

Using Ancient mtDNA to Reconstruct the Population History of Northeastern North America

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ABSTRACT Mitochondrial DNA (mtDNA) was extracted and analyzed from the skeletal remains of 44 individuals, representing four prehistoric populations, and compared to that from two other prehistoric and several contemporary Native American populations to investigate biological relationships and demographic history in northeastern North America. The mtDNA haplogroup frequencies of ancient human remains from the Morse (Red Ocher tradition, 2,700 BP) and Orendorf (Mississippian tradition, 800 BP) sites from the Central Illinois River Valley, and the Great Western Park (Western Basin tradition, 800 BP) and Glacial Kame (2,900 BP) populations from southwestern Ontario,

change over time while maintaining a regional continuity between localities. Haplotype patterns suggest that some ancestors of present day Native Americans in northeastern North America have been in that region for at least 3,000 years but have experienced extensive gene flow throughout time, resulting, at least in part, from a demic expansion of ancestors of modern Algonquian-speaking people. However, genetic drift has also been a significant force, and together with a major population crash after European contact, has altered haplogroup frequencies and caused the loss of many haplotypes. *Am J Phys Anthropol* 137:14–29, 2008. © 2008 Wiley-Liss, Inc.

Many early anthropological studies using mitochondrial DNA (mtDNA) attempted to answer broad questions regarding the homeland of North America's earliest colonizing populations, or the number, timing, and route of migrations to the New World (e.g., Schurr et al., 1990; Torroni et al., 1992, 1993; Shields et al., 1993; Merriwether et al., 1995; Merriwether and Ferrell, 1996). These studies, however, were conducted with a limited understanding of genetic diversity and its geographic and temporal distribution across the Americas. In recent years, regional studies have begun to investigate genetic diversity more extensively (Malhi et al., 2001, 2003, 2004; Bolnick and Smith, 2003; Eshleman et al., 2004; Shook, 2005; Johnson and Lorenz, 2006). These studies have shown that populations within a region, even those not related by language or culture, often exhibit genetic similarity suggesting that local populations have maintained a significant amount of gene flow limiting the influence of genetic drift.

Studies of ancient burial populations can complement regional studies of genetic variation among contemporary Native American populations by establishing how and when these regional patterns developed. Most studies of ancient populations, like those of the Columbia Plateau (Schultz et al., 2001; Malhi et al., 2004), Southwest (Parr et al., 1996; Carlyle et al., 2000), and Northeast (Stone and Stoneking, 1998), demonstrate genetic continuity between prehistoric and extant groups in those areas. However, most of the ancient populations included in these studies are less than 2,000-years-old, leaving unanswered the question of how and when these regional patterns developed. One notable exception is the western Great Basin, where prehistoric populations display a distribution of haplogroup frequencies that significantly differs from that of the populations that now live there, supporting the hypothesis of a prehistoric

intrusion into the region and incomplete replacement by Numic-speaking people (Kaestle and Smith, 2001). A significant level of admixture between Numics and the prior indigenous population probably accompanied the "replacement" of the pre-Numic inhabitants of the western Great Basin (Kaestle and Smith, 2001).

This study examines how mtDNA patterns have changed throughout prehistory and across geographic space within the Northeast by comparing genetic patterns of ancient populations from two localities (the Central Illinois River Valley and southwestern Ontario) at two different time periods (~800 and ~2,800 BP). The Northeast is a roughly defined region bordered on the north by the boreal forest, on the west by the plains, and somewhat arbitrarily on the south by the southern margin of the Ohio watershed (Trigger, 1978; Fig. 1).

This region is an optimal location for studies of ancient DNA in part because its modern populations' mtDNA structures have already been described (Malhi et al., 2001; Shook, 2005), and it is the homeland of the largest ancient population studied to date ($n = 108$; Stone and Stoneking, 1998), the Norris Farms Oneota.

