Genetic Analysis and the Peopling of the New World

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Genetic analysis has become increasingly utilized to describe the prehistoric populations that archaeologists attempt to understand. As specific genetic markers are analyzed, genes begin to outline population histories with detailed precision. Analysis of both mitochondrial DNA (mtDNA) and Y-chromosomes, for example, has produced a wealth of information regarding the peopling of the New World, a highly debated issue. These methods have been employed to identify the source population of the initial migration, in addition to shedding light on the timing and number of migrations implicated by modern genetic variation. Interestingly enough, much of the mtDNA and Y-chromosome analyses support contrastive interpretations of the peopling of the Americas regarding the timing and number of migrations. Using the genetic analyses applied to the peopling of the Americas as data, it is necessary to examine the maternally and paternally based lines of research in concert with the archaeological data to provide an accurate model of the peopling of the New World.

There are a few major points of contention in archaeological discussions of the peopling of the New World. Researchers disagree on the when the initial migration occurred and whether population of the Americas involved a single multiple migrations. In addition, various models have been developed to demonstrate how the migration(s) actually occurred. The identity of the source population of the initial migrants as Siberian is highly probable and widely accepted. The specific region from which the migration stemmed, though, is left mainly for DNA analysis to discern.

Sometimes called the “Blitzkrieg” model (see Figure 1), the traditional viewpoint of the peopling of the Americas assumes a single migration following big game across the Bering Strait into Alaska around 11,000 BP. This model conventionally views the
extinction of megafauna as a result of migrating peoples expanding into a totally
unoccupied area and mainly depending on unaware megafauna for subsistence (Martin,
1973). These migrants would have traveled through an ice-free corridor near Alberta that
separated the continental Cordilleran and Laurentide glaciers, essentially funnelling early
populations into the US. As population expansion and migration is likely to avoid high-
cost pathways in favor of less strenuous routes, computer simulation of migration based
on geographic and climatic factors channeled early migrants into the American Midwest,
from which they supposedly spread to the east, west and south (Anderson and Gillam,
2000). As this expansion led migrants into environments with untapped natural resources,
populations would not have been limited by carrying capacity. As a result, this model
requires a rapid wave of migration paired with a sudden population increase. The Clovis
projectile point, often considered a diagnostic artifact of this initial migration, is a large
lanceolate, with usually parallel sides. Fluted with an incurvate base, it sometimes has
ground basal edges (Cambron and Hulse, 1990). Corrected radiocarbon dates for Clovis
of 13,400 BP and that the Clovis projectile point appears almost contemporaneously in
North and South America raises a few issues about population expansion and migration
times (Fiedel, 1999). Whether the Clovis distribution marks the expansion of a people or
the expansion of a technology throughout an already extant population is of key
importance. If the Clovis distribution marks diffusion of technology throughout an
existing population, the time frame for migration must be adjusted to allow populations to
reach South America. Additionally, Pre-Clovis sites in North and South America have
been excavated and yielded in reliable radiocarbon dates that require an earlier entry.
Monte Verde, Chile, for example, has reliable $^{14}$C dates around 12,500 BP, calling for a
new interpretation of migration (Dillehay, 1997). Simulations of different expansion strategies following the ice-corridor migration model suggest that the hemisphere could have been populated in around 1,700 years, requiring an early entry between around 13,000 – 14,700 BP. A “leap-frogging” process in which population growth results in balkanization of social groups presents a likely scenario for this rapid expansion. According to this notion, as population growth continued in settled areas, other groups pushed out of their original settlements by overpopulation would have “leap-frogged” past other groups, moving beyond settled areas. This would have maintained migration pathways southward and towards the periphery (Anderson and Gillam, 2000)

