	1620	2200	1600	1475	355	2200	450	201	0	1312.11	1021.52
Xiongnu	392	1165	281	338	190	1162	1808	2058	1458	1333	213	2058	592	59	59	0	608.04
Egiin Gol	392	1165	281	338	190	1162	1808	2058	1458	1333	213	2058	592	59	59	0	0

The upper triangle portion of the matrix contains spherical distances measured in kilometers; the lower triangle contains temporal distances.

Figure 7.21 shows the Ward's clustering and **Figure 7.22** shows the NJ tree for the Chinese comparative series. The clustering algorithm produced with Ward's method shows two clusters. The first, though with low supporting bootstrap values (32%), connects the Mongolian samples. Again, the Chandman, Egiin Gol, and Mongol Turk collective are strongly supported as is the modern Mongolian, Mongol period, and Xiongnu samples. The second cluster includes all of the Chinese samples, with a general distinction between samples from Inner Mongolia and Xinjiang. The NJ tree supports these results, though some Mongolian samples seem to be outgroups of the Chinese, specifically Qinghai and Liaoning (NE China).

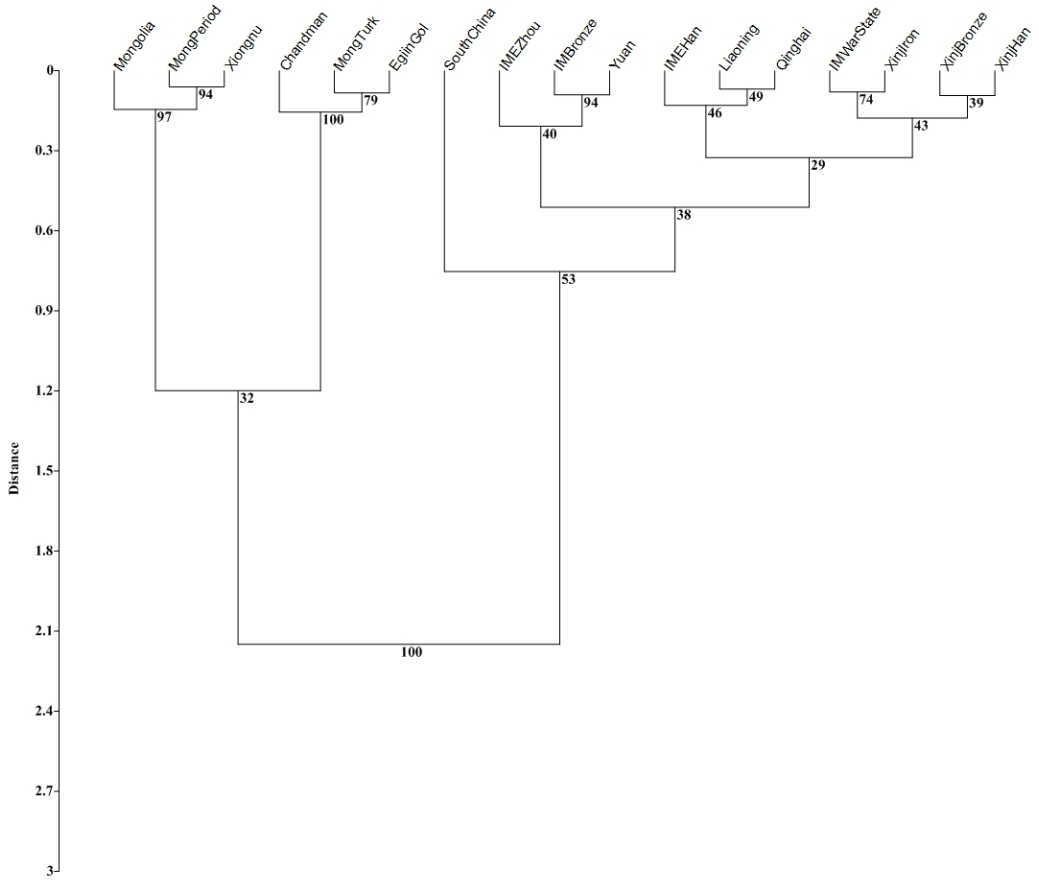

FIGURE 7.21. Ward's Hierarchical Clustering, Chinese Comparative Series with bootstrap values after 1000 replicates. R-matrix distances used to construct tree.

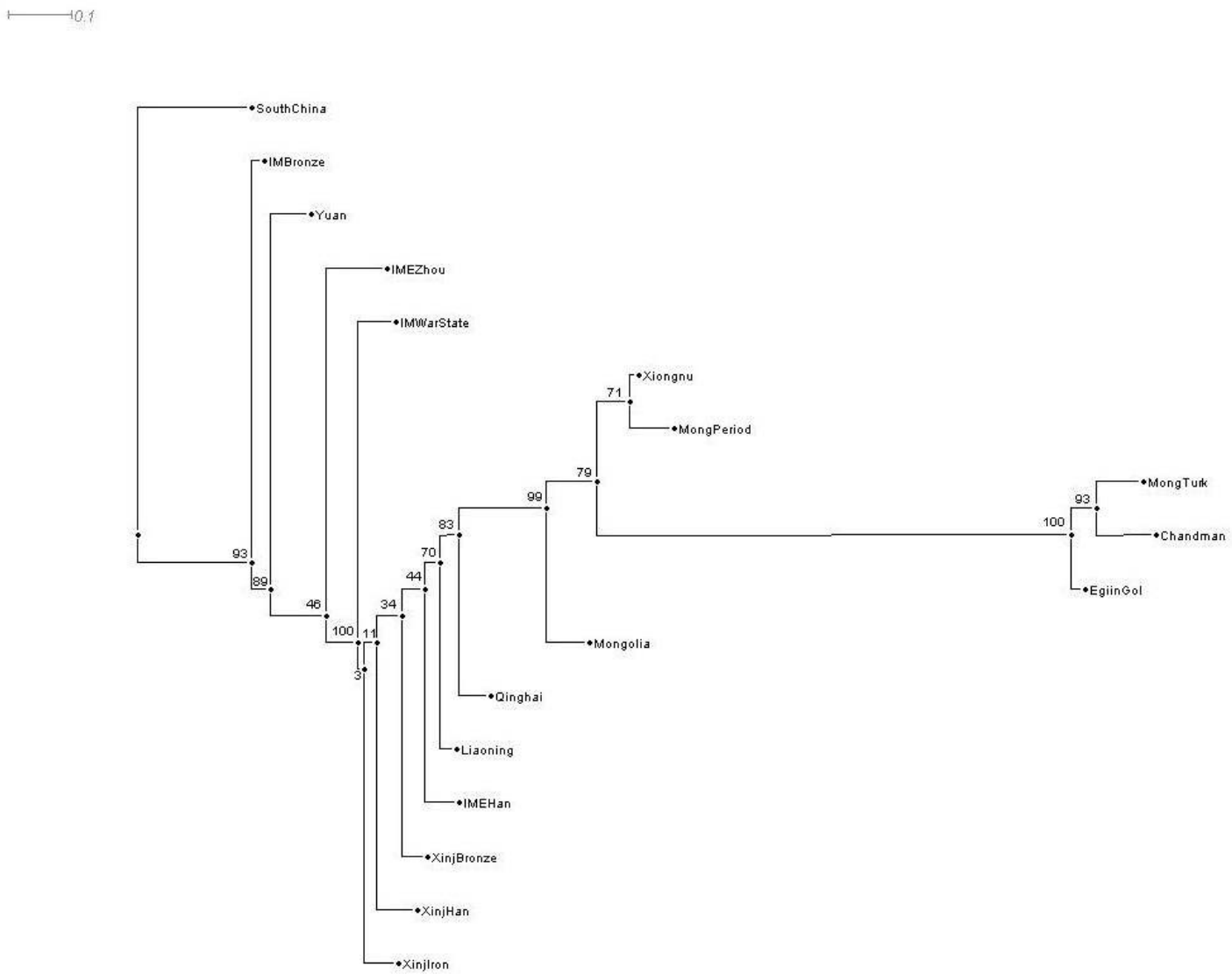

FIGURE 7.22. Neighbor-joining tree for Chinese comparative series using R-matrix distances.

Central Asian Comparative Series: **Table 7.16** shows samples and sample sizes used in the Central Asian comparative series. Again, this analyses was performed in order to assess variation between samples on a regional level.

TABLE 7.16. Samples included in Central Asian comparative series.

Population	N
Chandman	26
Chuvash	17
India	12
Kazakh	12
Kyrgyz	27
Mongolia	38
MongolPeriod	28
MongolTurk	8
Turkmen	17
Uighur	15
Uzbek	15
Xiongnu	33
EgiinGol	19
Total	267

Figure 7.23 shows the results from the principal coordinate analysis. Similar to the Chinese series analysis, the Chandman, Egiin Gol and Mongol Turk samples are outliers. In this analysis, the Xiongnu, Medieval Mongol period sample, and the modern Mongolian sample cluster in with most of the Central Asian samples. India is separated along EV2. It appears that the Kazakh sample is the closest related of all the Central Asian crania. The principal component plot (**Fig. 7.24**) has all of the Mongolian series as being outliers. When PC1 is plotted against PC3 (**Fig. 7.25**), the Xiongnu and Mongolian Medieval sample are clustered with the Turkmen and Chuvash samples. When PC2 is plotted against PC3 (**Fig. 7.26**), all of the Mongolian samples cluster together in the same quadrant.

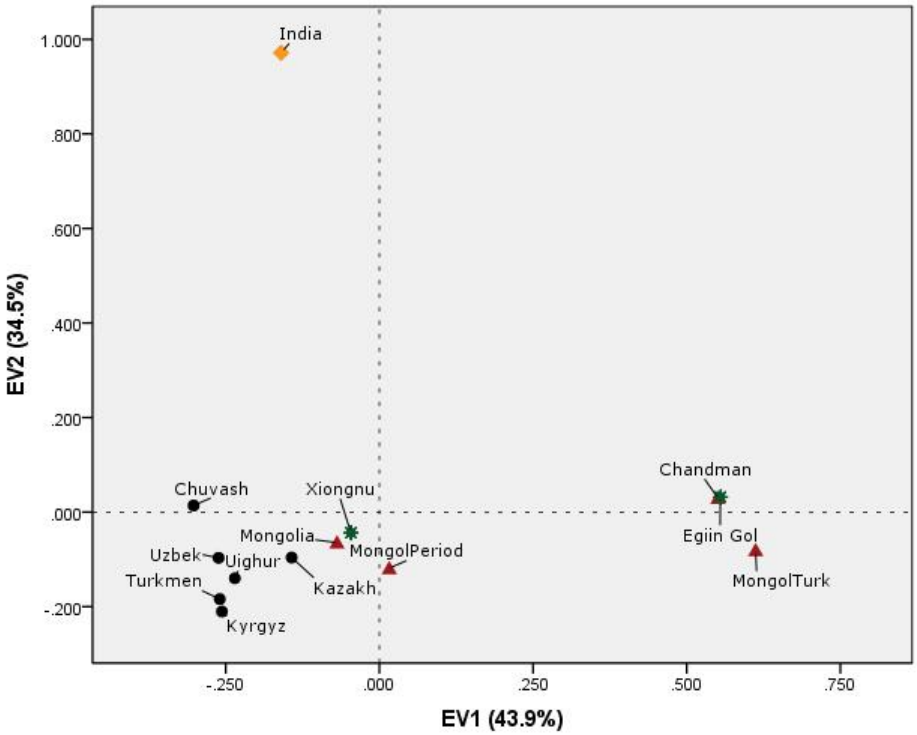

FIGURE 7.23. Principal coordinate plot, Central Asian series.

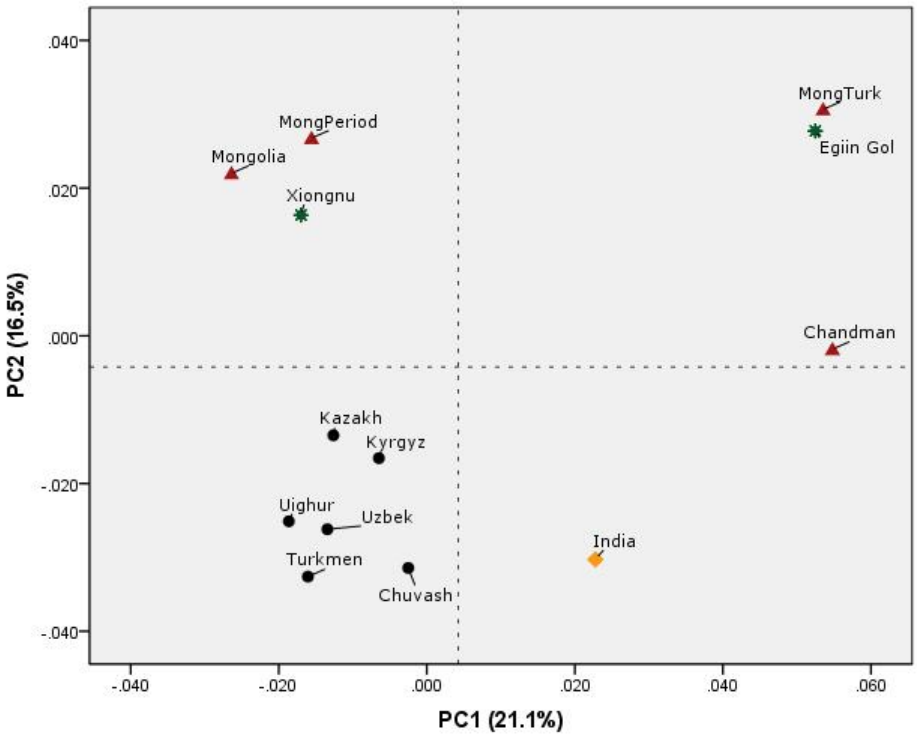

FIGURE 7.24. PC1 plotted against PC2, Central Asian series.

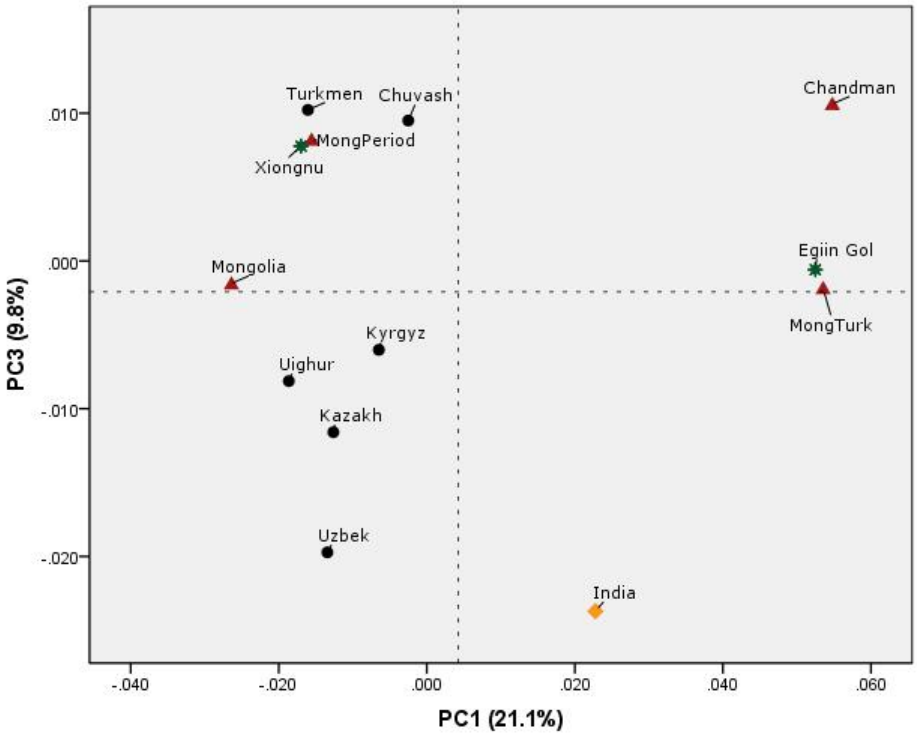

FIGURE 7.25. PC1 plotted against PC3, Central Asian series.

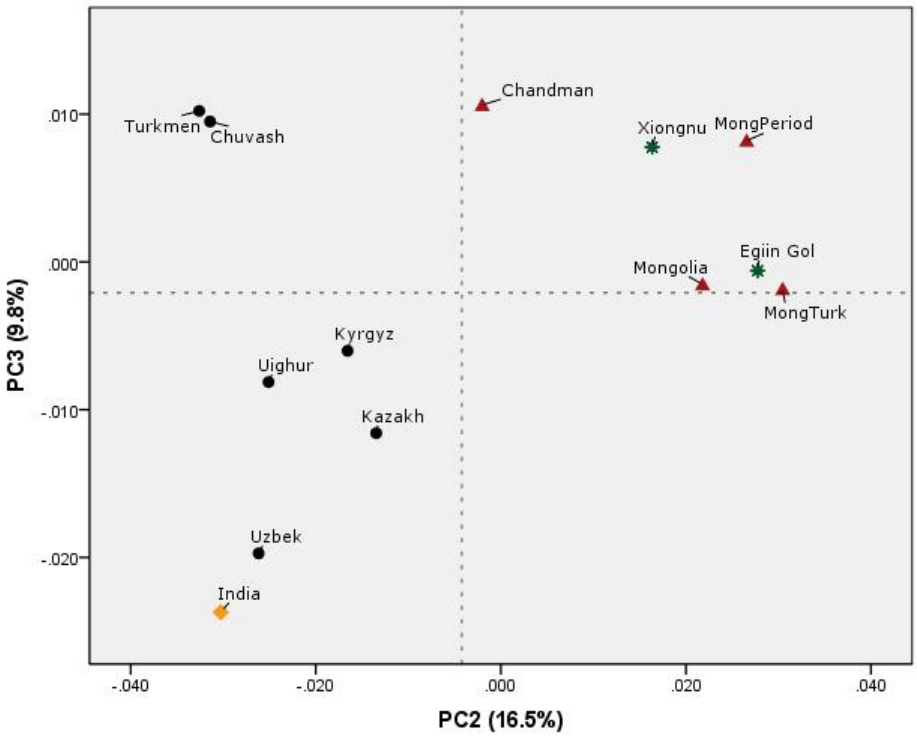

FIGURE 7.26. PC2 plotted against PC3, Central Asian series.

