New Developments in the Origins and Evolution of Native American Populations

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The history and origins of the earliest Americans have commanded the interest of anthropologists for many years. The following factors stimulate this interest. North and South America were the last large uninhabited landmasses that ice age humans settled. The two continents span all latitudes, harbor all ecological zones, and contain many natural barriers to the free movement of people. Despite the barriers to free movement, archaeology shows that Native American people mastered these challenges and filled the landscape in a narrow time window. By the time Christopher Columbus arrived, Native Americans possessed a rich diversity of cultures and languages that likely arose in situ during the course of population growth and adaptation to new environments. Genetics and physical anthropology have played prominent roles in studies of the origin and evolution of Native American populations (Spuhler: Genetic, linguistic, and geographical distances in native North Americans. In: Huizinga J, editor. The assessment of population affinities in man. Oxford: Clarendon Press, 1979; Crawford: The origins of native americans: evidence from anthropological genetics. New York: Cambridge University Press, 1998; Salzano and Bortolini: The evolution and genetics of Latin American populations. New York: Cambridge University Press, 2002).

The following six articles in this issue present the leading edge of genetic studies. Each paper was presented in a symposium entitled ‘New Developments in the Origins and Evolution of Native American Populations’ that was held at the 79th annual meeting of the American Association of Physical Anthropologists, held in Albuquerque, NM, April 15, 2010. The symposium was dedicated to Francisco M. Salzano who has been a prominent leader in the field for over 50 years.

HISTORICAL OVERVIEW

In the 19th and early 20th century, anthropologists connected Native Americans with Asian ancestors by comparing physical traits. The framework for this research was primarily race and taxonomy, with the goal of classifying people into descent groups (Powell, 2005). Geneticists moved studies of Native Americans forward in the 1960s and 1970s when technologies enabled electronic computations and the genotyping of a modest number of blood groups, serum proteins, and red cell enzymes. With the genetic data, the scientific questions shifted from taxonomy and race to population process. Many studies emphasized the balance between gene flow and random genetic drift among villages and local residential units within tribes (Neel and Salzano, 1964, 1967; Niswander et al., 1970; Neel, 1972). Over the past three decades, anthropological geneticists have focused their studies on mitochondrial DNA (mtDNA) and the Y chromosome. Phylogeography became the dominant organizing principle. A good deal of attention has gone to several questions related to the original peopling of the Americas. How many prehistoric migrations into the Americas were there? When did they occur? Where did the ancestral populations reside before entering the Americas?

The first major finding from mtDNA was that most Native American mtDNA haplotypes belong to one of four ancestral lineages (labeled A, B, C, and D) and that members of these lineages are found widely throughout the Americas (Schurr et al., 1990; Torroni et al., 1992). These mtDNA lineages are present sporadically throughout populations in Asia. They occur together in populations in Mongolia, Tibet, and Siberia (Kolman et al., 1996). This distribution suggests that Northern Asian populations are the most closely related to Native Americans. By the mid-1990s, researchers proposed that a single migration from Asia into the Americas could parsimoniously explain the distribution of the four principal mtDNA lineages and should be the favored scenario for the peopling of the Americas (Merriwether et al., 1995; Bonatto and Salzano, 1997). Studies of Y chromosome DNA came soon after mtDNA studies and provided a source of independent genetic information about the peopling of the Americas. Researchers identified two principal founding lineages, designated C and Q, which are ubiquitous among Native American men. Similar to the situation with mitochondrial lineages, the common Y chromosome lineages in the Americas appear with high

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frequencies in northern Asian people. Like mtDNA, the Y chromosome data are consistent with a single migration of people into the Americas. mtDNA and Y chromosomes indicate an entry date that is in the range 15,000–18,000 years ago (Mulligan et al., 2004; Zegura et al., 2004).

Until recently, geneticists had not conducted a broad survey of autosomal DNA variation in Native Americans. Several small studies including HLA haplotypes, short tandem repeat loci (STR), and Alu polymorphisms supported the recent single-wave interpretation of the peopling of the New World (Mulligan et al., 2004). The first large-scale study of autosomal diversity in Native Americans analyzed a large battery of STR loci (Wang et al., 2007). The authors confirmed the gradient of decreasing genetic diversity as a function of geographic distance from the Bering Strait, which increases the genetic distance from Siberians. This pattern reflects the southward dispersal of Native Americans from the northwestern tip of the Americas (Bortolini et al., 2002). The large scale study of STR polymorphisms also found novel evidence that supports the theory that migrating peoples traversed the continents using coastal routes rather than inland routes.