The ice-corridor route has been widely accepted but as criticisms are levied at this traditional model, though, other feasible alternatives have begun garner support. In response to data suggesting that the ice-free corridor may not have been as easily traversed as the Blitzkrieg model assumes, a coastal route was proposed by Fladmark (1979). Populations would have effectively “leap-frogged” down the Western coast of the Americas, permitting a rapid expansion while depending upon more conservative rates of population growth than the ice-corridor route (Fix 2002). A coastal route would allow for an earlier migration of at least 13,500 BP since it would not be drastically affected by climate changes, namely the geographic expanse of continental glaciers. Potential archaeological sites along this coastal route would now lie well below sea level due to warming trends since the Pleistocene, lessening the chance of tracing the route southward. Some question this model based on the fact that no direct technological evidence has been found insinuating seafaring capabilities. Australia was occupied, though, at least 40,000 years ago by seafaring individuals, rendering a long-distance sea
voyage at least possible. Also, given the big game focus of the traditional model, lithic analysis would have been dominated by a concentration on large bifacial projectile points used to hunt terrestrial game rather than smaller utensils like blades and burins that may have proved useful in maritime settings (Fladmark 1979). This claim has been echoed by some researching coastal areas in the early Holocene (Cassidy et al., 2004) but this argument is not considered highly viable here. The coastal route is a viable alternative to traditional model that deserves consideration.

Linguistic data are also utilized to help integrate archaeological data and DNA testing. Greenberg et al. (1986) classified the indigenous languages of North and South America relative to dental and genetic evidence. Their analysis resulted in a tripartite division that separated indigenous languages into Amerind, Na-Dene, and Aleut-Eskimo, representing three migrations into the Americas. The Amerind language family corresponds to the first migration based on its high internal linguistic diversity and the fact that it is centered in South America. The second migration spread the Na-Dene family from Northwest Canada and Alaska to California, Oregon, and the American Southwest. Even with a less peripheral distribution than the Amerind family, the Na-Dene family still has more internal differentiation. The third and most recent migration consisted of the Aleut-Eskimo language family and is distributed throughout Alaska and Northwest Canada. Looking at genetic and dental data as secondary to linguistic analysis, the Amerind migration was estimated around 11,000 BP, the Na-Dene around 9,000 BP, and between 2,900 and 5,600 BP for Aleut-Eskimo. The Aleut-Eskimo migration is too recent and hence is not deemed applicable for discussion of the peopling of the New World. This classification embodied an integration of the research of multiple fields,
offering a holistic view. Though their linguistic classification was controversial and their
discussion of genetic data is antiquated, their work was taken as a type of working
hypothesis against which all DNA analyses have been compared.

Mitochondrial DNA is a circular DNA molecule consisting of 16,569 known base
Pairs (see Figure 2). It is not recombinant, meaning that it does not involve a meiotic
union of paternal and maternal genes, solely reflecting the maternal lineage. mtDNA also
mutates 5-10 times faster rate than nuclear DNA, making it a useful tool for analyzing
population histories (Bailliet et al., 1994). Recovery of mtDNA from prehistoric
specimens is relatively unproblematic and retrieval from living populations is
uncomplicated, making the technique more applicable. As the mutations are identified
and traced throughout a population, a picture of prehistoric genetic variation develops,
illuminating the evolutionary history of a population based on haplogroups. A haplogroup
is defined on the basis of a specific mutation that is well established and widely
distributed among individuals of a population. These identified haplogroups can then be
divided further into haplotypes, usually based on restriction fragment length
polymorphisms (RFLP). RFLP’s are a result of restriction enzymes that cut DNA
sequences at specific points, priming them for electrophoresis and allowing a comparison
to be made showing patterns among individuals based on the length of the DNA
fragments (Eshleman et al., 2003). When prehistoric mtDNA is compared to analysis of
living populations, a genetic distance can be ascertained between prehistoric and living
populations based on mitochondrial variation and comparative frequencies within
populations. These comparisons rely on a certain degree of cultural continuity within
regions and assume no admixture with genetically different populations, like Europeans.
Though it is a reliable testing procedure, there are some limitations to mtDNA analysis. Genetic variability of populations over a number of generations is mathematically assessed and hence dependent upon making certain assumptions about population size, gene flow, and gene drift, all of which can be difficult if not impossible to evaluate in prehistoric populations. In addition, the degree of gene flow between living Amerindians and Europeans adds a complication to establishing haplotype frequencies. The extent to which this affects results depends entirely on the researchers’ sample choice, meaning that they are responsible for sampling from appropriate indigenous populations that show a history of genetic isolation from other unrelated groups. Assuming modern populations are pristine without any intermingling of foreign genes may be, at times, naïve, but it is the researcher’s duty to select the appropriate sample. Whereas contamination of prehistoric samples makes the results totally obsolete, cautiously conducted laboratory analysis eliminates this possibility (Kaestle and Horsburgh, 2002). One limitation inherent in mtDNA analysis is not due to faulty samples or methodology but the DNA itself. Only the maternal line is traceable through this method, palatably excluding 50% of the population and not being able to recognize social organizations that would reflect dissimilar pathways for men and women. There is always the possibility that lineages have become extinct and would not show up in modern populations, which raises the issue of the legitimacy of comparing ancient and modern populations. It should be noted that though the introduction of sources of error is possible, it does not necessarily unconditionally invalidate the conclusions drawn from mtDNA analysis.