TABLE 7.17. Mahalanobis distances for Central Asian comparative series.

Population	Chandman	Chuvash	India	Kazakh	Kyrgyz	Mongolia	MongPer	MongTurk	Turkmen	Uighur	Uzbek	Xiongnu	Egiin Gol
Chandman	*0.00*												
Chuvash	0.864910	*0.00*											
India	1.482620	1.156173	*0.00*										
Kazakh	0.588091	0.334382	1.168628	*0.00*									
Kyrgyz	0.826139	0.295878	1.431984	0.132881	*0.00*								
Mongolia	0.674155	0.489642	1.174296	0.051608	0.238238	*0.00*							
MongolPeriod	0.517447	0.473033	1.311411	0.104585	0.237711	0.040566	*0.00*						
MongolTurk	0.158675	1.065998	1.723812	0.636144	0.798498	0.573243	0.492173	*0.00*					
Turkmen	0.766048	0.181767	1.388665	0.147297	0.037252	0.282877	0.306632	0.807184	*0.00*				
Uighur	0.674326	0.183707	1.260395	0.055769	0.055068	0.200230	0.216496	0.762947	0.018589	*0.00*			
Uzbek	0.746485	0.257010	1.162212	0.009586	0.050956	0.170334	0.226866	0.784119	0.061718	0.006994	*0.00*		
Xiongnu	0.548338	0.376887	1.107620	0.045154	0.218639	0.028744	0.027729	0.544654	0.254668	0.165335	0.156078	*0.00*	
Egiin Gol	0.064250	0.846364	1.402564	0.563036	0.744157	0.521664	0.435751	0.016055	0.737495	0.662807	0.713371	0.439975	*0.00*

Table 7.17 shows the Mahalanobis distances between the Central Asian comparative cranial series. These results are similar to the PC plots, however, the third closest sample to the Xiongnu are Kazakh. There is a shared history between modern Mongolians and the peoples of Kazakhstan, and it appears that the Xiongnu nomads were possibly linked to the Kazakh people as well.

Table 7.18 shows the results from the Relethford-Blangero analysis. Here, the Xiongnu lie between the Kazakh and Uighur samples for r_{ii}, or distance to the centroid. They have a negative residual variance, while the Egiin Gol sample retains a positive residual variance. The F_{ST} value is also higher than compared to the Chinese analysis, indicating greater biological divergence among samples of Mongolian and Central Asia.

TABLE 7.18. Relethford-Blangero results for Central Asian comparative series.

		Within-Group Phenotypic Variance			
Population	r_{ii}	Observed	Expected	Residual	SE
Kazakh	0.05145	0.793	1.088	-0.295	0.020161
Xiongnu	0.06143	0.860	1.077	-0.217	0.011025
Uighur	0.08547	0.820	1.049	-0.229	0.020368
Uzbek	0.09186	0.923	1.042	-0.119	0.020909
MongolPeriod	0.09767	0.913	1.035	-0.122	0.014701
Mongolia	0.10264	0.848	1.030	-0.181	0.012634
Turkmen	0.14203	0.856	0.984	-0.128	0.022984
Kyrgyz	0.14965	0.811	0.976	-0.165	0.018063
Chuvash	0.26015	0.657	0.849	-0.192	0.029870
EgiinGol	0.30846	0.894	0.793	0.101	0.030380
Chandman	0.36831	1.312	0.725	0.587	0.027942
MongolTurk	0.39643	0.861	0.692	0.169	0.054817
India	0.96939	0.828	0.035	0.793	0.066434

F_{ST} = 0.237303
h^2 = 1.0
V_{GW} = 0.875

Table 7.19 shows the temporal and geographic distance matrices constructed to test correspondence between biological, spatial, and temporal distances.

TABLE 7.19. Temporal and geographic distances for Central Asian comparative series.

Population	Chandman	Chuvash	India	Kazakh	Kyrgyz	Mongolia	MongPer	MongTurk	Turkmen	Uighur	Uzbek	Xiongnu	Egiin Gol
Chandman	0	3041.63	3029.21	1837.91	1657.02	1109.40	1109.40	852.52	2827.12	767.62	2323.13	1138.27	591.91
Chuvash	2450	0	4928.03	1598.73	2570.67	4085.47	4085.47	3879.84	2063.38	3150.28	2028.04	4147.24	3543.38
India	2450	0	0	3347.26	2367.83	3249.61	3249.61	3008.36	3212.75	2316.31	2981.34	3130.48	3219.94
Kazakh	2450	0	0	0	979.69	2945.98	2945.98	2671.48	1168.76	1665.57	762.12	2964.39	2425.03
Kyrgyz	2450	0	0	0	0	2635.05	2635.05	2322.19	1317.28	1093.00	851.30	2606.66	2210.42
Mongolia	2450	0	0	0	0	0	0.00	317.01	3889.06	1558.90	3385.20	137.21	543.86
MongolPeriod	1850	600	600	600	600	600	0	317.01	3889.06	1558.90	3385.20	137.21	543.86
MongolTurk	1725	725	725	725	725	725	125	0	3585.71	1242.52	3082.87	294.35	383.44
Turkmen	2450	0	0	0	0	0	600	725	0	2393.10	504.68	3877.33	3414.57
Uighur	2450	0	0	0	0	0	600	725	0	0	1901.40	1520.08	1197.55
Uzbek	2450	0	0	0	0	0	600	725	0	0	0	3375.02	2910.05
Xiongnu	392	2058	2058	2058	2058	2058	1458	1333	2058	2058	2058	0	608.04
Egiin Gol	392	2058	2058	2058	2058	2058	1458	1333	2058	2058	2058	0	0

The upper triangle portion of the matrix contains spherical distances measured in kilometers; the lower triangle contains temporal distances.

Figure 7.27 shows the results from applying R-matrix distances to a clustering algorithm using Ward's method, and **Figure 7.28** shows the results from the NJ tree using R-matrix distances for the Central Asian cranial series. Both trees result in a consensus where the Chandman, Egiin Gol and Mongol Turk are distant outliers to India, while the Xiongnu, modern Mongolian, and Mongol period show a more distant relationship to other Central Asian groups.

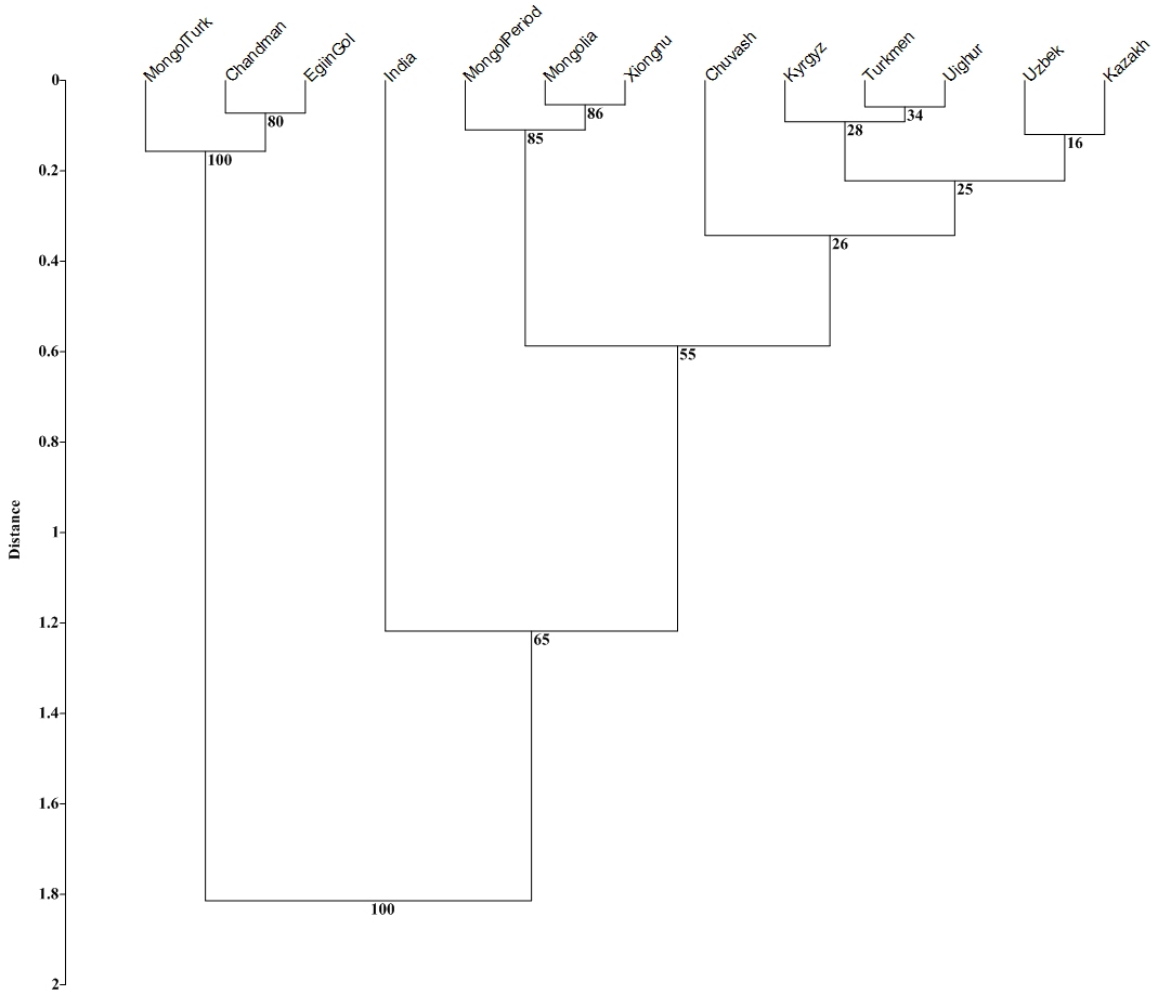

FIGURE 7.27. Ward's Hierarchical Clustering Tree for Central Asian comparative series with bootstrap values after 1000 replicates. R-matrix distances used to construct tree.

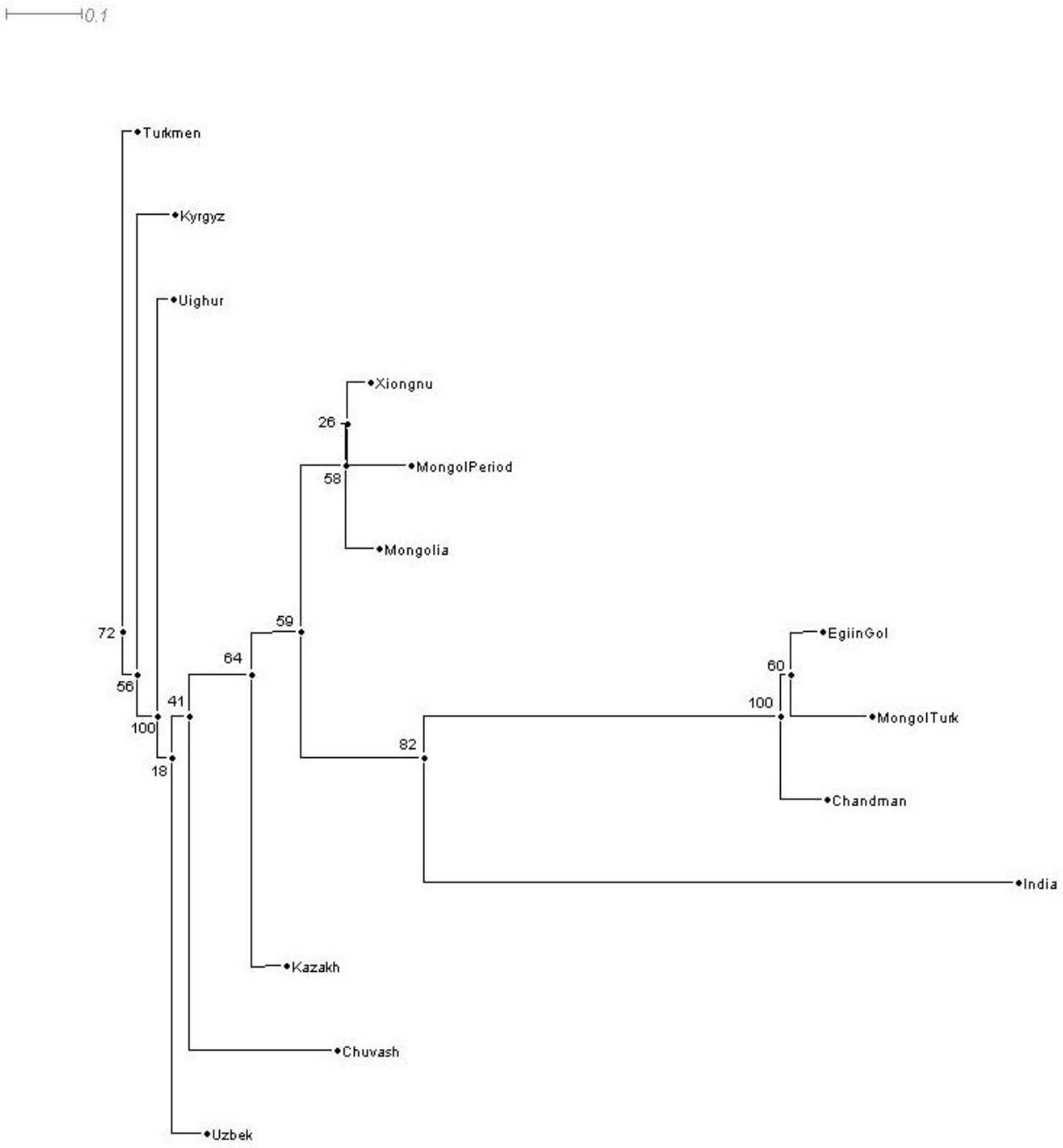

FIGURE 7.28. Neighbor-joining tree for Central Asian comparative series. R-matrix distances used to construct tree.

Siberian Comparative Series: **Table 7.20** shows the samples and sample sizes used in the Siberian cranial comparative series.

TABLE 7.20. Samples used in Siberian comparative analysis.

Population	N
Buryat	19
Chandman	26
Chuckchi	6
Evenks	17
EarlyIronSiberia	30
IronAgeTuva	19
Kalmyk	33
Mongolia	38
MongolPeriod	28
MongolTurk	8
Oruchi	17
Pazyryk	46
SiberianBronze	10
Tagar	18
Tuva	36
Ulchi	15
MedievalSiberia	47
Xiongnu	33
EgiinGol	19
Yakut	31
Total	496

Figure 7.29 shows results from the principal coordinate analysis. Once again, the Chandman, Mongol Turk, and Egiin Gol series are severe outliers in comparison to the other groups. The first two eigenvectors account for 71.6% of the variation. The Medieval period Mongol sample and the Xiongnu are once again clustered in close proximity, however, the modern Mongolian sample is close to the Yakuts of northeast Siberia, the Early Iron Age Siberian, the Iron Age Tuvan, and the Pazyryk sample from southern and western Siberia. The principal components analysis (**Fig. 7.30**) is slightly different, though accounts for only 32.1% of the variation on the first two PCs. In this plot, the Mongolian samples are all isolated.

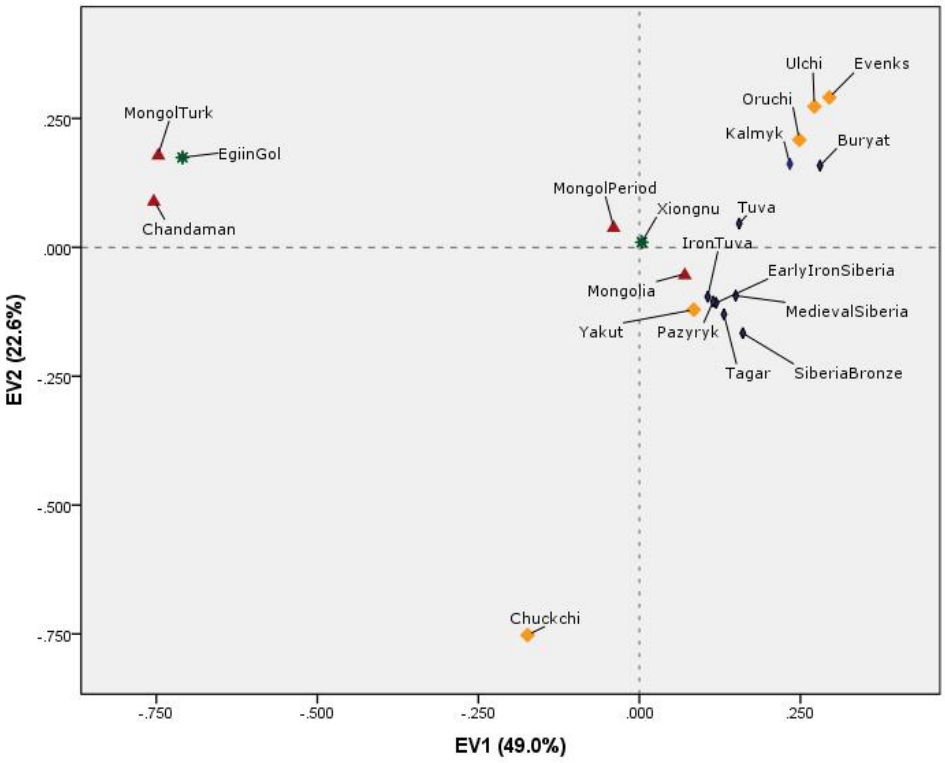

FIGURE 7.29. Principal coordinate plot, Siberian cranial comparative series.