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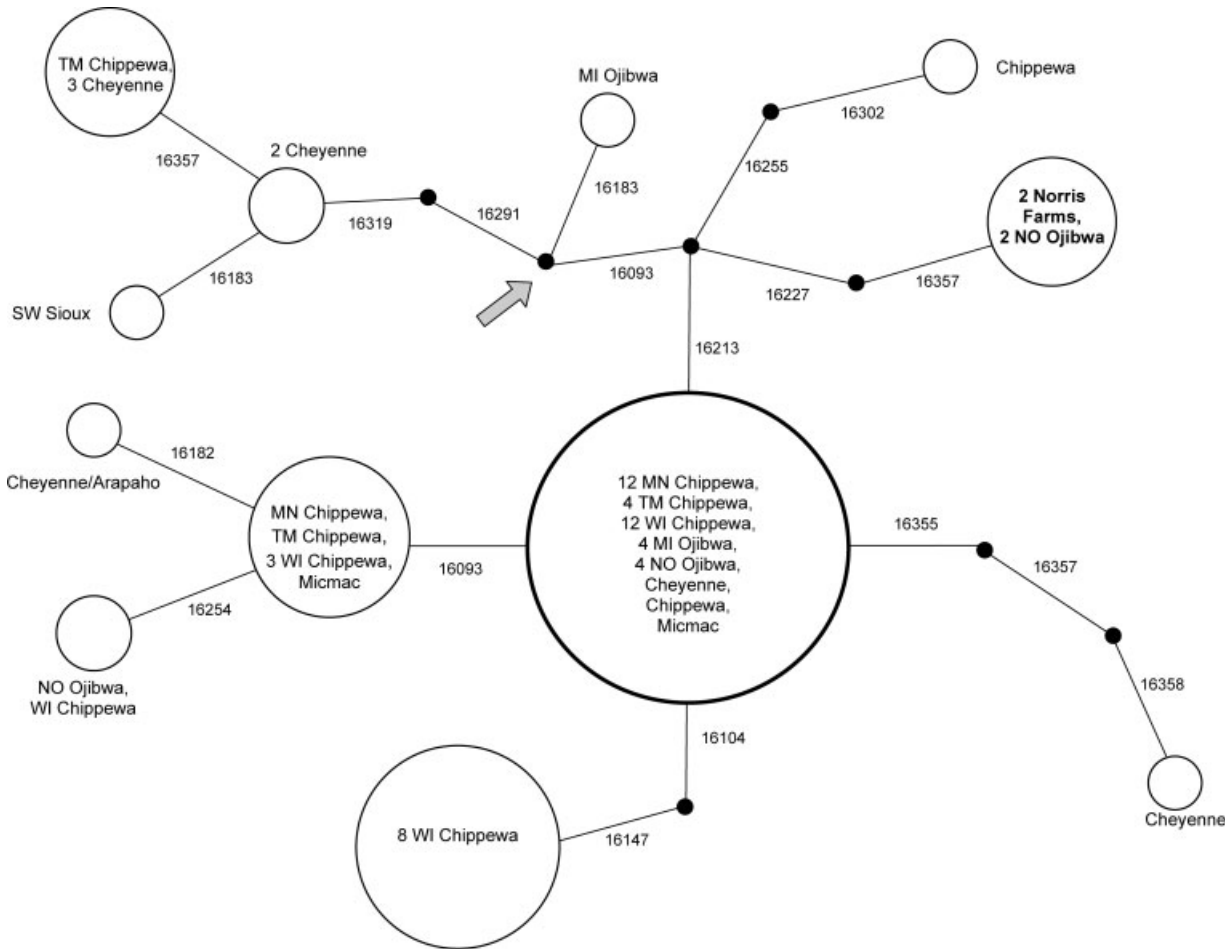


Fig. 7. Haplogroup X haplotype network. One likely founding New World type (Shook, 2005) is marked by a thicker border, and differs from the Anderson et al. (1981) sequence at nps 16093, 16183, 16189, 16213, 16223, and 16278. The founding type suggested by Brown et al. (1998) is not represented by a sample in this study. It is represented, however, by a hypothetical ancestral haplotype indicated by an arrow.

and/or other cultural exchange, might be responsible for the similar haplotypes and haplogroup frequencies across the region. Additionally, this pattern of regional continuity for mtDNA is consistent with patrilocal mating patterns, like those exhibited by most Algonquian and Siouan speakers (Bolnick et al., 2006). Other genetic markers (autosomal or Y chromosome) would need to be employed to fully evaluate the extent of this regional continuity.