The mutations common in mtDNA of prehistoric Native American populations
have been analyzed and 5 major haplogroups, or clades, of haplotypes have been established, resulting in haplotypes A, B, C, D, and X. The haplotypes have different geographic distributions in extant Amerindian populations, marked by either high concentrations or clinally distributed frequencies. Classified by a mutation at nucleotide position (np) 16111 involving a C→T transition and including HaeIII at np 633, Haplogroup A has high frequencies in Canada, the Eastern US, and central Mexico. Haplogroup B exhibits high frequencies in the Western and Midwestern US and is identified by a 9 base pair deletion. Uniformly distributed throughout NA aside from a decrease in Alaska, Haplogroup C is typified by an addition of AccI at np 13262 and a deletion of NichII at np 13259. Haplogroup D is characterized by a deletion of AluO at np 5176 and has higher frequencies in Alaska paired in consistently lower frequencies in the rest of NA. These four haplogroups were thought to represent all haplogroup variation until haplogroup X was defined in both modern and ancient populations (Eshleman et al., 2003; Smith et al., 1999; Malhi and Smith, 2002). Involving a G→A mutation at np 16,213 and an addition of AccI at np 14465, haplogroup X is found in high concentrations in the Great Lakes and Greenland with moderately lower frequencies elsewhere, though the high concentration in Greenland may be a function of the chosen sample and methodology of analysis (Malhi et al., 2002). The varying geographic distributions of the haplogroups is best considered as a result of isolation by distance because research suggests that intratribal genetic homogeneity is greater than intraregional genetic homogeneity, evidence which some view as indicative of an early tribalization in prehistoric populations (Lorenz and Smith 1996). This model requires groups to aggregate relatively soon after initial migration(s) and assumes that later tribalization
would have resulted in greater intraregional diversity as increased gene flow occurred among populations pre-tribalization. In some studies (Malhi et al., 2002) haplogroups A, C, and X demonstrated higher estimates of diversity than B or D, whereas others (Lorenz and Smith, 1996) report that genetic diversity of haplogroups B and D are comparable with A, C, and X. Genetic diversity in the southeast is markedly smaller than any other geographic region, representing the devastating repercussions of Spanish contact. Thirty-six percent of ancient haplotypes are shared with modern Native Americans, evidencing continuity, and 29.6% of haplotypes are shared among modern Native American individuals, supporting the widespread distribution of the haplotypes and their applicability to the study of prehistoric populations (Malhi et al., 2002).