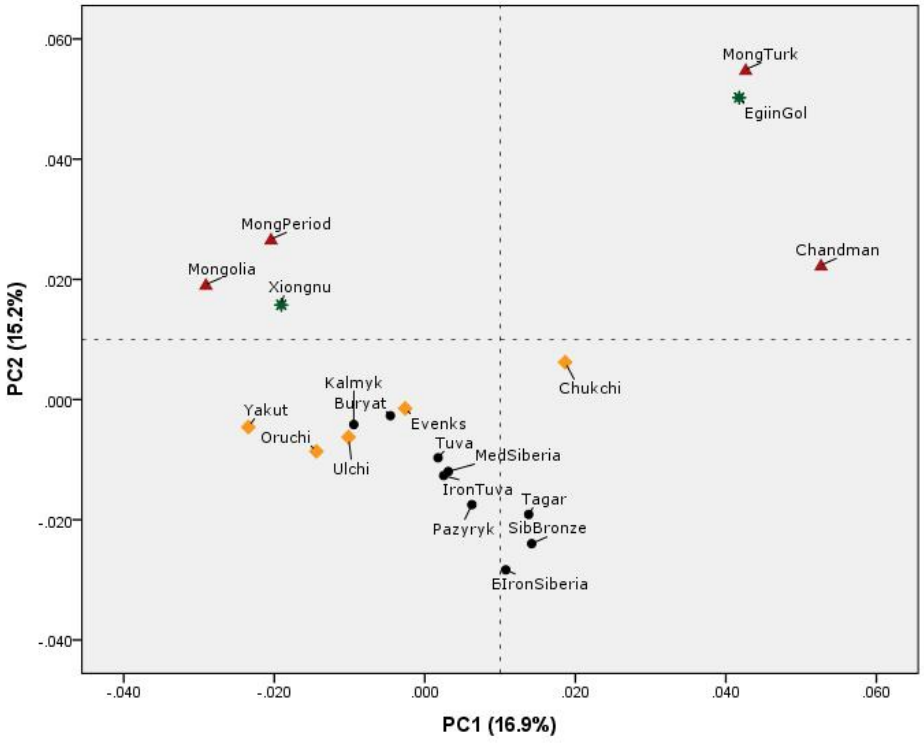

FIGURE 7.30. PC1 plotted against PC2, Siberian cranial comparative series.

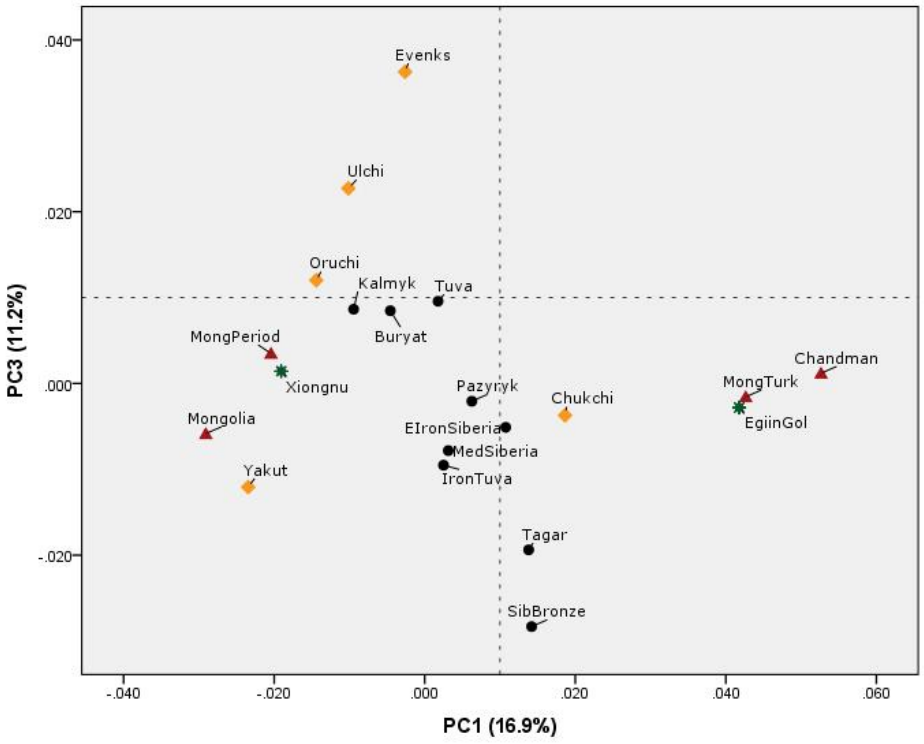

FIGURE 7.31. PC1 plotted against PC3, Siberian cranial comparative series.

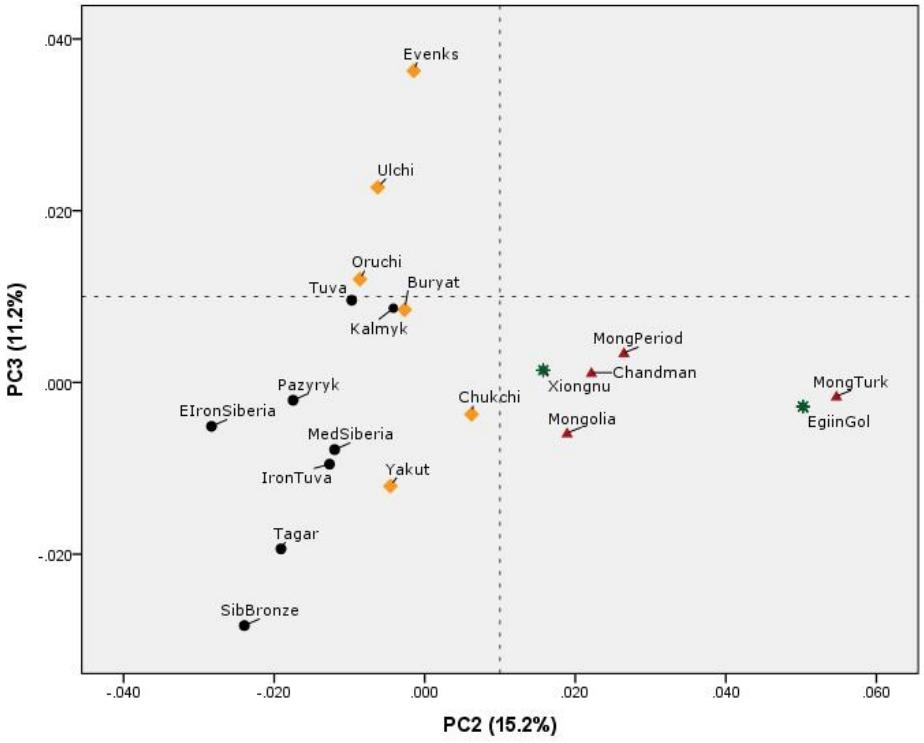

FIGURE 7.32. PC2 plotted against PC3, Siberian cranial comparative series.

When PC 1 is plotted against PC3 (**Fig. 7.31**), the Mongol Period, Xiongnu, and modern Mongolian sample become less isolated with the modern Mongolian sample clustering close to the Yakut sample. When PC2 is plotted against PC3 (**Fig. 7.32**), PC2 segregates all of the Mongolian samples into the same quadrant.

Table 7.21 shows the results from the Relethford-Blangero analysis (h^2=1.0). Here the Xiongnu fall between the Tuvan sample from southern Siberia and the Late Bronze Age/Early Iron Age Pazyryk sample from western Siberia for distance from the centroid (R_{ii}). F_{ST} is similar to other analyses with a value of 0.21, indicating moderate biological divergence between groups. Once again, the Egiin Gol, Chandman, and Mongol Turk display greater than expected extralocal gene flow compared to the other groups in the analysis, which have negative residuals.

TABLE 7.21. Relethford-Blangero analysis of Siberian cranial series.

		Within-Group Phenotypic Variance			
Population	r_{ii}	Observed	Expected	Residual	SE
IronTuva	0.03035	0.810	1.098	-0.288	0.01228
MedievalSiberia	0.05315	0.767	1.072	-0.305	0.00828
Pazyryk	0.06795	0.761	1.055	-0.294	0.00931
Xiongnu	0.07187	0.979	1.051	-0.071	0.01155
Tuva	0.07922	0.848	1.042	-0.194	0.01143
EarlyIronSiberia	0.07966	0.802	1.042	-0.240	0.01274
Mongolia	0.08900	0.925	1.031	-0.106	0.01166
Kalmyk	0.09171	0.746	1.028	-0.282	0.01279
MongolPeriod	0.10111	1.055	1.018	0.037	0.01465
Tagar	0.11106	0.745	1.006	-0.261	0.01975
Yakut	0.12428	1.027	0.991	0.035	0.01513
Oruchi	0.13872	0.857	0.975	-0.118	0.02236
Buryat	0.15851	0.769	0.953	-0.183	0.02217
SiberiaBronze	0.16342	0.807	0.947	-0.140	0.03284
Ulchi	0.17309	0.851	0.936	-0.085	0.02637
Evenks	0.24274	0.654	0.857	-0.203	0.02845
EgiinGol	0.53282	1.014	0.529	0.485	0.03857
MongolTurk	0.60641	0.959	0.446	0.513	0.06501
Chandman	0.62938	1.444	0.420	1.024	0.03551
Chuckchi	0.74912	0.960	0.284	0.676	0.08374

F_{ST} = 0.214678
h^2 = 1.0
V_{GW} = 0.889

TABLE 7.22. Mahalanobis distances for Siberian comparative series.

Population	Buryat	Chandman	Chuckchi	Evenks	EISiberia	IronTuva	Kalmyk	Mongolia	MongPer	MongTurk	Oruchi	Pazyryk	SibBrz	Tagar	Tuva	Ulchi	MedSib	Xiongnu	EgiinGol	Yakut
Buryat	*0.00*																			
Chandman	1.207929	*0.00*																		
Chuckchi	1.136789	1.357254	*0.00*																	
Evenks	0.121302	1.312743	1.350063	*0.00*																
EarlyIronSiberia	0.228358	0.811282	0.833124	0.381152	*0.00*															
IronTuva	0.213477	0.804623	0.791765	0.331079	0.008339	*0.00*														
Kalmyk	0.023263	1.052640	1.124908	0.077174	0.173270	0.125545	*0.00*													
Mongolia	0.276154	0.921752	0.793143	0.337539	0.245108	0.116386	0.172734	*0.00*												
MongolPeriod	0.335061	0.722524	0.938842	0.356986	0.283756	0.141280	0.206543	0.049909	*0.00*											
MongolTurk	1.085806	0.140938	1.335701	1.204909	0.935891	0.849223	0.999262	0.809864	0.651554	*0.00*										
Oruchi	0.033065	1.130227	1.180529	0.066700	0.223521	0.211166	0.043626	0.235671	0.314452	1.015912	*0.00*									
Pazyryk	0.271706	0.829647	0.852433	0.371400	0.033407	0.000000	0.175478	0.133388	0.163250	0.919579	0.261730	*0.00*								
SiberiaBronze	0.289309	0.999631	0.896170	0.543567	0.046213	0.051212	0.244259	0.320438	0.410715	1.050658	0.316271	0.105036	*0.00*							
Tagar	0.264201	0.871335	0.876486	0.434084	0.025663	0.023182	0.212184	0.248467	0.314954	0.999648	0.299240	0.061672	0.051498	*0.00*						
Tuva	0.109941	0.907313	0.954257	0.199385	0.113685	0.087756	0.100454	0.183781	0.161929	0.910741	0.149136	0.106831	0.243328	0.160391	*0.00*					
Ulchi	0.077614	1.198389	1.330821	0.030401	0.273183	0.218488	0.046105	0.271890	0.342438	1.078558	0.000000	0.279921	0.377046	0.321766	0.170275	*0.00*				
MedievalSiberia	0.164286	0.933288	0.746965	0.283573	0.045819	0.001537	0.122619	0.134890	0.179425	0.897380	0.161151	0.051703	0.099727	0.091396	0.087212	0.203758	*0.00*			
Xiongnu	0.314988	0.743887	0.906934	0.324328	0.209454	0.085749	0.162351	0.037892	0.022052	0.715021	0.270091	0.102244	0.321478	0.220824	0.176178	0.281061	0.144268	*0.00*		
EgiinGol	1.024305	0.079651	1.238494	1.042723	0.839699	0.765368	0.885271	0.753038	0.593229	0.000038	0.937133	0.836321	1.011794	0.890324	0.840650	0.963177	0.836338	0.620662	*0.00*	
Yakut	0.338839	0.894525	0.797989	0.438024	0.209241	0.130698	0.210438	0.057809	0.162541	0.946111	0.277516	0.122124	0.283596	0.203009	0.242439	0.357604	0.192591	0.101852	0.844399	*0.00*

TABLE 7.23. R matrix distances for Siberian comparative series.

Population	Buryat	Chandman	Chuckchi	Evenks	EISiberia	IronTuva	Kalmyk	Mongolia	MongPer	MongTurk	Oruchi	Pazyryk	SibBrz	Tagar	Tuva	Ulchi	MedSiberia	Xiongnu	EgiinGol	Yakut
Buryat	0.158510																			
Chandman	-0.210021	0.629378																		
Chuckchi	-0.114579	0.010623	0.749122																	
Evenks	0.139973	-0.220314	-0.179102	0.242737																
EarlyIronSib	0.004908	-0.051121	-0.002170	-0.029376	0.079663															
IronTuva	-0.012308	-0.072447	-0.006146	-0.028995	0.050838	0.030351														
Kalmyk	0.113480	-0.165775	-0.142037	0.128638	-0.000947	-0.001740	0.091713													
Mongolia	-0.014323	-0.101688	0.022489	-0.002902	-0.038223	0.001482	0.003989	0.088999												
MongolPer	-0.037721	0.003981	-0.044306	-0.006571	-0.051492	-0.004910	-0.006861	0.070099	0.101108											
MongolTurk	-0.160443	0.547425	0.009916	-0.177880	-0.124908	-0.106230	-0.150569	-0.057227	0.027983	0.606411										
Oruchi	0.132080	-0.181066	-0.146345	0.157377	-0.002571	-0.021049	0.093401	-0.003978	-0.037314	-0.135392	0.138717									
Pazyryk	-0.022625	-0.066162	-0.017683	-0.030359	0.057101	0.050976	-0.007910	0.011778	0.002902	-0.122611	-0.027534	0.067945								
SibBronze	0.016310	-0.103417	0.008186	-0.068705	0.098435	0.071279	0.005436	-0.034010	-0.073094	-0.140414	-0.007068	0.063164	0.163419							
Tagar	0.002685	-0.065449	-0.008152	-0.040144	0.082530	0.059115	-0.004706	-0.024204	-0.051393	-0.141088	-0.024731	0.058667	0.111491	0.111060						
Tuva	0.063893	-0.099359	-0.062959	0.061285	0.022598	0.010906	0.035238	-0.007783	0.009198	-0.112556	0.034399	0.020166	-0.000346	0.014943	0.079217					
Ulchi	0.126994	-0.197960	-0.204304	0.192714	-0.010213	-0.007523	0.109350	-0.004899	-0.034119	-0.149527	0.156113	-0.019441	-0.020267	-0.018807	0.041017	0.173092				
MedievalSiberia	0.023689	-0.125378	0.027655	0.006159	0.043499	0.040984	0.011124	0.003631	-0.012581	-0.118907	0.015360	0.034698	0.058423	0.036409	0.022580	0.011244	0.053154			
Xiongnu	-0.042303	-0.021318	-0.042970	-0.004859	-0.028959	0.008237	0.000617	0.061489	0.075464	-0.018369	-0.029751	0.018787	-0.043093	-0.018946	-0.012544	-0.018048	-0.009621	0.071872		
EgiinGol	-0.166487	0.541274	0.021724	-0.133583	-0.113608	-0.101098	-0.130368	-0.065609	0.020350	0.569597	-0.132798	-0.117777	-0.157777	-0.123221	-0.114306	-0.128632	-0.125182	-0.007985	0.532821	
Yakut	-0.028027	-0.070436	0.037704	-0.035505	-0.002651	0.011965	0.002775	0.077733	0.031422	-0.107712	-0.007262	0.035049	0.002050	0.016163	-0.019473	-0.030118	-0.007580	0.047148	-0.093651	0.124276

Table 7.22 shows the Mahalanobis distances for the groups in the Siberian comparative cranial series. The Xiongnu sample is closely related to the Medieval period Mongolians, modern Mongolians, followed by the Iron Age Tuvans, the Yakut and the Pazyryk sample. The Egiin Gol sample is closest to the Chandman, Mongol Turk, Medieval Period, and Xiongnu samples. **Table 7.23** shows the R-matrix distances, which are similar to the Mahalanobis distances. Here, the Xiongnu exhibit positive values (biological similarity) also with the Kalmyk Mongol sample. The Egiin Gol sample shows some similarity for R-matrix values with the Chuckchi. **Table 7.24** shows the temporal and geographic distances for the Siberian comparative series.

TABLE 7.24. Temporal and geographic distances for Siberian comparative series.