NEW DEVELOPMENTS

The six following papers present recent research on the origins and evolution of Native American populations. It is worthwhile to point out some of the new data that they present and the directions that they forge. Direct observations on DNA form the basis of all genetic marker analyses that they report. Two papers add to the large body of research on Y-chromosomes and mtDNA. Santos et al. find a new Y-chromosome marker that is useful for deciphering the local history of Andean populations. Raff et al. analyze an amazing compilation of mtDNA extracted from ancient archaeological materials (aDNA). Three papers focus on high density autosomal polymorphisms. The same set of STR polymorphisms are the materials in two papers, by Hunley and Healy on genetic admixture, and Ramachandran and Rosenberg on geo-ecological structure (Wang et al., 2007; Wang et al., 2008). These STR data are now the common denominator of many studies on human population structure and history throughout the world (Cann et al., 2002; Rosenberg et al., 2002, 2005; Wang et al., 2007). Kidd et al. present extensive new data on single nucleotide polymorphisms, both individually and as markers of haplotype systems. Finally, de Azevedo et al. depart from the study of genetic markers by focusing their efforts on cranial morphology. Their interpretations are evolutionary, and they advocate an integrative model to explain the evolution of both morphology and genes.

To illustrate the new insights from these papers, it is useful for us to introduce a framework of five historical phases in the history of American Indian populations. Each phase will have had an effect on the architecture of Native American genetic diversity. First, American Indians possess many polymorphisms that occur in all human populations. These polymorphisms are ancient and arose before the geographic expansion of our species and the formation of continental and local populations. Second, founder effects that occurred as modern humans migrated out of Africa and filled the remaining habitable continents had a profound effect on all non-African people, including the Native Americans (Ramachandran et al., 2005). By virtue of the founder effect process, Native Americans share small founder pools with the original out-of-Africa migrants and the first modern human migrants into Asia. Third, American Indians descend from a small set of founders who lived at, or just before, the time that they migrated to the Americas (Tamm et al., 2007; Kitchen et al., 2008; Mulligan et al., 2008). Fourth, the processes that led to the development of new cultures and languages most likely restricted genetic contact among different American Indian populations. This increased genetic differentiation among groups (Spuhler, 1972; Spuhler, 1979; Salzano et al., 1986; Hunley and Long, 2005; Hunley et al., 2007; Cabana et al., 2008). Fifth, European contact caused a nearly total population collapse; entire groups were lost, formerly distinct populations amalgamated, and genetic admixture with non-Indians occurred (Mulligan et al., 2004). Each phase in the historical saga that culminated in the Native American people superimposed its effects on the effects of the earlier phases. This challenges analysis because each event had the potential to deform or erase the genetic signature of earlier events.

The study by Judith Kidd and collaborators achieves considerable breadth because it addresses both the relationships among Native American populations and the relationships between Native Americans and populations living in Europe, African, and the Pacific Islands. This intra- and intercontinental design can potentially pick up signals from each of the five historical phases that shaped Native American genetic diversity. The impressive set of sampled populations and DNA markers in this study reproduced some findings from earlier large scale genetic studies, but also provided new conclusions. The most notable findings from this study confirm some of the canonical findings that have emerged in recent years. First, there is a general North to South reduction in genetic diversity, which agrees with the probable migration patterns. Second, Native American populations are more closely related to Central Asian populations than to East Asian populations. Third, the ancestral gene pool underlying Native American populations diverged early from contemporary Eurasians. Finally, a variety of different statistical analyses performed by Kidd and collaborators failed to find clear patterns of relationships among Native American populations. A few outlier populations dominated these analyses; each of these populations is a well-known genetic isolate.

de Azevedo et al. tested competing models about the initial peopling of the Americas using data from craniofacial morphology. Their study focuses on the interface between the third and fourth historical phases that shaped genetic diversity in Native Americans. The best explanation of their data came from a model that postulates a single migration into the Americas followed by recurrent gene flow among Asian and the American populations in the Arctic region. Like the finding of Kidd et al., de Azevedo et al.'s craniofacial analyses raise the possibility of substantial in situ evolution in Beringia before the Native American ancestors spread throughout the American continents.

Raff et al. focus on the evolution of Native American populations in the fourth phase that shaped diversity in Native Americans; the period between the initial peopling of the Americas and European contact in the fifteenth century. Their analysis of aDNA alleviated the need for them to filter postcontact effects from their data. Of course, aDNA presents its own analytical challenges. Most notably, the sample sizes vary widely
among groups, and it is unclear how samples that are diachronic in origin relate to either living or past populations. Nonetheless, the authors draw judicious conclusions. They find compelling evidence that Native American populations genetically differentiated soon after they entered into the Americas. They see overall continuity of genetic populations within regions, with a few exceptions that may reflect the genetic signatures of large-scale population movements in pre-Columbian times.