Comparing haplotype distributions and frequencies of both ancient and modern Native Americans with frequencies and distribution on a global scale begins to shed some light on the identity of the founding populations of the New World. Migration models support a founding population from the vicinity of Siberia so, ideally, shared haplotypes should be found in both societies. Initially, this wasn’t the case as all haplogroups excluding X were found to exist in modern Siberian populations. Haplogroups A- D found in Siberia supported a Siberian founding population but categorized haplogroup X as an anomaly (Uinuk-Ool et al., 2003). Haplogroup X is also present in European foundations, further confounding the matter as some began investigating the possibility of an early European migration (Brown et al., 1998). Eventually, mtDNA analysis confirmed that haplogroup X was present in Asia, specifically the Altai area in Southern Siberia, located to the south of Lake Baikal. The Altains were found to exhibit all five haplogroups, making them a very likely founding population source. In addition, the
European haplotype X differs from the Native American, and the Altains seem to represent an intermediary form between the two different mutations, possibly hinting at a western migration of European peoples followed by a much later migration of Altai peoples to the Americas (Derenko et al. 2001). However, further analysis of the X haplotypes now insinuates that the Altains exhibit recent acquisition of a unique mutation that characterizes Amerindians, leading some to consider alternate staging areas. An earlier Near Eastern origin for this mutation has been proposed, eventually filtering into founding populations near the Altai region (Reidla et al., 2003). Paired with the notion that now-extinct founding lineages may explain some of the “leftover” variation seen in modern Amerindian populations, the outline of a clear-cut genetic history based on mtDNA is problematic.

The diversity of haplogroups is recognized as either representing anywhere from one to four migrations. Hypothetically, multiple migrations should result in geographically distinct areas and varying levels of genetic variability within haplogroups relative to timing of migration. A single migration should be reflected in haplogroup distributions and frequencies that extend beyond linguistic or geographic borders. Paired with early tribalization, it would result in more intratribal than intraregional genetic diversity (Lorenz and Smith, 1996). Most mtDNA analysts seem to advocate a single west-to-east migration that contained all of the haplotypes currently found in Native American populations based on intraregional haplotype frequencies and the clinal distribution of haplogroups A, B, and X (see Figures 3-5) (Malhi et al., 2002). A distinction should be made, though, between initial migration and population expansions, both of which can account for similar geographic distributions. Supposed lower levels of
genetic diversity, as some have suggested for haplogroup B, might indicate a recent expansion, not necessarily a second migratory wave. The timeframe of migration based on mtDNA ranges from 40,000 – 11,500 BP. An important feature of many of the mtDNA time estimates is that they mark divergence from an Old World lineage, not necessarily a migration to the New World, overestimating the timeframe. With this in mind, an early migration estimate based on mtDNA analysis would provide a conservative entry date of at least 12,000 BP with Clovis marking the expansion of a migrating group (Eshleman et al., 2003; Reidla et al., 2003). Others supporting multiple migrations seem to suggest an initial pre-Clovis entry between 18,000 – 15,000 BP with a subsequent migration occurring around 12,550 BP (Schurr and Sherry, 2004), accounting for genetic variability, early Pre-Clovis South American sites, and even the varying geographic distribution of haplogroups.

Y-chromosome analysis has also been used to investigate the peopling of the New World. The Y-chromosome is one of two chromosomes that define sex during meiotic recombination. Though recombination occurs at some points on the chromosome, there is also a non-recombinant Y-chromosome (NRY) portion that is valuable when outlining evolutionary population histories. Similar to mtDNA, analyzing the successive mutations on the non-recombinant portion will result in definition of haplogroups and haplotypes. Distinguishing the haplotypes often relies on identifying and differentiating single nucleotide polymorphisms (SNP) in an effort to gauge variation, another key aspect of NRY. NRY analysis is worthwhile due to its non-recombinant status and the fact that it traces evolutionary linkage through the paternal line, presenting a different model of migration than mtDNA analysis, which focuses on the maternal line. In some cases it
could offer a contrast between evolutionary histories of males and females of a given population though in this case its use is geared towards establishing a timeframe, source population, and number of migrations.