Population	Buryat	Chandman	Chuckchi	Evenks	EISiberia	IronTuva	Kalmyk	Mongolia	MongPer	MongTurk	Oruchi	Pazyryk	SibBrz	Tagar	Tuva	Ulchi	MedSiberia	Xiongnu	EgiinGol	Yakut
Buryat	0	1490.97	3309.58	1360.66	1781.42	1178.73	4661.52	795.54	795.54	1037.02	1656.44	1842.33	1781.42	1486.18	1178.73	1847.30	1781.42	925.17	955.04	1543.80
Chandman	2450	0	4477.93	769.02	859.14	328.03	3392.89	1109.40	1109.40	852.52	3090.92	418.89	859.14	357.40	328.03	3335.41	859.14	1138.27	591.91	2588.70
Chuckchi	0	2450	0	3848.15	4135.39	4165.24	6338.08	4104.74	4104.74	4331.51	3020.61	4635.53	4135.39	4253.47	4165.24	2506.09	4135.39	4231.94	4141.53	1889.31
Evenks	0	2450	0	0	424.45	609.58	3304.99	1423.92	1423.92	1312.50	3006.54	791.81	424.45	439.94	609.58	3123.59	424.45	1518.02	929.73	1984.55
EarlyIronSib	2350	100	2350	2350	0	860.05	2881.40	1762.02	1762.02	1596.03	3430.38	656.04	0.00	516.21	860.05	3545.21	0.00	1838.27	1230.65	2313.66
IronTuva	2350	100	2350	2350	0	0	3599.44	919.98	919.98	736.18	2803.78	669.92	860.05	377.16	0.00	3026.02	860.05	983.82	377.24	2276.41
Kalmyk	0	2450	0	0	2350	2350	0	4498.54	4498.54	4242.02	6311.52	2974.01	2881.40	3230.39	3599.44	6399.39	2881.40	4530.85	3961.74	4873.33
Mongolia	0	2450	0	0	2350	2350	0	0		317.01	2080.69	1526.96	1762.02	1296.94	919.98	2420.07	1762.02	137.21	543.86	2320.03
MongolPer	600	1850	600	600	1750	1750	600	600	0	317.01	2080.69	1526.96	1762.02	1296.94	919.98	2420.07	1762.02	137.21	543.86	2320.03
MongolTurk	725	1725	725	725	1625	1625	725	725	125	0	2397.66	1270.03	1596.03	1097.72	736.18	2732.76	1596.03	294.35	383.44	2503.52
Oruchi	0	2450	0	0	2350	2350	0	0	600	725	0	3473.68	3430.38	3135.50	2803.78	540.77	3430.38	2133.78	2506.25	2130.53
Pazyryk	2350	100	2350	2350	0	0	2350	2350	1750	1625	2350	0	656.04	394.37	669.92	3688.09	656.04	1557.05	999.70	2761.10
SibBronze	2500	50	2500	2500	150	150	2500	2500	1900	1775	2500	150	0	516.21	860.05	3545.21	0.00	1838.27	1230.65	2313.66
Tagar	2400	50	2400	2400	50	50	2400	2400	1800	1675	2400	50	100	0	377.16	3322.63	516.21	1357.20	753.50	2373.44
Tuva	0	2450	0	0	2350	2350	0	0	600	725	0	2350	2500	2400	0	3026.02	860.05	983.82	377.24	2276.41
Ulchi	0	2450	0	0	2350	2350	0	0	600	725	0	2350	2500	2400	0	0	3545.21	2495.53	2774.70	1877.59
MedievalSiberia	600	1850	600	600	1750	1750	600	600	0	125	600	1750	1900	1800	600	600	0	1838.27	1230.65	2313.66
Xiongnu	2058	392	2058	2058	292	292	2058	2058	1458	1333	2058	292	442	342	2058	2058	1458	0	608.04	2455.13
EgiinGol	2058	392	2058	2058	292	292	2058	2058	1458	1333	2058	292	442	342	2058	2058	1458	0	0	2273.52
Yakut	0	2450	0	0	2350	2350	0	0	600	725	0	2350	2500	2400	0	0	600	2058	2058	0

The upper triangle portion of the matrix contains spherical distances measured in kilometers; the lower triangle contains temporal distances.

Figure 7.33 shows the results from the hierarchical clustering algorithm (Ward's method) using R-matrix distances, and **Figure 7.34** shows the NJ tree resulting from R-matrix distances. As usual, Chandman, Mongol Turk, and Egiin Gol are clustered together with high bootstrap values. In this case, the Bronze age cluster is an outgroup to the extreme NE Siberian Chuckchi group. The modern Mongol, Xiongnu, and Mongol period samples are closer to Bronze and Iron Age western and southern Siberian groups than they are with the NE groups, with the exception of the modern Mongolian sample clustering with the Yakut sample, though, like the global analysis, there is low support value at that node.

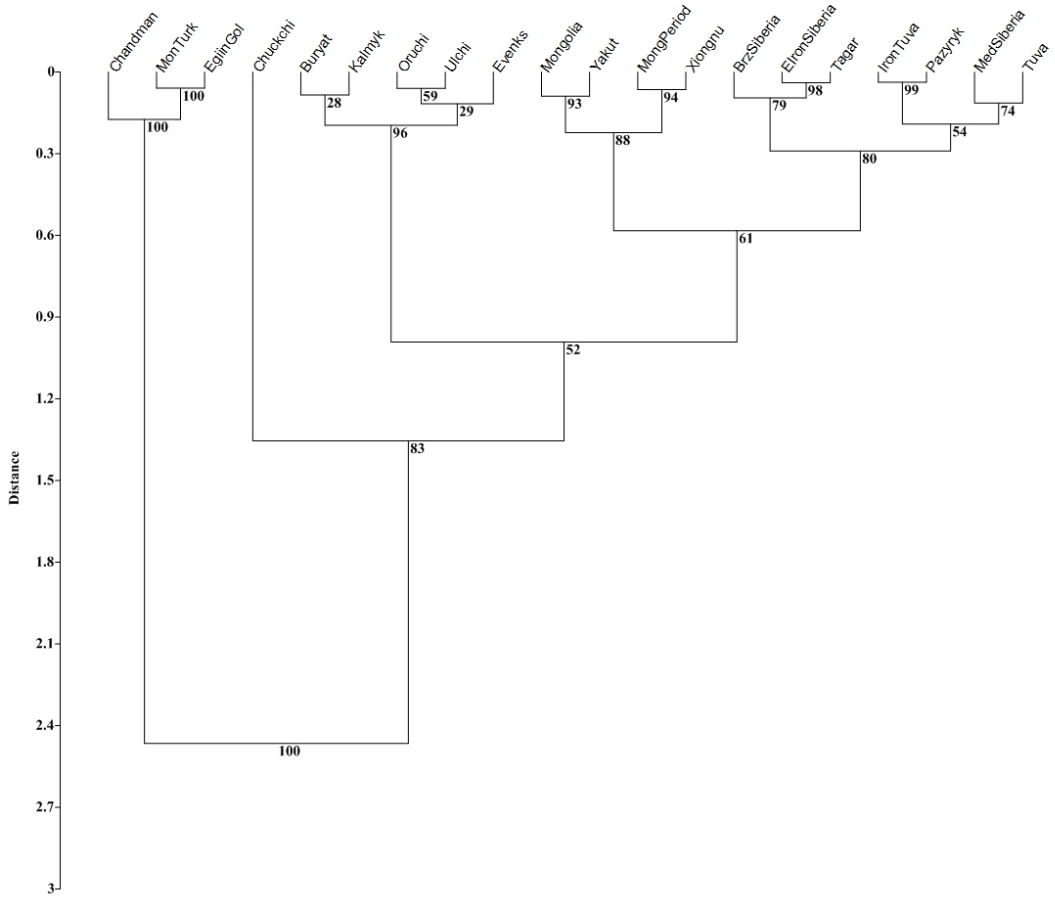

FIGURE 7.33. Ward's Hierarchical Clustering, Siberian comparative series with bootstrap values after 1000 replicates. R-matrix distances used to construct tree.

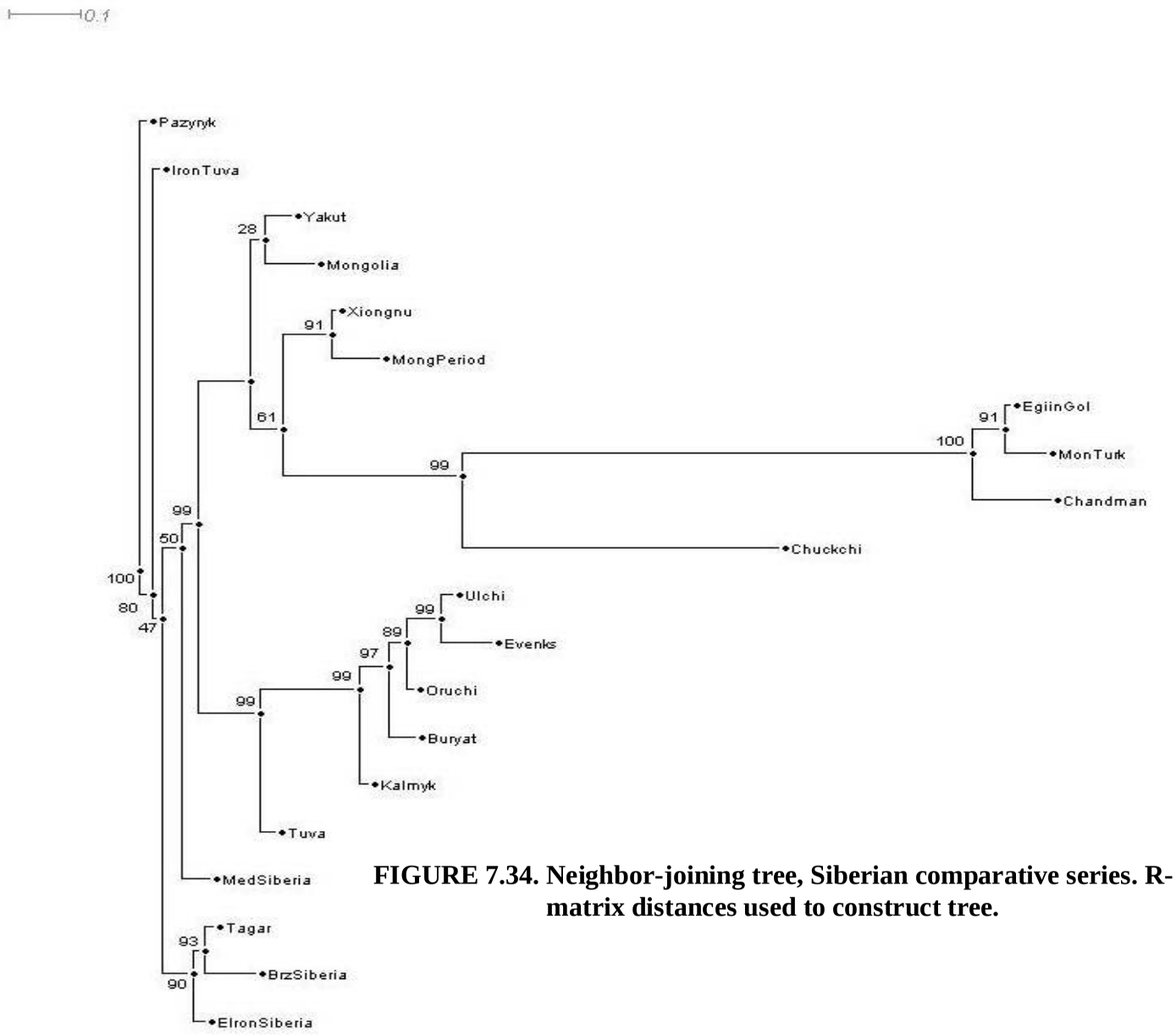

FIGURE 7.34. Neighbor-joining tree, Siberian comparative series. R-matrix distances used to construct tree.

Manly Regression Tests: Manly randomization tests were performed in order to account for possible principal components (shape variables) that are correlated with either median latitude, longitude, or time period. **Table 7.25** shows the significance values from these tests. 1000 randomizations were performed on the Y (independent) variables – latitude, longitude, and time. Those PC scores that accounted for 95% of the variation were used as dependent variables in the analysis. This amounted to using the first 30 PCs for each level of analysis. Significance was assessed using a *t*-test of the absolute value of the regression coefficient at $\alpha = .05$.

The global series, using a total of 50 groups, yielded several PCs with significant values for latitude. These include PCs 6, 14, 15, 27, 29. Interestingly, several more PCs were significant for longitude. These include PCs 4, 6, 7, 9, 11, 15, 21, 22 and 30. For time period, PCs 1, 5, 9 and 14 were significant at the $\alpha = .05$ level. When the test was performed using only the Mongolian and Chinese samples (n = 17), fewer PC scores were significant. These included PC10 for latitude, PCs 8, 15, 16, 19 and 25 for longitude, and only PC21 for time. For the Central Asian comparative series (n = 13), there were no PC scores that were significant for either latitude or longitude, however, PC21 was also significant for time. PCs 16 and 19 were significant for latitude in the Siberian cranial comparative series, while PC 15 was significant for longitude. Temporal period had more significant PC scores for the Siberian series, with PCs 3, 5, and 14 significant at $\alpha = .05$. Those significant PC scores will be discussed further in the discussion chapter in the context of localized craniofacial shape changes.

TABLE 7.25. Manly Tests for PC correlation with latitude, longitude, and time. Values shown are exact p values using 1000 randomizations.

Latitude	PC1	PC2	PC3	PC4	PC5	PC6	PC7	PC8	PC9	PC10
Global	0.207	0.345	0.254	0.922	0.105	0.006 *	0.979	0.183	0.673	0.866
Chinese	0.603	0.340	0.065	0.354	0.143	0.665	0.068	0.095	0.509	0.007 *
Central Asian	0.631	0.774	0.331	0.622	0.639	0.360	0.859	0.784	0.899	0.440
Siberian	0.804	0.287	0.517	0.981	0.526	0.478	0.468	0.632	0.556	0.183
	PC11	PC12	PC13	PC14	PC15	PC16	PC17	PC18	PC19	PC20
Global	0.675	0.339	0.802	0.019 *	0.015 *	0.773	0.530	0.431	0.776	0.913
Chinese	0.933	0.152	0.071	0.988	0.998	0.634	0.913	0.510	0.268	0.414
Central Asian	0.973	0.269	0.466	0.476	0.635	0.484	0.786	0.746	0.388	0.995
Siberian	0.207	0.480	0.846	0.843	0.719	0.041 *	0.999	0.138	0.019 *	0.305
	PC21	PC22	PC23	PC24	PC25	PC26	PC27	PC28	PC29	PC30
Global	0.976	0.522	0.952	0.244	0.553	0.891	0.001 *	0.257	0.038 *	0.397
Chinese	0.783	0.734	0.688	0.599	0.087	0.677	0.572	0.468	0.803	0.677
Central Asian	0.328	0.872	0.609	0.722	0.993	0.319	0.558	0.076	—	—
Siberian	0.476	0.944	0.240	0.681	0.163	0.930	0.818	0.610	0.661	0.886
Longitude	**PC1**	**PC2**	**PC3**	**PC4**	**PC5**	**PC6**	**PC7**	**PC8**	**PC9**	**PC10**
Global	0.287	0.220	0.779	0.001 *	0.753	0.001 *	0.001 *	0.636	0.039	0.886
Chinese	0.584	0.122	0.568	0.297	0.129	0.764	0.853	0.016 *	0.196	0.788
Central Asian	0.333	0.207	0.285	0.595	0.931	0.755	0.505	0.391	0.971	0.409
Siberian	0.993	0.740	0.593	0.964	0.617	0.892	0.937	0.978	0.937	0.157
	PC11	PC12	PC13	PC14	PC15	PC16	PC17	PC18	PC19	PC20
Global	0.012 *	0.511	0.900	0.549	0.017 *	0.601	0.226	0.849	0.742	0.332
Chinese	0.077	0.551	0.703	0.715	0.005 *	0.015 *	0.082	0.663	0.052 *	0.739
Central Asian	0.710	0.384	0.804	0.932	0.552	0.579	0.855	0.846	0.710	0.632
Siberian	0.070	0.171	0.912	0.575	0.045 *	0.151	0.098	0.475	0.864	0.274
	PC21	PC22	PC23	PC24	PC25	PC26	PC27	PC28	PC29	PC30
Global	0.003 *	0.051 *	0.367	0.056	0.423	0.475	0.629	0.797	0.673	0.001 *
Chinese	0.098	0.587	0.690	0.762	0.041 *	0.402	0.247	0.078	0.715	0.130
Central Asian	0.262	0.194	0.065	0.222	0.550	0.138	0.577	0.971	—	—
Siberian	0.715	0.128	0.190	0.084	0.270	0.776	0.965	0.121	0.354	0.479
Time	**PC1**	**PC2**	**PC3**	**PC4**	**PC5**	**PC6**	**PC7**	**PC8**	**PC9**	**PC10**
Global	0.021 *	0.322	0.838	0.155	0.002 *	0.066	0.180	0.055	0.052 *	0.356
Chinese	0.171	0.878	0.983	0.789	0.294	0.910	0.306	0.220	0.201	0.523
Central Asian	0.277	0.869	0.463	0.568	0.599	0.800	0.639	0.824	0.718	0.853
Siberian	0.244	0.063	0.007 *	0.297	0.003 *	0.647	0.657	0.883	0.096	0.055
	PC11	PC12	PC13	PC14	PC15	PC16	PC17	PC18	PC19	PC20
Global	0.503	0.664	0.088	0.024 *	0.102	0.069	0.710	0.409	0.638	0.424
Chinese	0.973	0.914	0.752	0.810	0.982	0.450	0.419	0.981	0.679	0.713
Central Asian	0.887	0.894	0.671	0.648	0.927	0.996	0.515	0.614	0.708	0.758
Siberian	0.583	0.066	0.841	0.042 *	0.702	0.589	0.909	0.817	0.086	0.431
	PC21	PC22	PC23	PC24	PC25	PC26	PC27	PC28	PC29	PC30
Global	0.999	0.863	0.624	0.099	0.287	0.548	0.918	0.603	0.063	0.871
Chinese	0.044 *	0.483	0.137	0.051 *	0.478	0.601	0.177	0.228	0.457	0.734
Central Asian	0.04 *	0.079	0.063	0.207	0.613	0.185	0.995	0.938	—	—
Siberian	0.460	0.264	0.892	0.474	0.657	0.151	0.159	0.751	0.254	0.971

Mantel Correspondence Tests: The results from the Mantel matrix correspondence tests are shown in **Table 7.26**. The results shown are using absolute time (median site date), geographic distance in kilometers, and Mahalanobis distances between samples.