Hunley and Healy evaluate Native American genetic diversity at the fourth and fifth historical phases in American evolution. In particular, they look at the impact of European admixture on making inferences regarding founder effects and gene flow in the pre-European contact era. Their baseline hypothesis is that the serial founder effects model for the global populations will explain Native American diversity as a geographic extreme. From this starting point, they seek to find the effects of European admixture, and specifically to determine whether the overlay of European admixture has wiped out earlier signals of Native American population structure. They find that European ancestry in Native Americans follows a North to South gradient. Since Europeans harbor more genetic diversity than Native Americans, this pattern of admixture has created a gradient in genetic diversity that could be easily mistaken for serial founder effects as the initial migrants in the Americas radiated South from their entry point in the far Northwest. Nonetheless, Hunley and Healy find that admixture has not altered a hierarchical structure of diversity, and that local gene flow, between neighboring groups has had surprisingly little impact on macro-geographic patterns of genetic diversity in the Americas.

Ramachandran and Rosenberg contribute a highly original article. They find a pattern of greater genetic differentiation in Native American populations along lines of longitude than that in Eurasian populations along lines of latitude. This pattern supports a proposition by Jared Diamond (Diamond, 1997). Namely, the orientation of continents influenced the movement of people, and ultimately the transmission of genes and cultural traits. Their conclusion applies to the fourth historical phase in the evolution of genetic diversity in Native American populations. A standout feature of this article is the introduction of new methodology to find clines in genetic diversity based on waypoints in migration, and rotations of geographical coordinates. This sophisticated methodology is likely to find many applications future anthropological genetics studies.

**FRANCISCO M. SALZANO**  
Professor Francisco M. Salzano has been one of the most prolific contributors to the study of the origins and evolution of Native American people. To summarize his many published articles and books would exceed the scope of this brief introduction. Yet, it is important to point to a few of his highest impact contributions. First, Dr. Salzano conducted some of the first studies on human population genetics that reflected the influence of population thinking and the modern evolutionary synthesis of the 1940s. In doing so, he pulled a biologist’s perspective into the domain of physical anthropology. He presented this approach in his earliest studies on the Caingang Indians of southern Brazil (Salzano, 1961a,b,c). The structure of these papers shows the profound influence of T. Dobzansky’s famous Genetics of Natural Populations series of papers. Dr. Salzano and several other investigators used a similar format in studies of many other Native American tribes (Neel and Salzano, 1964, 1967; Neel et al., 1964; Niswander et al., 1970; Workman and Niswander, 1970; Neel, 1972). Second, Dr. Salzano and his collaborators were among the first to analyze the genetic consequences of fluidity in preindustrial human demographic and social structure. One of their cogent observations was that local populations may appear endogamous at any specific moment, but this appearance is deceptive because fissions and fusions govern their long term dynamics. Moreover, groups of related individuals form the units of diffusion among local groups. Kin-structured migration subsequently became a basic component of human genetic demography (Fix, 1978, 2004; Smouse et al., 1981; Rogers, 1987; Hunley et al., 2008). Another ground breaking conclusion from Dr. Salzano’s studies on Native American genetic diversity was that the amount of genetic variability in a small inbred tribe comes close to that in contemporary urban populations (Neel and Salzano, 1967). This observation was later to become the lynchpin in Richard Lewontin’s argument against the significance of race in human genetics and taxonomy (Lewontin, 1972).

Third, Dr. Salzano and his colleagues in South America performed a prodigious amount of field work. They collected data from more than 20 Native American populations (Salzano and Callegari-Jacques, 1988). While they published their own interpretations of these data, they made their samples and results available to other researchers. Our current understanding of the origins and evolution of Native American Populations owes much to these data collections, as will all future investigations.

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**LITERATURE CITED**


The new study shows that the remains found there belonged to members of a previously unknown Native American population, whom academics have named “Ancient Beringians”. Credit: Ben Potter. Direct genetic traces of the earliest Native Americans have been identified for the first time in a new study. The genetic evidence suggests that people may have entered the continent in a single migratory wave, perhaps arriving more than 20,000 years ago. The findings are being published in the journal Nature and present possible answers to a series of long-standing questions about how the Americas were first populated. There's a New Theory About Native Americans' Origins. An Eskimo harpoons a whale in the Bering Sea off Alaskan shores. Chlaus Lotscher / Getty Images. By Tanya Basu. The researchers looked at sequenced DNA from bones as well as the sequenced genomes of Native American volunteers with heritage from not only the Americas but also Siberia and Oceania, says according to Rasmus Nielsen, a computational geneticist at the University of California, Berkeley, and one of the authors of the study. The researchers contacted people whose heritage indicated they were of Amerindian or Athabascan—the two ethnic derivations of Native Americans’ descent. Specifically, they looked at their mitochondrial DNA (mDNA), which is passed from mother to child.