Like any form of genetic analysis, there are certain limitations to NRY that must be understood in order to appropriately assess the results. It only presents data on the paternally inherited traits, and hence must be analyzed within a larger framework that considers other analyses. The accuracy of NRY analysis is very much a function of having the right sample size and choosing the right sample. As with the mtDNA, accurate NRY interpretation relies on the pristine nature of indigenous populations, forgoing the possibility of genetic admixture. This can lead to a misinterpretation of the results, especially considering male-based inheritance as immigrant men tend to mate with native women (Bortolini et al., 2003). In addition, NRY relies heavily on mathematical calculations of lifespan and population increase to generate an accurate evolutionary timeframe, which may or may not include accurate mathematical assumptions. NRY is a valid means to test evolutionary histories of populations, but, like any genetic analysis, raises questions about the validity of having to make a number of assumptions about demographics paired with assuming continuity between modern and ancient populations.

There are three major haplogroups considered in NRY analysis in the Americas. One haplogroup, P-M45, can actually be broken down into two haplotypes, M45a and M45b, which have varying geographic distributions: M45a is distributed throughout the Americas while M45b seems to appear in only North America. Together, they account for 29% of Native American Y-chromosomes. The Q-M3 haplogroup, the most frequent in Native American populations, is a derived version of P-M45. It is also the most widely
dispersed haplotype, accounting for 66% of all Native American Y-chromosomes (Lell et al., 2002) and occurring in a north-south oriented cline with increased frequencies in South America (Schurr and Sherry, 2004). The most recently discovered haplogroup is Q-M242, another derivative of P-M45 that diverged before the Q-M3 split, probably more accurately reflecting an appropriate timeframe for initial migration (Seielstad et al., 2003). It occurs throughout Native American populations, with higher frequencies in South America (Bortolini et al., 2003). The geographic distribution of the three haplogroups, then, reveals a slightly different pattern for North and South America. Part of this discrepancy is due to smaller sample sizes in some of the studies considered here, which in some cases would tend to over represent South American frequencies at the expense of North American populations (Schurr and Sherry, 2004).

The source population estimates based on the haplotype frequencies and distributions of modern populations mostly advocates a Central Asian or Siberian emigration. Q-M3 is found only on the Chukotka peninsula of Siberia, a possible staging area for the migration (Lell et al., 2002). The global distribution of P-M45 haplotypes reflects a divergent history, linking P-M45a with Central Siberian populations and P-M45b with southern and eastern Siberian populations (Schurr and Sherry, 2004). P-M45b is interesting because it also is scattered throughout Europe, raising questions about possibly erroneous judgment of the genetic “purity” of the indigenous sample (Lell et al., 2002). In addition, the fact that P-M45b is one of the most frequent haplotypes in Europe led some scholars to question whether this expansive dispersal is a function of genetic admixture with Europeans throughout recent history and not characteristic of founding populations at all (Tarazona-Santos and Santos, 2002). Though recognition of this
possibility is key to interpretation, so is the fact that P-M45b is present in Siberian populations. A parsimonious solution in alignment with the rest of the genetic data would rely on an emigration of Siberian peoples without incorporation of European Y-chromosomes. The Q-M242 haplogroup is found in Central Asia where it apparently diverged pre-migration from its parental P-M45 haplogroup. Taken collectively, Y-chromosome analysis supports Middle and eastern Siberia as possible source populations (Bortolini et al. 2003).