Significant correlations were found between when all 50 groups (global series) was included for biological and geographic distance (r = 0.33819, p = 0.0006), and between the Mongolian samples and Central Asian series for biological distance and spatial distance (r = 0.40558, p = 0.0126). Partial correlations were also calculated for samples using one of the matrices held constant. Only the Central Asian biological versus spatial (with temporal held constant) was significant (r = 0.41399, p = 0.0117). All other correlations were not significant at the 0.05 level.

TABLE 7.26. Mantel matrix tests showing Mongolian samples against the Chinese, Central Asian, Siberian and the entire Global cranial series. Mahalanobis distance matrix was used as morphological matrix.

Chinese Series	Partial	**r**
BIO x GEO		0.25325 (0.10809)
BIO x TEMP		−0.00173 (0.98850)
GEO x TEMP	Biological	0.14868 (0.21878)
BIO x GEO	Temporal	0.25636 (0.10129)
BIO x TEMP	Spatial (GEO)	−0.04117 (0.74533)
Central Asian Series	**Partial**	**r**
BIO x GEO		0.40558 (0.01260)*
BIO x TEMP		0.17934 (0.33237)
GEO x TEMP	Biological	−0.09133 (0.58204)
BIO x GEO	Temporal	0.41399 (0.01170)*
BIO x TEMP	Spatial (GEO)	0.20038 (0.27547)
Siberian Series	**Partial**	**r**
BIO x GEO		0.17066 (0.35036)
BIO x TEMP		0.08631 (0.14479)
GEO x TEMP	Biological	0.01451 (0.82922)
BIO x GEO	Temporal	0.16886 (0.35636)
BIO x TEMP	Spatial (GEO)	0.08261 (0.18988)
Global Series	**Partial**	**r**
BIO x GEO	Temporal	0.33819 (0.00060)*
BIO x TEMP	Spatial (GEO)	0.12618 (0.13889)

* significance in parentheses calculated after 10,000 permutations in a two-tail test (α = 0.05) distribution.

CHAPTER 8

DISCUSSION & CONCLUSIONS

Researchers and scholars have been interested in Mongolian culture and pre-history for many years. However, only recently have American researchers begun to unravel the complex history of Mongolia, in terms of archaeology, linguistics, ethnicity, ecology, and biological diversity, both of its people and its places (Sabloff, 2011; Schurr and Pipes, 2011). This dissertation has attempted to bridge a gap in our collective knowledge of a place and people few in the West truly understand. Though narrow in scope, I explored the origins of a steppe polity that emerged during the Late Bronze Age (circa second century BC) in an effort to better understand how the people of the modern nation-state of Mongolia are connected to nomads who lived over 2000 years ago. My results have shown, as has been shown previously by historians, linguists, and archaeologists that the people of Mongolia are as complex and nuanced as the landscape in which they reside. That is, the formation of Mongolia involved a series of complex demographic processes that included a diverse mix of confederations, clans, tribes, and families. To understand how these processes have shaped the biological diversity of modern peoples, we need to dig deep into the past and ask how patterns of kinship and confederation forged the biogenetic makeup seen today.

Those nomads of the Late Bronze and Early Iron age who formed the polity that became known as the Xiongnu are very much a part of the fabric that connects modern-day Mongolian people to the larger regions of Inner Asia. As a region known for the movement of peoples and cultures, marked by wars and territorial conquest, my results have shown the Xiongnu to be composed of several different biologically distinct

peoples. This finding, however, should come as no surprise to anyone who has studied the Central and Inner Asian steppe region. Archaeologists and molecular anthropologists have recovered traces of those interactions on numerous local and regional levels. Here, I have attempted to place the Xiongnu into a larger regional context by quantifying their population history and structure, and have unraveled a set of complex interactions and origins. Only by studying the people in the past are we able to address the origins and affinities of modern-day Mongolian peoples.

Using the data I have presented in this dissertation, along with biological data gleaned from other sources, I have attempted to describe the process by which Mongolian populations came into being, both in terms of chronology and geography. This chapter will place the Xiongnu into a context in which we will better be able to discuss their origins and relationships with groups throughout Inner Asia. I will discuss my results and compare those results to the appropriate literature, in both a biological and archaeological context. I will use my results to place the Xiongnu into a context that makes the people who composed this incipient steppe polity as important, if not more important, than Mongolian connections to the Mongol Empire of the 13th and 14th centuries. I will conclude this dissertation with directions for future research into Mongolia's prehistoric past, and how researchers can continue to collaborate in an effort to shine a light on a region that has hitherto been mostly disregarded in the annals of biological anthropology.

Xiongnu Population Structure

One of the goals of this dissertation was to assess within-group population structure of the Xiongnu. Was this group composed of several distinct biological groups? How can we interpret within-group variability of the Xiongnu? My assumption was that

the Xiongnu were composed of one biological group who would not show significant within-group heterogeneity, and further, that they would resemble, craniometrically, other Mongolian samples, regardless of temporal context. To test this assumption, I applied several indirect multivariate tests, including discriminant function analysis, Ward's clustering on individual Mongolian crania, and principal components tests after subjecting individual crania to general Procrustes superimposition using three-dimensional landmarks. I also applied a model-based approach to clustering that uses what is known as the Bayes Information Criterion to estimate the number of clusters inherent in the data (Schwarz, 1978; Fraley and Raftery, 2002). To test for within-group diversity, I used the model-based approach of Relethford and Blangero (1990) and accompanying measure of genetic divergence (F_{ST}).

Overall, the results from the model-based classification for the Mongolian samples indicate only one cluster in the data with little uncertainty in classification according to the Bayes Information Criterion. Keribin (1998) has shown the BIC to be consistent with the choice and number of clusters in the data. However, the difference in BIC value for two clusters was more than 100. According to Kass and Raftery (1995), differences of less than 2 in the BIC score are insignificant, while differences of more than 10 are often regarded as strong evidence for more than one component. This leaves open the possibility for two clusters, with the 'Egiin Gol' cluster that was obtained from the hierarchical analysis potentially valid.

Using these results, I then tested the Mongolian dataset using discriminant function analysis with *a priori* sample names (**Table 7.2**). Similar to the PCA, the Mongol Turk, Chandman, and Egiin Gol samples were separated along the first

eigenvector. Using a classifying procedure, the results indicate that although the aggregated Xiongnu sample classifies individuals into other groups, the Egiin Gol sample only classifies with itself (66.7% of the time), with the Turk sample (5.6% of the time) and with the Chandman sample (27.8% of the time). Using these results, I did not include the Egiin Gol sample in the pooled Xiongnu cluster in subsequent analyses to avoid biasing the data.

The R-matrix values between Mongolian samples are similar to the multivariate analyses (**Table 8.1**). The Relethford-Blangero (**Table 8.2**) analysis indicates less diversity (based on the R_{ii} values (distance to the centroid) within the two Xiongnu groups and greater diversity for the Mongol Bronze sample. These results also indicate that most of the Mongolian samples (except the Bronze) show negative residuals, indicating a degree of biological isolation. The Egiin Gol sample shows the highest negative residuals, possibly indicating some degree of biological, or cultural, isolation. The F_{ST} (genetic divergence) for these samples is relatively low (0.07), which indicates a low biological divergence between the samples.

TABLE 8.1. R-matrix and D^2 values for the Mongolian samples used in this dissertation.

Population	MongolTurk	MongolBronze	Mongolia	MongolPeriod	Xiongnu	EgiinGol
MongolTurk	*0.077192*	0.257271	0.249929	0.235064	0.232449	0.058960
MongolBronze	-0.024896	*0.130287*	0.347111	0.322821	0.207035	0.133009
Mongolia	-0.042540	-0.064584	*0.087656*	0.104472	0.066059	0.221131
MongolPeriod	-0.043567	-0.060898	0.026961	*0.070738*	0.055140	0.190915
Xiongnu	-0.058596	-0.019341	0.029832	0.026832	*0.038066*	0.146567
EgiinGol	0.029908	0.019431	-0.045946	-0.039296	-0.033459	*0.041584*

Positive values indicate a closer relationship than negative values than on the average. The diagonals are the R_{ii} values (distance to the centroid). Mahalanobis distances are along the upper right diagonal.

TABLE 8.2. Relethford-Blangero analysis for Mongolian samples used in this dissertation.

		Within-Group Phenotypic Variance			
Population	r_{ii}	Observed	Expected	Residual	SE
Xiongnu	0.03807	0.978	1.027	-0.049	0.007
EgiinGol	0.04158	0.890	1.023	-0.133	0.010
MongolPeriod	0.07074	0.975	0.992	-0.017	0.009
MongolTurk	0.07719	0.966	0.985	-0.019	0.021
Mongolia	0.08766	0.954	0.974	-0.021	0.006
MongolBronze	0.13029	1.168	0.929	0.239	0.012

F_{ST} = 0.074
V_{GW} = 0.988
h^2 = 1.0

It is clear from these results that there are possibly two separate clusters of biologically distinct individuals, at least on the basis of craniofacial variability. The first contains the Egiin Gol, Chandman, and Mongol Turk samples, the second contains the modern Mongolian, Mongol Period (Medieval) and the aggregated Xiongnu samples. The Xiongnu polity was composed of elite agents in an effort to control an administrative territory that stretched from Xinjiang Province in China to south Siberia. The Egiin Gol sample may represent an isolated element within Xiongnu society, while the other pooled Xiongnu sample may include individuals who composed the majority of its peoples. Or, perhaps the Egiin Gol sample is not entirely Xiongnu, and those individuals should be considered a part of the Turk (Uighur) Empire that dominated parts of Mongolia during the 8th and 9th centuries A.D. The pooled sample shows a clear relationship to both the Mongol period sample and the modern Mongolian sample. This finding would suggest that at least some individuals who composed the Xiongnu steppe polity are connected biologically to peoples who composed the Mongol Empire under Chinggis Khan, and to individuals who now compose the modern nation-state of Mongolia.

The Egiin Gol sample is difficult to interpret. It appears similar to both the Mongol Turk period sample, which is small (N=8) and the Chandman sample from

Bronze Age Mongolia (I will refer to this as the 'Egiin Gol cluster' throughout the discussion). Using traditional craniometric traits, Brace et al. (2001) and Seguchi (2004) showed that the Chandman sample did not cluster close to modern Mongolian samples. In fact, both their aggregated Xiongnu (Hunnu in their analyses) sample and the Chandman sample are more similar to modern Native Americans from the Great Lakes region, as well as prehistoric Archaic Period samples from North America. I have not sampled any Native American crania, and therefore am unable to test this interpretation, however, when plotted with the Jomon and Ainu, the Chandman do cluster closest to those groups, which are thought to represent an ancestral relationship to Native American groups (Seguchi et al. 2011). Tumen (2006) used traditional craniofacial traits and found that the Chandman sample resembles individuals representative of the Tagar culture of southern Siberia. This is certainly plausible since these two groups shared similar cultural features. The results from my analysis did not bear this relationship out, although higher dimension PCs did exhibit a relationship of the Chandman and Pazyryk people, who are closely related to the Tagar people (see below).

The Egiin Gol sample comes from a cemetery in northern Mongolia called Borkhan Tolgoi (Wright, 2006). The sample is named after the valley where several cemeteries are located and archaeologists have surveyed extensively (Wright, 2006). The cemetery of Borkhan Tolgoi has been examined previously by several researchers (Murail et al., 2000; Keyser-Traqui et al., 2003a, 2003b; Bennett and Kaestle, 2006; Crubezy et al., 2006; Wright et al., 2009; Ricaut et al., 2011). This site (necropolis) was used during the entire Xiongnu period and contains the remains of 84 graves containing

skeletal material from 99 individuals buried from the third century B.C. to the second century A.D.

The necropolis was arranged into three main sections that roughly correspond to temporal ordering as measured through AMS carbon-14 dating (**Fig. 8.1**). Section A is the oldest followed by Section B and Section C. Section A contains a number of "double burials" near graves marked with higher status individuals. This practice was quite common among peoples of the Scytho-Siberian tradition, including the Sakka (Yakuts) and the Bronze Age Pazyryk culture of the Gorny Altai in southern Siberia (Chikisheva, 2000; Ricaut et al., 2004a, 2004b; Amory et al., 2006). This practice has been reported in Murail et al. (2000) who investigated part of the Egiin Gol cemetery.

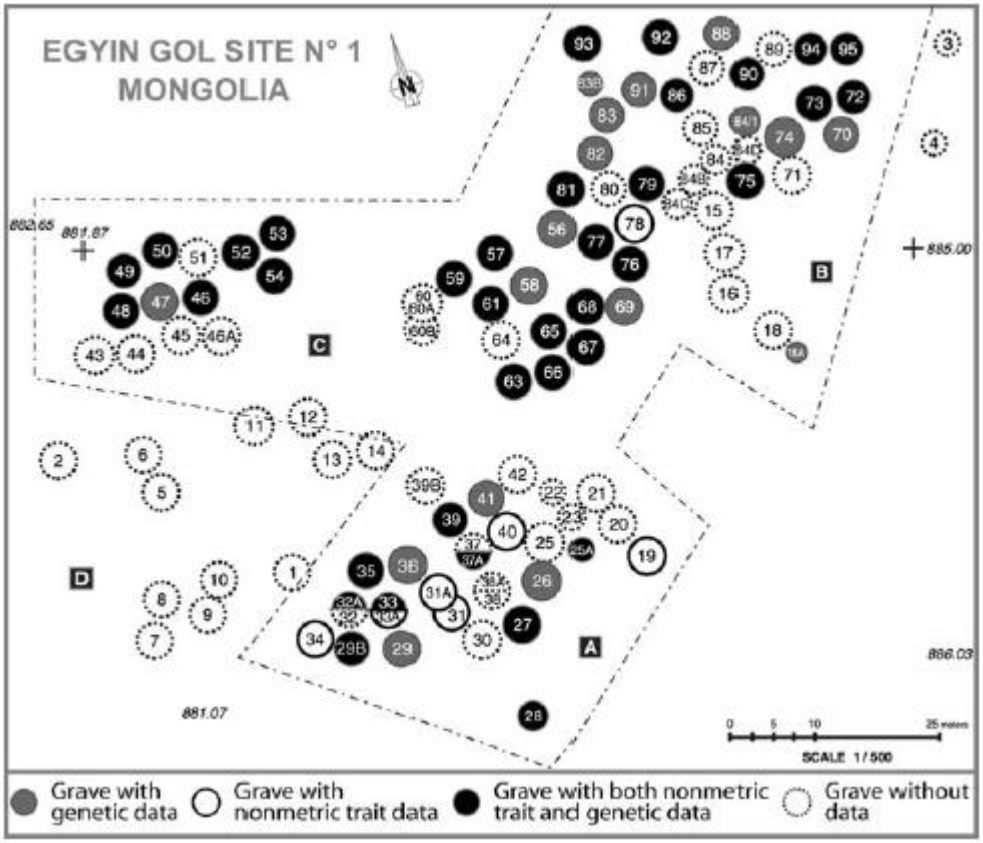

FIGURE 8.1. Egiin (Egyin) Gol necropolis (adapted from Ricaut et al. 2010, pp. 359).

Section C of the necropolis is interesting as it corresponds to the end of the cemetery's use and may be associated with a Turkish influence of the Xiongnu tribe (Keyser-Tracqui et al., 2003a). Based on STR (short-tandem repeat) genetic markers (autosomal and Y chromosome) and mtDNA, Keyser-Tracqui et al. (2003a) found distinct signatures unique to this section of the necropolis. Specifically, they found markers of a Turkish origin and a characteristically kin grouping in Section C that would seem to indicate a demographic shift in the necropolis toward the end of the Xiongnu Empire. Ricaut et al. (2010) also detected this unique signature in Section C using nonmetric cranial traits. Bennett and Kaestle (2006) also investigated the cemetery using mtDNA and included a greater diversity of populations that may be representative of the individuals buried at Borkhan Tolgoi. Using pairwise genetic distance (F_{ST}) derived from mtDNA HVSI sequences to calculate between pairs of populations, these authors found the individuals buried in the Egiin Gol cemetery showed close affinity with other East Asians, including Chinese Han, Northeastern Chinese, Mongolian, and Japanese. This finding is similar to what Keyser-Tracqui et al. (2003) found using haplogroup data (89% of sequenced individuals belonged to Asian specific haplogroups).

Bennett and Kaestle (2006) tested the observation of a possible Turkish component in Section C of the cemetery. They conclude that the subdivision within the cemetery may be more superficial in nature, and point out some methodological issues of making such a comparison due to differences in sample sizes for the two subdivisions (only 8 individuals for Section C). Their results indicate that the separated Egiin Gol sample (Sections AB combined, Section C) are similar to the reported results for the overall sample (including all sections), though they do note the existence of some

differences, which they do not expand upon in the article. Their observation of no significant differences does not entirely discredit Keyser-Tracqui et al.'s (2003a) conclusion as Bennett and Kaestle's (2006) analysis is only based on mtDNA (**Table 8.3**), and not autosomal or Y-STR data.

TABLE 8.3. Results of minimum F_{ST} values from Bennett and Kaestle (2006) study.

Match to Egyin Gol	F_{ST}
Xinjiang Han	0.01289
Mongolian	0.01586
Shandong	0.01630
Japanese	0.01755
Liaoning	0.01857
KirghizLL	0.01972
Korean	0.02271
Ewenki	0.02288
UighurXJ	0.03123
Kazakh	0.03355
Lambadi	0.05030
Lobana	0.07208
Bcqsa	0.08292
Tunisian	0.08344
Parsi	0.08908
Pushtoon	0.08955
Uttar Pradesh	0.09998
Pakistan	0.10157
RomaB	0.13780
Moroccan	0.15186
Cuman	0.15466
Armenian	0.16654
Georgian	0.16772
Hungarian	0.17357
Catalan	0.17485
Slovakians	0.18742
Icelander	0.19261
Biaka	0.44516

The methodology used by Bennett and Kaestle (2006) is also somewhat questionable. Though pairwise F_{ST} is a valid and informative approach to testing population divergence and gene flow, a more appropriate measure would be to use Slatkin's R_{ST} (Slatkin, 1995). This measure has similar properties to the classical measure of Wright's F_{ST}, but was developed because F_{ST} is used for loci with low mutation rates. This is not the case for microsatellite alleles (such as the hypervariable region of mtDNA)

with higher mutation rates. Slatkin's (1995) simulation results suggest that F_{ST} bias produces closer estimates of genetic similarity than distances estimated using R_{ST}. Though the authors cite relevant literature associated with some of the problems of using F_{ST} (Long and Kittles, 2003), and also include separate estimators of genetic distance (N_{ST}, D_a) in which they find similar results, using R_{ST} may have been more appropriate in this case.

I used a similar methodology as Bennett and Kaestle (2006) using pairwise F_{ST} from the quantitative characters (r_{ii} distances to centroid – analogous to genetic F_{ST}) to test the Mongolian samples affinity with all of the populations included in the analyses. These results are found in **Tables 7.9 – 7.11**. The analysis of 3D geometric morphometric traits used in this dissertation also indicates a distinction among the Egiin Gol sample from the pooled Xiongnu sample. Clearly from the cluster analysis, the Egiin Gol individuals show an affinity with the small sample of Mongol Turk from the 8th century A.D. Therefore, although the crania used in this dissertation have not been carbon dated to give an exact time since burial, nor can we correlate these crania with those used in the Ricaut et al. (2010) and Keyser-Tracqui et al. (2003) studies, they are most likely a separate population from the pooled Xiongnu sample, composed of individuals from various other places in Mongolia. Whether these individuals were distinct to the Egiin Gol necropolis toward the end of the Xiongnu Empire, or were a part of the administrative polity over a longer period will remain unknown until further sampling and larger sample sizes are achieved.

As for the pooled Xiongnu sample, it appears there is a strong connection to both the Mongol Period sample, which dates to the time of Chinggis Khan's reign (12th

century AD), and to the modern Mongol sample. Based on F_{ST} values, the pooled Xiongnu display an affinity with Xinjiang Chinese samples. Therefore, although there may have been a component of the Xiongnu that were not entirely Mongol, there is a strong connection for at least some segment of the Xiongnu society. This finding shows a clear line of descent from the Xiongnu polity through to those people now inhabiting the modern nation-state of Mongolia.

As for the population structure of the Chandman sample using F_{ST} values, it is similar to the Egiin Gol sample, Czech, and Jomon, followed by progressively larger values for all of the other populations. This is not an uncommon trend for the Chandman sample. Other studies (Seguchi 2004) have shown this distinction using traditional craniometrics. An interesting result is seen in how far away the Pazyryk sample is from the Chandman. On the basis of material goods located at the Chandman excavation (Miller 2009), the grave style and artifact analysis show similarities with the Pazyryk culture of southern Siberia (Chikisheva 2000). At least on the basis of the group sampled here, there is not any biological similarity to those Altaian nomads of southern Siberia, at least in terms of F_{ST}.

The Mongol Period F_{ST} values are interesting in that the closest groups are quite different. These include a Bronze Age sample from Inner Mongolia, an Uzbek sample from Central Asia, and an Early Iron Age sample from Siberia. These groups are not contemporaneous with the Mongol Period sample, but could indicate how diverse the Mongol Period individuals are – made up of Chinese, Central Asian, and Siberian populations. The modern Mongol sample also shows a mix of similar groups, though,

interestingly, other East Asian groups, such as South Chinese and Japanese are further away.

Xiongnu Population History

The origins and history of the Xiongnu are complex and multilayered as seen in the analysis of the group's population structure. The population history of the Xiongnu (and Mongolia) has been analyzed in a hierarchical manner. Where do these samples fit into the larger regional context? How do these samples compare when analyzed on a smaller, more local scale? I have compared the Mongolian samples to a large dataset consisting of groups from around the globe, and separately to three regions: China, Siberia, and Central Asia.

Global Comparison: The results from the global comparative analysis using 50 populations show two trends. The first places the Egiin Gol cluster on a branch with the Jomon and Ainu of Japan, and separated at a greater distance from the Xiongnu and Mongol Period samples. The principal component plots shown in **Figures 8.2** and **8.3** were drawn from group principal component means using the variance-covariance matrix. Plots have been scaled by their eigenvectors. In order to visually present the phenetic affinities among the series using PCA, D^2 (Mahalanobis distance) values were used to construct a Minimum Spanning Tree (MST) that was superimposed on the morphospace expressed as the first two or three principal components (Hartigan, 1975). Principal component plots are included throughout the discussion to allow greater ease in interpreting the results. The first three components account for 50.5% of the variance within the sample. It is clear from these plots the separation of the 'Egiin Gol' cluster from the other Mongolian samples.

Interestingly, this cluster as detected using Ward's clustering, also places these groups together, however, they are on a larger branch that includes groups from East Asia, Western Europe, Southern Asia, and Africa. The Mongol sample (modern), Mongol Period, and aggregated Xiongnu sample fall on a separate cluster that includes Siberian and Central Asian groups. Unfortunately, I was unable to include Native American samples to show the relationship of the Mongolian samples to groups in the New World. It has been proposed by many researchers using both quantitative characters (Brace et al., 2001; Hanihara and Ishida, 2005; Gonzalez-Jose et al., 2008; Hanihara, 2008; de Azevedo et al., 2011; Hubbe et al., 2011) and genetics (Kolman et al., 1996; Karafet et al., 1999; Santos et al., 1999; Zegura et al., 2004; Starikovskaya et al., 2005; Tamm et al., 2007; Adachi et al., 2011; Dulik et al., 2012) that Asia, and in particular southern Siberia, have been large contributors to the ancestral gene pool of Native American peoples.

The Ward's clustering could also be indicative of the peopling of greater East Asia. It has been suggested on the basis of genetic and morphological data, the peopling of East Asia occurred along a southern route during the Paleolithic, however, some studies have suggested a Neolithic contribution to Northeast Asian groups from West Eurasia and Central Asia (Derenko et al., 2007a; Zhong et al., 2011). This is evident in the clustering of southern Siberian and Mongolian samples with Central Asian and Northern European populations. Based on modern samples using the Y chromosome and mtDNA, researchers have suggested a contribution of Neolithic expansions on Northeast Asian populations, including indigenous Siberian and Mongolian groups. Major Y chromosome haplogroups Q and R that derive from Western Eurasian populations have been detected in Northeast Asian groups, while several mtDNA haplogroups (H, V, and

X) show signatures of postglacial expansion into Northeast Asia (Reidla et al., 2003; Derenko et al., 2007). These distinctive signatures have also been found in ancient skeletal remains from places such as southern central Siberia, the Tarim Basin and Xinjiang province in China (Lalueza-Fox et al., 2004; Ricaut et al., 2005; Keyser et al., 2009; Li et al., 2010; Zhang et al., 2010).

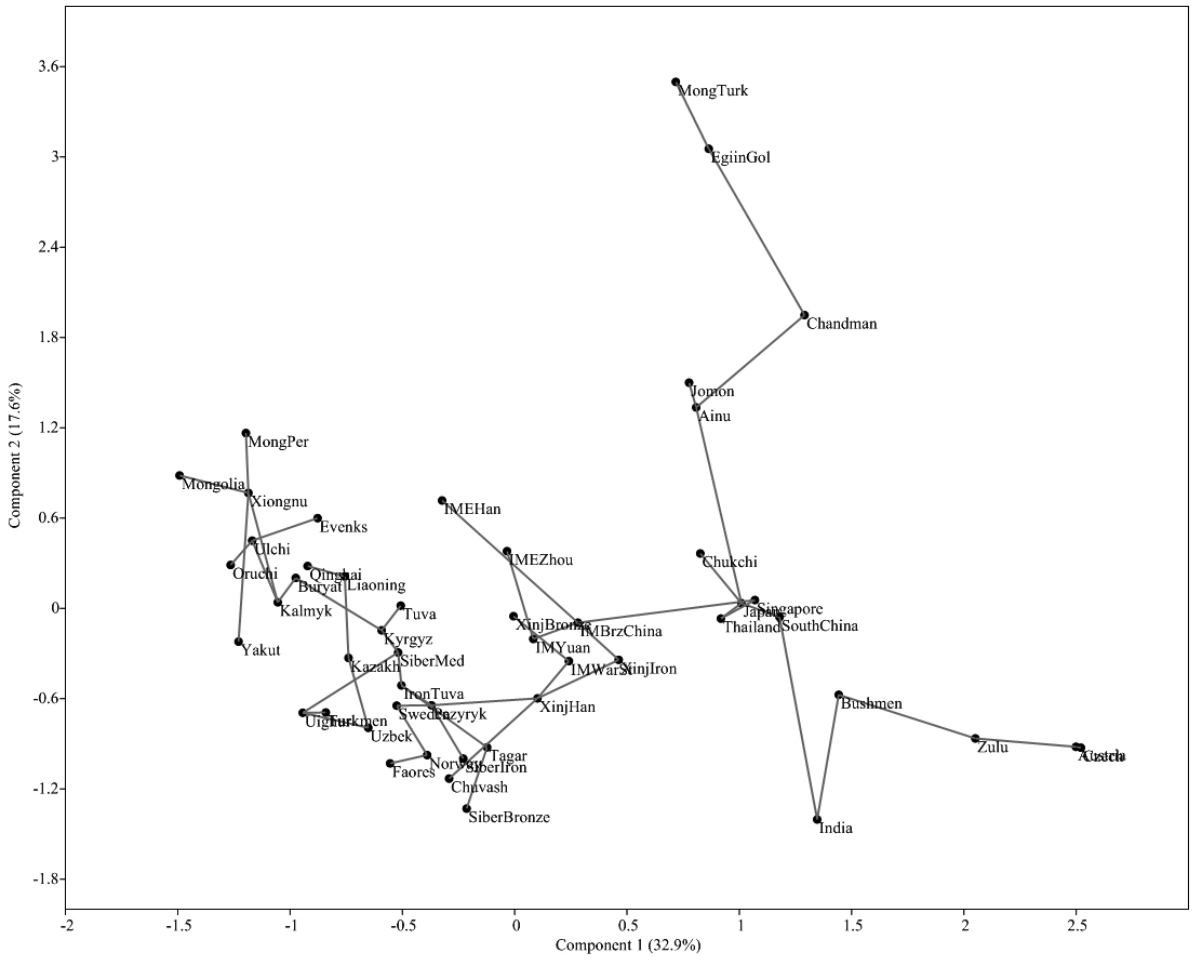

FIGURE 8.2. Minimally spanned ordination of sample PC scores for the first two principal component axes for global cranial series.

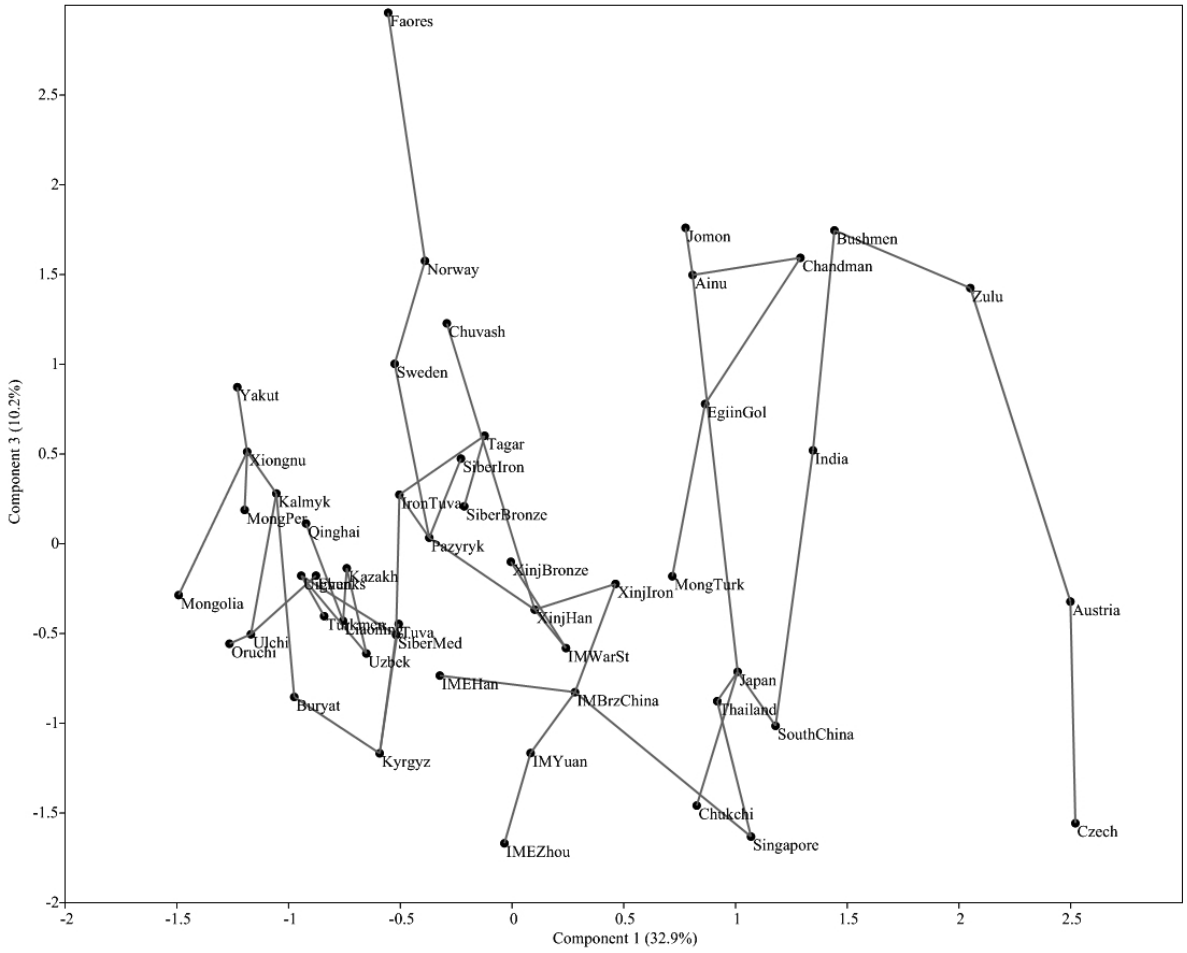

FIGURE 8.3. Minimally spanned ordination of sample PC scores for axes one and three for the global cranial series.

Similar to previous studies that have used a global distribution of samples to infer biological (phenotypic) variability between groups (Manica et al., 2007), this study has shown a positive significant correlation between biological distance (using Mahalanobis distances) and geographic distance between samples using a partial Mantel test ($r = 0.33819$, $p = 0.006$) when controlling for time.

A multiple regression approach was used to account for which PCs (95% of the variance) that might be correlated with one of three variables: latitude, longitude, and time. The global analysis revealed several PC scores that were strongly and significantly

correlated ($p < .05$) with latitude: 6, 14, 15, 27, 29. PC 6 is accounting for shape changes in upper facial breadth, cranial height and length; PCs 14 and 15 are accounting mostly for orbital and malar height; PCs 27 and 29 account mostly for nasal height and breadth. Interestingly, more PCs were significant for longitude. These include PCs 4, 6, 7, 9, 11, 15, 21, 22 and 30. PCs 4, 6, and 7 account for shape changes in cranial length; PCs 9, 11, and 15 account for cranial height and upper facial height; PC 21 is a change in jugale (more forward projecting), PC 22 accounts for change in glabella (perhaps a sex size factor), and PC 30 accounts for change in the nasal region. PCs 1, 5, 9 and 14 are shape and size changes related to time within the samples.

The greater number of variables correlating with longitude is interesting. In a recent study, Ramachandran and Rosenberg (2011) found that latitude tended to contribute more to the genetic differentiation in Eurasia. This conflicts with these results, however, more samples are included in this study than what was used in the Ramachandran and Rosenberg (2011) study.

Chinese Comparison: The Mongolian samples were then compared to just populations from China and Japan. Again, it is shown the 'Egiin Gol' cluster is separating those groups from all others in the analyses. Plotting these groups using principal components analysis and minimum spanning tree also shows this cluster for the fist two axes (**Fig. 8.4**), however, plotting the second component against the third component reveals a closer association for the pooled Xiongnu and the Liaoning sample from Northeastern China (**Fig. 8.5**). The first three components account for 79.3% of the variance.

The Liaoning sample dates from the second to third century AD. According to archaeological and historical evidence, the individuals buried at this cemetery might be

the descendants of the Donghu people, who, according to Chinese accounts, were a Mongolic nomadic people who occupied northeastern China and were conquered by the Xiongnu in 150 BC. These people were later broken into the Xianbei Empire, who have historic ties to the Xiongnu and modern Mongolian peoples (Di Cosmo, 2002). There is also a close relationship with samples from Qinghai, which span several periods, ranging from the Han through the Jin (265 – 420 CE). This area has also historically been a melting pot of peoples, having been home to the Xianbei, Turkic, Chinese Han, and Mongols. In the PC plots, the Xiongnu also group together with samples from the Eastern Han Empire (25 – 220 CE). This sample (Eastern Han) comes from several locations in Inner Mongolia.