Whereas some researchers support a single migration, Y-chromosome analysis mostly supports two major male migrations into North America based on NRY distributions in modern populations. The genetic heterogeneity or homogeneity of the founding population is dependent upon model choice. The multiple migration model was proposed by Lell at al.(2002) to correlate the haplogroup divergence estimates with the geographic distribution. As originally outlined, the initial migration stemming from southern Middle Siberia would have introduced haplogroups P-M45, Q-M242, and Q-M3 around 20,000 – 30,000 BP. Discovery of the haplotype Q-M242, the ancestral form of Q-M3, resulted in a revised upper limit of 15,000 to 18,000 BP based on reconsideration of population size and population growth rates (Seielstad et al., 2003). The second migration bringing M45b and RPS4Y-T, a minor haplotype, would have consisted mainly eastern Siberians, occurring around 7,000 – 9,500 BP. The single migration model centers on the P-M45b haplotype, which Tarazona-Santos and Santos (2002) view as attributable to European admixture and hence irrelevant to the peopling of the Americas. This migration would have stemmed from the central southern Siberia and included all of the haplotypes.
Though researchers are starting to integrate mtDNA and Y-chromosome analysis, much of the research was conducted and hence interpreted independently. Considering that both genetic analyses present varying possibilities, an integration of migration models, timing and number of migrations, and identification of the source population is absolutely necessary. The source population can be considered first. Both mtDNA and NRY analysis place founding populations in south-central Siberia near the surrounding areas of Lake Baikal and the Altai region. All of the haplotypes of both mtDNA and NRY have been found south-central Siberia and though the proposed regional affinities may differ on the matter of a few kilometers, the populations are geographically close enough in proximity to constitute a breeding population.

Two migrations from Siberia would sufficiently account for both genetic variability in the Americas and the pre-Clovis presence of archaeological sites in the New World. An earlier coastal migration consisting of populations from near south-central Siberia (near Lake Baikal) around 18,000 - 15,000 BP could possibly have brought mtDNA haplogroups A – D and NRY haplogroups P-M45a, Q-M242, and Q-M3 (Schurr and Sherry 2004). This could correspond to Greenberg et al.’s (1986) migration of the Amerind peoples. Terrestrial passage through the Cordilleran and Laurentide glaciers would have been nearly impossible at this point, necessitating an alternate route. An initial coastal route was favored over the land route because it would release migrant populations from demanding rapidly increasing population while allowing more than enough time to reach South America. Bands are organized into relatively small, mobile groups whose lifestyle does not exactly facilitate extremely rapid population expansion. Many of the computer simulations (Anderson and Gillam, 2000) following an ice-
corridor route actually impose population growth that exceeds any rates seen in observation of modern band-level societies. Observations that modern band-level societies rarely approach carrying capacity due to a pattern of resource density-dependent dispersal would not necessarily insinuate a rapidly increasing population expansion would, not supporting the Blitzkrieg land route (Steele et al., 1998). Computer simulations replicating dispersal rates suggest that an initial land route would result in a severe bottleneck effect, depleting genetic variation within around 30 generations (Fix, 2002). This would have effectively made early land-route migrants doomed to genetic a homogeneity that is simply not supported by either mtDNA or Y-chromosome analysis, which shows considerable variation. Early tribalization as suggested by Lorenz and Smith (1996) would fit into this model, as a coastal technology would also allow for considerable gene flow and diffusion of technology between populations as migrants would be able to interact with groups beyond their immediate inhabitation area. This could account for the later expansion of Clovis technology throughout an already extant population. A second migration from southeastern Siberia (the Altai region) occurring around 11,000 BP could have brought the remaining mtDNA haplogroup X, the NRY haplogroup M-45b, and other minor NRY haplogroups like the RPS4Y-T mentioned earlier 9Schurr and Sherry, 2004). Possibly corresponding with Greenberg et al.’s Na-Dene linguistic group, this migration could possibly have exploited the interior ice-free corridor, which would have been passable at this time. This would explain the high concentration of haplogroup X in the Great Plains, which would reflect channeling into the American Midwest.
The interpretation presented here hinged upon integration of different fields and made certain assumptions regarding migration models. Further research will likely continue to integrate mtDNA and Y-chromosome analysis, including novel interpretations as haplotypes are further delineated and the geographic distributions mapped. Hopefully a more satisfying understanding of migration patterns will fill in the gaps regarding a coastal or ice-free corridor route as the Pre-Clovis archaeological becomes more established.
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