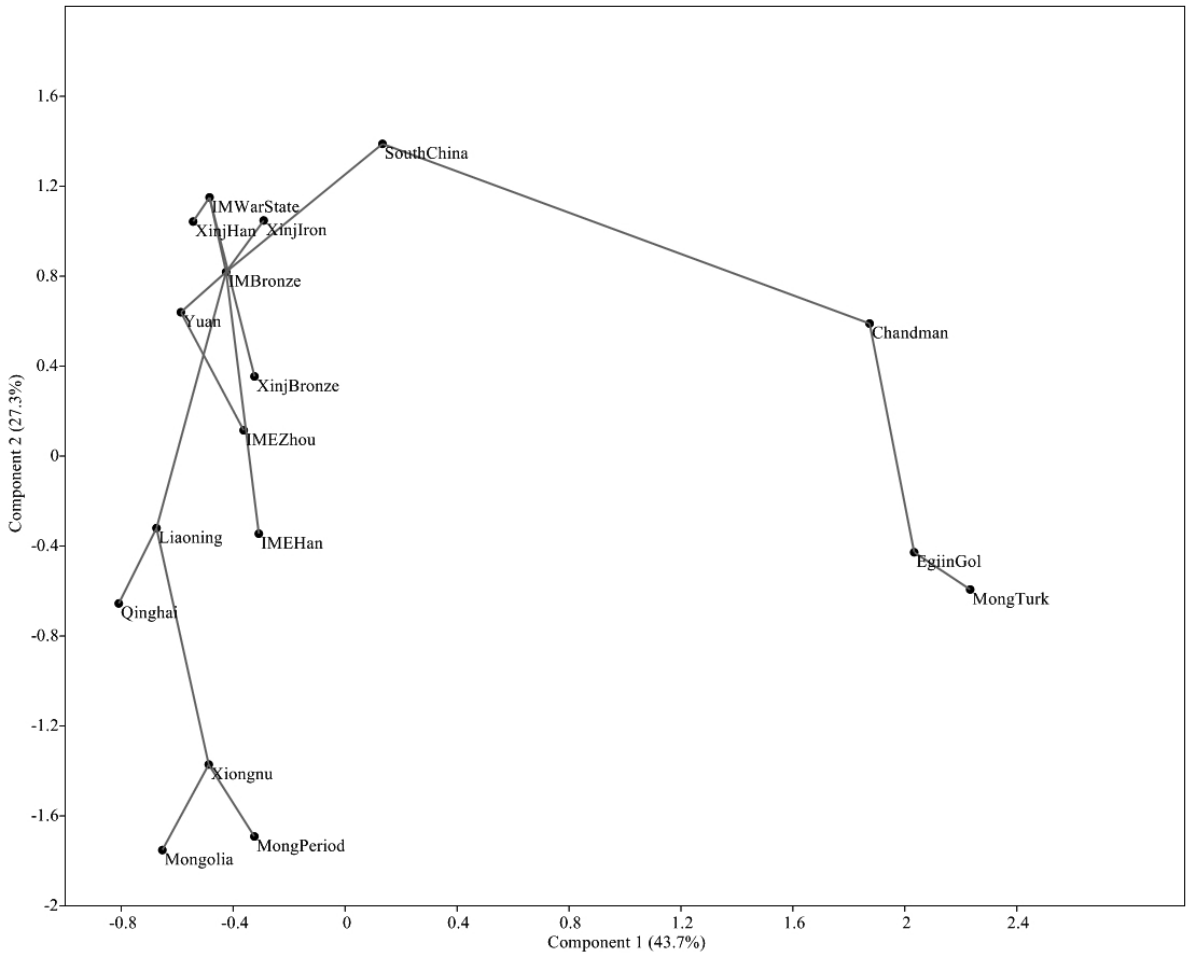

FIGURE 8.4. Minimally spanned ordination of sample PC scores for the first two principal component axes for Chinese comparative series.

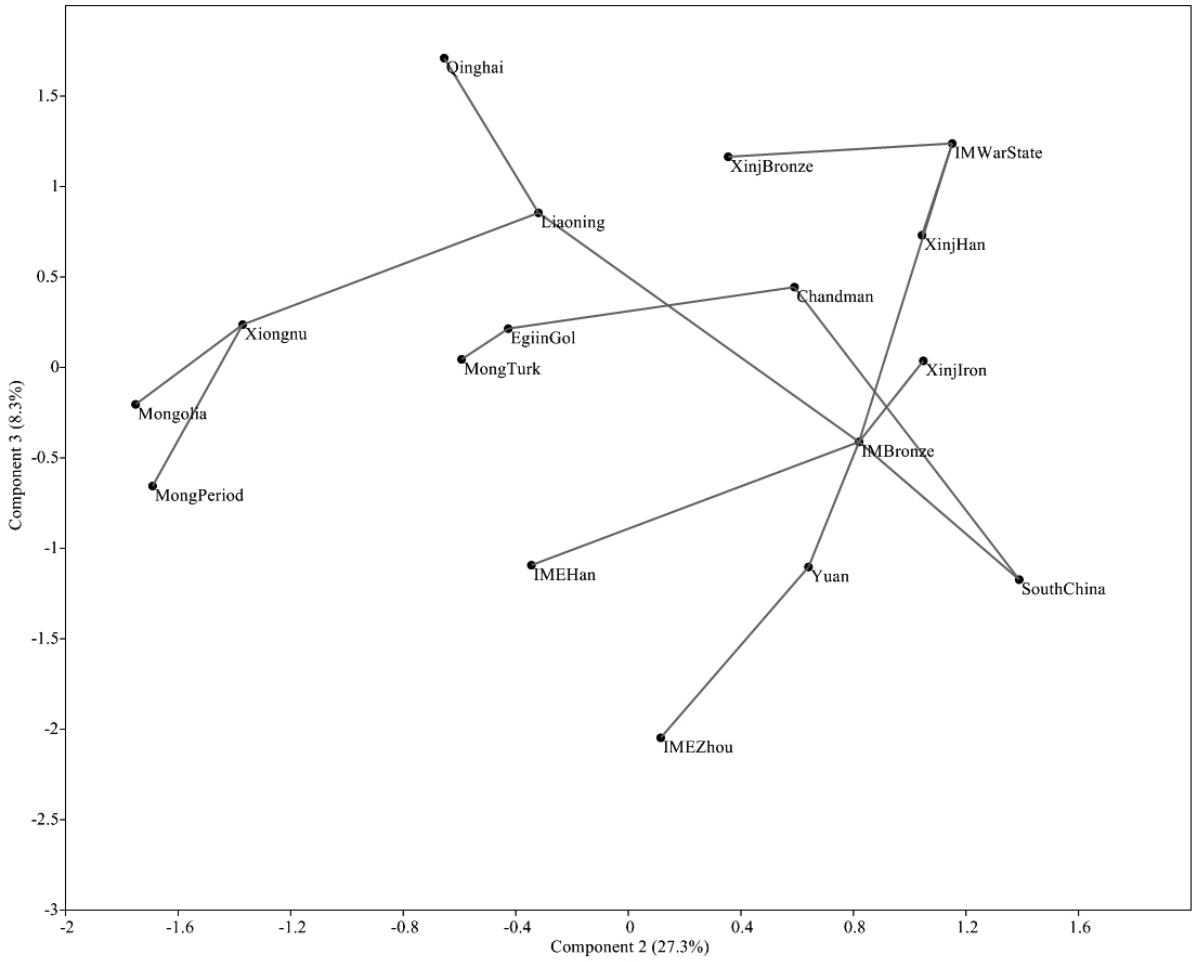

FIGURE 8.5. Minimally spanned ordination of sample PC scores for principal component axes two and three for Chinese comparative series.

The affinity between the Xiongnu sample and the Qinghai, Liaoning, and the Eastern Han is also seen in the distance matrices (Mahalanobis and R-matrix). After the Mongol Period and modern Mongolian sample, the next closest D^2 distances are the Liaoning (0.108), the Eastern Han (0.128), and the Qinghai (0.129). Though not shown, the R-matrix shows this observation as well, as all three of these samples are positive, indicating some level of gene flow among these groups. Interestingly, the modern Mongolian sample shows a similar trend for biological distances.

The results from the Relethford-Blangero analysis indicate some biological divergence (F_{ST} = 0.21) using a value of 1.0 for the narrow-sense heritability (minimum genetic distance). Using an average value of 0.55 yield a higher estimate of F_{ST} of 0.33. The 'Egiin Gol' cluster shows relatively high heterozygosity (r_{ii} values), though a larger standard error for the Mongol Turk sample, most likely due to the small sample size. The Xiongnu aggregate and modern Mongol samples estimate a negative residual for both groups, which means there may be some isolation when compared with the Chinese groups, though this estimate is small (-0.024). The Mongol Period sample shows greater than expected residuals (positive), meaning this sample may be experiencing some degree of gene flow, though again, the residual is rather small (0.063).

Results from the cluster analysis with 1000 bootstrap replicates have the 'Egiin Gol' cluster with high bootstrap support values (100% for both), though the NJ tree has Egiin Gol as an outlier to the others (93% bootstrap value for the Chandman, Mongol Turk cluster), while Wards has the Bronze Age Chandman as an outlier with an Egiin Gol/Mongol Turk cluster (79% bootstrap value). All Mongolian samples occupy a separate cluster in both analyses, though in the NJ tree the Qinghai (83%), followed by Liaoning (70%) and Inner Mongolian Eastern Han (44%) are outliers to the Mongolian samples, with decreasing bootstrap values for the other Chinese samples. Wards clustering ties all of the Mongolian samples together and are a separate branch entirely from the Chinese samples. Interestingly, in the NJ tree, the Xiongnu and Mongol Period samples cluster together with high support value (71%) with a node that connects to the Egiin Gol cluster, with high support for the modern Mongol sample (99%) connected to all of the Mongolian samples.

To test for isolation by distance or temporal correlation within the samples, partial Mantel tests were conducted. The results from the Mantel tests do not show any significant correlations either for geography or time. Biological distance and geographic distance did have a positive correlation while biological distance and temporal distance had a negative correlation (as predicted under Konigsberg, 1990), however these values were not significant. There were a few individual PCs that did show correlation with significance for latitude (PC 10), longitude (PCs 8, 15, 16, 19), and time (PC 21).

Few studies published in English have examined Chinese craniometric variation. A recent study examined Chinese Neolithic crania to test for differences between North and South Chinese (Wu et al., 2012), while Chan (2011) explored the Bronze Age Anyang sample (1600 – 1046 BC) and its relation to modern peoples of Hong Kong and Thailand. Schmidt et al. (2011) used Chinese immigrants in a study to test origins and migration patterns to North America, but did not include many groups from northern China. Pietrusewsky (1990, 2008, 2010) routinely includes Chinese data in his studies, however, these studies tend to focus on broad trends in craniofacial morphology through time for large areas of Asia and island Southeast Asia. It is interesting to note that his inclusion of a Mongolian sample from Ulaanbaatar provided by Hrdlicka are extreme outliers in his analysis using canonical variate plots and UPGMA distances (Pietrusewsky, 2010).

There are, however, several studies that have extracted ancient DNA from the same samples tested in this dissertation. In the pairwise F_{ST} analysis conducted between groups, the Mongol Period sample showed a close affinity with the Chinese sample labeled “Inner Mongolian Bronze”. This sample is from a cemetery called Chengbozi and

is located in Ulan Hua town of Siziwang Banner in Inner Mongolia, northern China. Although labeled as Bronze Age (label taken from Chinese physical anthropologists), these individuals date from the Jin-Yuan period, approximately 800 yBP. Molecular analysis was conducted on 16 individuals from this same cemetery (Fu et al., 1007). These individuals belonged to the Wanggu tribe, who, according to Chinese historical and archaeological sources, played an important role in the founding of the Yuan dynasty, and helped Chinggis Khan destroy the Jin dynasty (Zhou, 2001).

There is some argument as to whether the Wanngu tribes were originally derived from Turkic tribes, or were one of the Mongolian tribes (Gai, 1991). The results from the genetic analysis indicate a diverse mix of haplogroup sequences, most shared with East Asian, Siberian, and Central Asian groups. However, like other studies, these individuals also shared some European specific haplogroups, signifying the complex nature of the maternal structure of groups in this area of the world. The similarity of the Mongol period sample and this Inner Mongolian sample would seem to indicate that the Wanggu tribe were a Mongolian tribe, at least in terms of shared biological affinity as seen in craniofacial morphology. Fu et al. (2007) hypothesize the Wanngu were of Turkic origin due to genetic similarity, however, this is based on a single line of evidence (mtDNA), and does not include other genetic systems, which might indicate a closer genetic similarity to the Mongols, who admixed with Turkic groups at least since the 6th century AD.

A similar study reports on the molecular analysis of remains from the Upper Capital city of Kublai Khan on the Jinlianchuan steppe of Inner Mongolia (Fu et al., 2009). These individuals were excavated from the Zhenzishan cemetery, which is located

in proximity to the Upper Capital. This sample is the same sample as that labeled "Inner Mongolia Yuan" in this dissertation. Archaeologists had suggested these individuals were part of the Han who lived close to the Upper Capital at the time of the Yuan dynasty, which was controlled by the Mongols under the leadership of Kublai Khan. However, physical anthropological analysis revealed features that would be shared with groups from Mongolia and elsewhere (Wei, 2004). Therefore, ancient DNA was extracted (mtDNA), and it was revealed that those individuals belonged to characteristic Asian maternal haplogroups shared with present-day Han Chinese (Yao et al., 2002b). However, some individuals had observed haplotypes distinctive of Mongols, Oroqen, and Ewenkis (Kong et al., 2003), which would reflect the interaction between the Han and Mongolian groups. This dissertation has shown through pairwise F_{ST} that both the Mongol period sample (0.042) and the modern Mongolian sample (0.064) are closely related to the Inner Mongolian Yuan (Upper Capital) sample.

To summarize the Chinese comparison, the pooled Xiongnu sample does show some similarity to several Chinese populations, especially to the Liaoning sample from Northeast China (Xianbei), and the Qinghai sample, located on the northwestern part of the Tibetan Plateau. Both of these locations have experienced significant demographic shifts, of which the inhabitants have been connected to the Xiongnu Empire. The 'Egiin Gol' cluster remains isolated in the analysis. The only population that shows any biological affinity are the Ainu and Jomon of Japan. The Mongol Period sample is closely related to several samples from Inner Mongolia. These connections are most likely the result of historical and demographic processes that brought the Mongols into contact with sedentary populations living in Inner Mongolia.

Central/Southern Asian Comparison: A separate analysis was carried out to compare the Mongolian and Xiongnu samples to a cluster of modern-day Central and Southern Asian samples. These include samples from India, Kazakhstan, Kyrgyzstan, Turkmenistan, Uzbekistan, a group of Uighur from western China, and a sample of Chuvash, a Turkic ethnic group now living in Russia. All of these groups speak a Turkic language. The PC plot with minimum spanning tree clearly shows a separation of the Mongolian samples on PC1 and PC2. The minimum spanning tree does indicate some similarity of the Xiongnu to the Kazakh sample, though still relatively distant. When PC2 is plotted against PC3 (**Fig. 8.6**), a much closer relationship is seen between Egiin Gol and the Kazakh sample. The first three coordinates account for 84.1% of the variance within the sample.

The Kazakhs are descendants of various Turkic tribes. When PC3 is plotted against PC4 (not shown), the Mongol Turk sample falls squarely into the Turkic cluster, although the variance for these components are low. When PC 1 is plotted against PC 3, both the Xiongnu and Mongol Period samples cluster close to the Chuvash and Turkmen. In the pairwise F_{ST} analysis, the Xiongnu sample is closely related to the Kazakh sample (0.006). In a study by Lalueza-Fox et al. (2004), the authors show that the Kazakh population is composed of West and East Eurasian mtDNA haplogroups. However, it wasn't until the expansion of the Xiongnu did the authors find traces of East Eurasian haplotypes – specifically haplogroups A and G2. The authors attribute these sequences to the migrations of the Xiongnu from Mongolia and Siberia. These observations bear further evidence for the influence of the Turkic tribes on the Mongolian populations, or vice a versa.

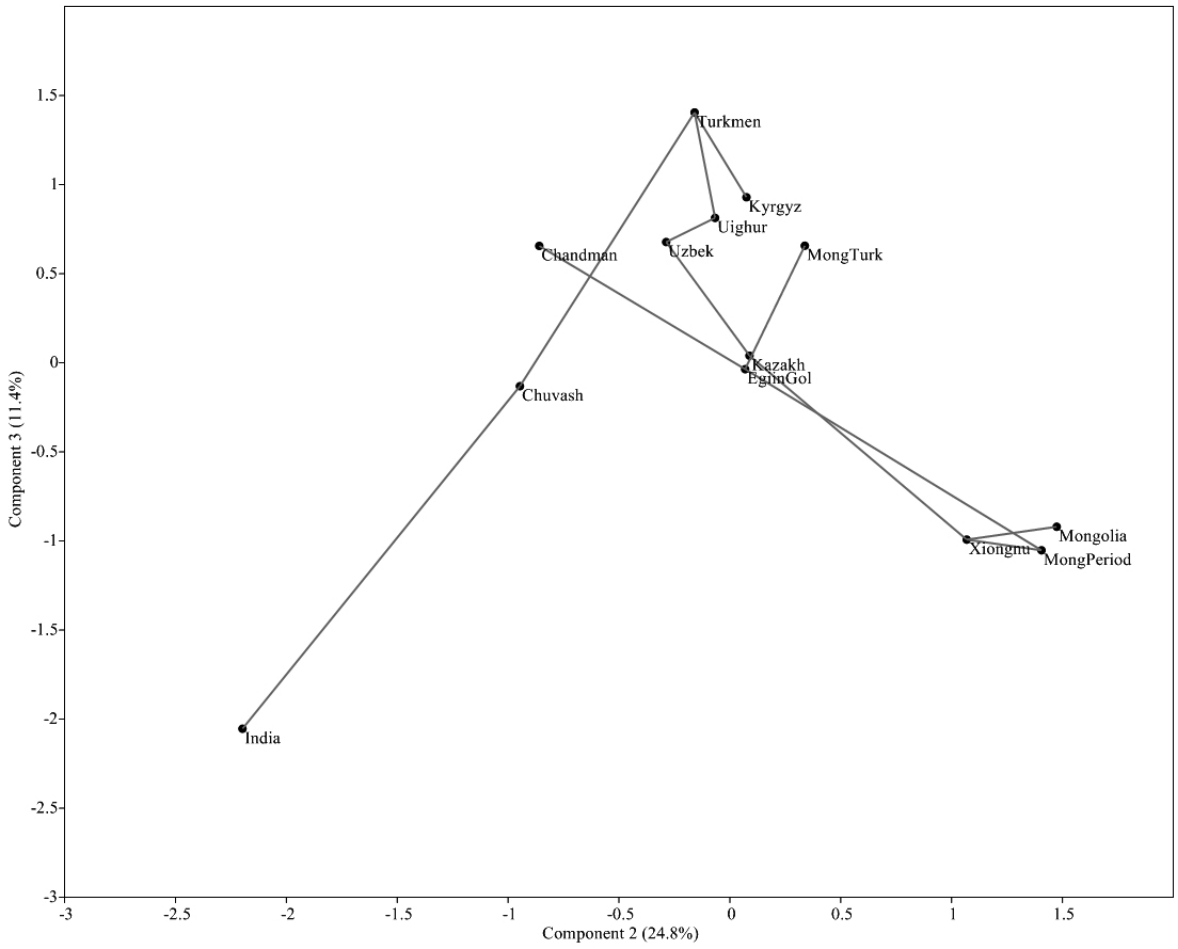

FIGURE 8.6. Minimally spanned ordination of sample PC scores for principal components two and three for Central Asian comparative series.

The results of the Relethford-Blangero analysis indicate more genetic differentiation than previously found when comparing the Mongolian samples to the Chinese samples. Overall, F_{ST} is higher than might be expected (0.24, $h^2 = 1$). All of the samples in the analysis, except the 'Egiin Gol' cluster and the India sample show negative residuals, or less than expected levels of heterozygosity. This result is interesting since Central Asia has been genetically determined to be one of the most diverse areas in the world (Comas et al., 1998; Wells et al., 2001; Comas et al., 2004; Quintana-Murci et al., 2004; Chaix et al., 2007, 2008).

The partial Mantel tests did indicate a significant positive correlation between these samples ($r = 0.412$, $p = 0.011$) for biological distance and geographic distance while holding temporal distance constant. This is another surprising result given the amount of genetic admixture seen between these groups. There were no PCs that were significant for either latitude or longitude, which is strange considering the significant correlation seen in the Mantel test. PC 21, as was seen in the Chinese comparison, was significant for time.

Overall, it would seem there is less similarity among Mongolian and Central Asian samples than that which is evidenced through genetic data. These data do suggest that the Turkic speaking groups, especially the Kazakhs, are related to some Mongolian samples, at least on axes of lesser variation. However, the Central Asian samples used here are modern, and may not reflect the biological diversity seen during the Bronze and Iron Age. Inclusion of such samples may change this relationship.

Siberian Comparison: Lastly, the Mongolian samples were separately analyzed against a number of geographically and temporally distinct groups from Siberia. These samples range from southern and Western Siberia to populations now inhabiting areas close to the Sea of Okhotsk. Several samples were included for temporal comparison. The results from the principal components analysis once again show the 'Egiin Gol' cluster to be an outlier compared to the other samples in the analysis when viewing PC1 and PC2. This is also seen in the clustering analysis. The Xiongnu show some relationship with the Yakut in the PC plots. The Chandman sample shows some affinity with the Pazyryk sample along PC3 (**Fig. 8.7**). The first three components account for 78.8% of the variance. The

partial Mantel tests for correlation were not significant for either time or geography. However, several individuals PCs were significant for time.

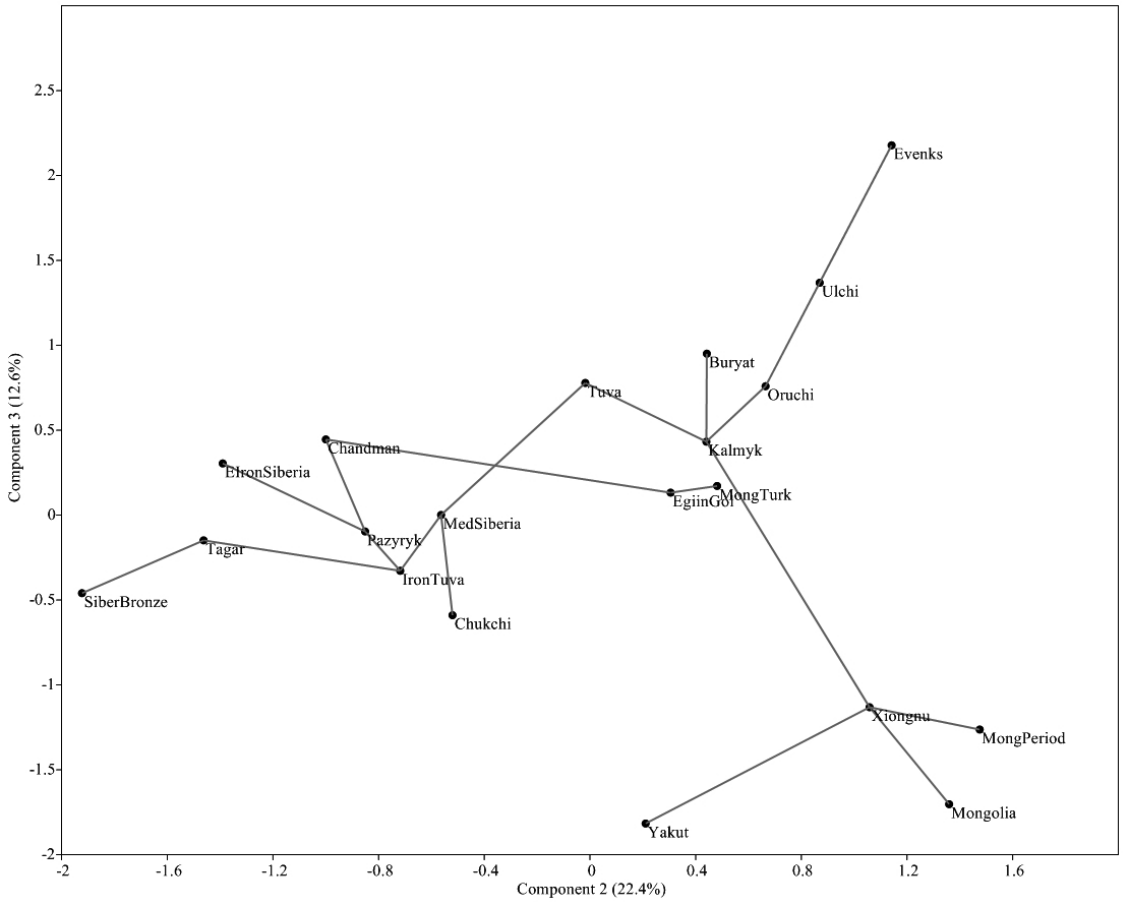

FIGURE 8.7. Minimally spanned ordination of sample PC scores for principal components axes two and three for Siberian comparative series.

The Yakuts (or Sakha as they call themselves) are a Turkic-speaking group with borrowed Mongolic words who reside in the modern republic of Yakutia, an autonomous region in Central and northeastern Siberia that is part of the Russian Federation. They are semi-nomadic cattle and horse breeders surrounded by Tungustic-speaking reindeer herders (Evenks and Evens) and hunter-gatherers (Pakendorf et al., 1999). This group has been studied extensively at the genetic level, including modern populations' autosomal loci (Pakendorf et al., 1999; Gouriev, 2004), mtDNA and Y chromosome diversity

(Pakendorf et al., 2003, 2006; Keyser-Tracqui et al., 2006; Zlojutro et al., 2008, 2009; Fedorova and Khusnutdinova, 2010), and ancient DNA analysis (Amory et al., 2006; Ricaut et al., 2006; Crubezy et al., 2010). Linguistic evidence suggests the Yakuts have ties to southern Siberian Altai-Sayan region Turkic speaking groups (Ruhlen, 1987) and migrated more recently to northeastern Siberia.

The evidence from mtDNA suggests the Yakuts are closely related to southern Siberian and Central Asian groups, which confirms a southern origin. The timing of their northward migration has been suggested as being caused by the expanding Mongol empire (Pakendorf et al., 2006). Both maternal (Zlojutro et al., 2008) and paternal (Pakendorf et al., 2006) lineages suggest a bottleneck event at around 800-1000 BP, very close to the founding of the Mongol Empire. These contacts are seen in the craniometric evidence presented in this dissertation.

The origin of the Yakut population is more complex. Several studies have characterized the ancient DNA of the Yakuts. The findings vary. Using ancient DNA taken from the Egiin Gol cemetery, Keyser-Traqui et al. (2006) compared modern Yakut DNA with the ancient DNA from the Xiongnu. These authors found no evidence, on the basis of Y chromosomal analysis, for a link between Xiongnu and Yakut, however, some of the Xiongnu individuals had shared mtDNA sequences with modern Yakuts. The lack of Y chromosome similarity could be the result of a significant loss of genetic diversity in the Yakuts after their contact with the Mongol Empire, which probably resulted in significant loss of males to the gene pool, or could simply result from genetic processes, such as genetic drift.

Amory et al. (2006) characterized the mtDNA of a single Yakut individual (dated 2300 yrBP) from the Altai-Baikal region near the Lena River and found the mitochondrial haplotype of this individual matched a woman buried at Egiin Gol cemetery. Crubezy et al. (2010) analyzed a more extensive sample of Yakuts from the 15th century. They found that the male lineage was composed of a small group of settlers from the Cis-Baikal region and that the maternal lineage was more diverse and composed of groups from different south Siberian origins. This dissertation has shown a direct link between the Yakut and Xiongnu based on craniofacial variability. This is seen in not only the principal coordinate plot, but also in the R-matrix and Mahalanobis distances. This evidence highlights an admixture event(s) between peoples now living in central and northeastern Siberia, and the Xiongnu.

The Chandman sample does connect to the Pazyryk sample when PC2 is plotted against PC3. The Pazyryk people represented a culture from southern Siberia (Altai) dating to the 5th to the 3rd centuries B.C. (Chikisheva, 2000, 2008). Archaeologically, the Pazyryk people trace their origins to the Scytho-Siberians of the 7th century BC. (Rudenko, 1970). The craniofacial variability of the Pazyryk people has been documented as being a mix of Eastern and Western Eurasian features. Chikisheva (2000) believes the 'Caucasoid' element present in the Pazyryk population is due to their relation with pastoral groups inhabiting the Central Asian steppe and/or the Near East (northeastern Iran, Turkmenistan, and southern Uzbekistan and Tajikistan). There may be some validity to this argument as seen in the global analysis used in this study.

The Pazyryk sample is part of a larger cluster that includes Uzbek and Kazakh samples in the global Ward's clustering tree. In addition, several studies have used

ancient DNA to characterize these southern Siberian populations. Keyser et al. (2009) analyzed ancient DNA from the Krasnoyarsk region of the Russian Federation on specimens dating from the 2nd century BC to the 4th century A.D. Their results indicate these people belong to Eastern Eurasian specific haplogroups connected to the eastward migration of Kurgan peoples. This result is similar to that obtained from Lalueza-Fox et al. (2004), who found on an ancient Kazakh sample that all of the specimens prior to the 7th century BC belong to European lineages. After that time, an influx of East and Northern Asian sequences (resulting from the Xiongnu Empire) appeared and continued to co-exist with these European-specific sequences.

Chikisheva (2000) finds it more difficult to account for the 'Mongoloid' traits within the Pazyryk sample. She believes the component comes from the steppe region of Mongolia and the Baikal region. She bases this conclusion on the observation that the Pazyryk people display "Paleosiberian" features associated with Neolithic tribes inhabiting the Lake Baikal region. It is interesting to note that in the R-matrix analysis, the Pazyryk sample does show positive residuals in comparison with the modern Mongol and the Mongol period samples. This is also reflected in the D^2 distances between these groups.

The Chandman sample is based on a site-type attributed to the Pazyryk people and known as the Chandman culture (Tseveendorj, 1980). The site is associated with kurgans, which are barrow chambers holding one or several individuals, and is located in northwestern Mongolia. However, as pointed out by Miller (2009), the sites ascribed as Chandman are drastically different from other traditions elsewhere in Mongolia. This is evidenced in the grave goods and assemblages of these Pazyryk culture people that differ

from other Mongolian traditions. Nonetheless, the Chandman sample used in this study does show some affinity with the Pazyryk sample of southern Siberia, thus offering a connection for further research.

The results from the Relethford-Blangero analysis show a higher level of biological differentiation (F_{ST}) than with other analyses. Interestingly, the pooled Xiongnu sample falls between the Pazyryk and Tuva sample for distance to the centroid. The Republic of Tuva is located adjacent to the northwestern portion of Mongolia. The Tuvan people share many cultural traits with both the Mongols and the Buriat people of the Baikal region. mtDNA and Y chromosome analysis indicate the Tuvan population to be closely related to other groups from the Altai, but distinct from groups inhabiting the Baikal area, such as Buriats and Mongols (Derenko et al., 2002b, 2006). The cluster analysis used in this dissertation shows two different results. Wards clustering places the Tuva sample next to an aggregated Western Siberian Medieval sample, and into a larger cluster that contains other southern Siberian samples and the Mongol samples (excluding the 'Egiin Gol' cluster). The NJ tree, in contrast, shows the Tuva sample to be an outgroup to Baikal samples (Buriat and Kalmyk) and northeastern Siberian groups (Ulchi, Oruchi, and Evenks). This cluster is supported with high bootstrap values (99%). This observation is also apparent in the R-matrix and D^2 distances, which indicate the Tuvan sample to be more differentiated.

Though the clustering analysis does not show a direct link between the Xiongnu and the Tuva sample, Chinese historical records imply the ancestors to modern Tuvans were known as the Dingling (Li, 2003). The Xiongnu conquered them in 51 BC, and in 85 AD, the Dingling joined with the Xianbei to defeat the Xiongnu, but most Dingling

were assimilated into northern Xiongnu tribes (Duan, 1988). The PC plots do indicate some affinity as seen in the MST from **Figure 8.7**.

The Kalmyk sample's placement is interesting in the cluster analysis. The Kalmyks are thought to be descendants of the Oyrats of western Mongolia (Nasidze et al., 2005). They now live along the banks of the Volga in eastern Russia and are thought to have migrated there around 300 years ago. Y chromosome and mtDNA analysis have shown a very close relationship with modern Mongol peoples (Nasidze et al., 2005). However, in the analysis of craniofacial variation presented in this study, the Kalmyk sample shows a stronger affinity to groups from northeastern Siberia. This is surprising considering that the so-called "Chinggis Khan" STR Y chromosome haplotype (Zerjal et al., 2003; Derenko et al., 2007b) is so prevalent among the Kalmyks (31.3%). This haplotype derives from the time of Chinggis Khan and is found throughout Eurasia. It is believed by several authors that the haplotype was dispersed from social selective processes attributed to the male linage of Chinggis Khan. However, the Kalmyks do show some similarity with the Tuvan sample, which is observed in the mtDNA (Derenko et al., 2000, 2002b), and the Buriat sample, which is observed in various genetic polymorphisms (Galushkin et al., 2001; Gilbert et al., 2010) and whole mtDNA sequencing (Derenko et al., 2007a). Another interesting result is found in the global analysis where the Kalmyks are clustered together with groups from Central Asia, such as the Uighur, Turkmen, and Kyrgyz. This could be represented of more recent gene flow with these groups, though their language is Mongolic, not Turkic.

It has been shown on the basis of DNA markers and craniofacial characters that the region of Siberia is quite heterogeneous. It appears the Mongolian samples show

some affinity with a few of these groups based on historical demographic processes (conquest and migration), from the time of the Xiongnu through to the Mongol period.

Conclusions

In trying to elucidate questions of origin and relationships to surrounding groups of the people who composed the Xiongnu polity, it is rather apparent the complex nature of group dynamics, historical demographic processes, and biological relationships that define the region of Inner Asia. I have shown in this dissertation the complex population structure of the Xiongnu, with the possibility that, at least for one cemetery in northern Mongolia during the Late Iron Age, a biologically distinct group, who may, or may not, have administered parts of the Xiongnu Empire. In terms of craniofacial diversity, the Xiongnu people were rather heterogeneous. One segment seems to be an outlier, possibly through cultural isolation, while the other segment seems to integrate into and define a continuity of populations that have inhabited modern-day Mongolia for at least the last 2000 years.

The population history of the Xiongnu is as complex as analyses of within-group structure. When compared to regional skeletal samples, it is not surprising that some of the individuals who were a part of the Xiongnu polity to show a clear biological relationship with groups inhabiting northeastern China and parts of what is today Inner Mongolia. This connection has been well documented by archaeologists working the region. Surprisingly, although there are some biological connections to Central Asia, it appears there are less so than to groups in China. This finding is similar to what has been proposed elsewhere, and the similarities to some groups in China as opposed to others,

should help shape further research by bioarchaeologists, physical anthropologists, and molecular biologists.

The relationship to groups in Siberia may be even more complex, due to the nature of the region of South Siberia with the numerous and varied cultures and peoples who have passed through there in the last 3000 to 4000 years. On the basis of craniofacial morphology, the Xiongnu may be connected to the Yakuts, though further analysis is certainly warranted. As for the Bronze Age Chandman sample, although in most analyses they are completely isolated (except showing a relationship to the Egiin Gol and Mongol Turk samples), they do show some similarity, at least on axes of lesser variation, with the Pazyryk nomads of the Altai.

All of these findings are preliminary. Of course, greater sample sizes are needed, in addition to new methods and hypotheses to be tested. The craniofacial traits represented here may only account for some of the variation seen in these groups. In addition, more analyses using ancient DNA may help clarify issues of origin and demography. For now, it appears the Xiongnu have been more exposed to the analyses of biology and biological anthropology, and we now know more than we did before we started about the origin of this incipient steppe polity who ruled over vast parts of Inner Asia during the Bronze and Iron Ages.

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