Explaining genetic change over time throughout the Northeast

It is known that populations were expanding demographically in the Northeast during the Late Archaic and Woodland periods, potentially in association with an Algonquian language expansion and/or the entry of maize agriculture to the region. Since modern Algonquian populations exhibit high frequencies of haplogroups that became increasingly common in the Northeast throughout the Woodland period (A, C, and X) and low frequencies of haplogroups that became increasingly rare in the same interval (B and D), it is possible that either the linguistic expansion of the Algonquian language family or the spread of maize agriculture not only facili-

tated gene flow, but was accompanied by a demic expansion involving significant levels of admixture with surrounding populations, leading to clinal changes in haplogroup frequencies in addition to the persistence of many ancient haplotypes. This same mechanism, "incomplete population replacement," has been invoked to explain the Numic expansion into the western Great Basin (Kaestle and Smith, 2001) and the absence of genes of Mesopotamian origin at the supposed terminus of the demic expansion that brought the Neolithic Revolution to western Europe (Renfrew, 2002). It is conceivable that the historical influence of this property of migration and population replacement has been underestimated.

Only 19.7% of the 61 ancient haplotypes in the Northeast are shared with extant individuals, somewhat lower than the rate (36%) estimated by Malhi et al. (2002) for all of North America. Additionally, ancient populations exhibit a higher frequency of unique haplotypes than modern populations and much of the diversity of haplogroups B and D have been lost, suggesting that genetic drift has been a significant force over time. As reported for Southeastern populations (Bolnick and Smith, 2003), it is also likely that much of the genetic diversity was lost after European contact when population levels dropped from between two and ten million to approxi-

mately a quarter million by the year 1890 as a result of disease, warfare, and depletion of resources (Snipp, 1989; Calloway, 1997; Jones, 2003).

The increase in frequency of haplogroup X, virtually absent or in very low frequencies in ancient populations throughout the Northeast, is also consistent with a genetic bottleneck after European contact. Haplogroup X has been detected in 3% of individuals from a Middle Woodland Hopewell site in Illinois (1,825–1,625 BP; Bolnick and Smith, 2007) and 6% of the Norris Farms individuals (Stone and Stoneking, 1998). Yet, ~700 years later, it is at extremely high frequencies in some Northeastern populations (50% in Minnesota Chippewa). Haplogroup X does, however, exhibit a relatively high level of haplotype diversity, more than might be expected to accumulate from recent (<100 years of) growth and diversification. Thus it is more likely that haplogroup X was present at higher frequencies in other prehistoric Northeast, or neighboring, populations and has gone undetected.

It is also likely that genetic drift has been a factor throughout the entire study period, not just post-European contact, given that only 15% of the Late Archaic haplotypes are shared with any other prehistoric or contemporary group. Genetic drift in the Northeast may have been fostered by the small size of the earliest populations, significant isolation of populations, and/or a significant historical event, such as warfare, famine, or disease, causing a decline in population size. Recent research using a model to simulate population variation over time suggests that genetic drift can account for significant change in haplogroup frequencies over time even in the presence of gene flow (Cabana, 2002). Thus, the cumulative effect of 3,000 years of low levels of genetic drift could account for a significant amount of evolutionary change in the Northeast.

CONCLUSIONS

The general pattern observed in the Northeast is one of sustained genetic regional continuity, likely due to significant female gene flow, whose genetic patterns shift gradually in relationship to time. Despite changing haplogroup frequencies, haplotype patterns suggest that some ancestors of present day Native Americans in northeastern North America have been in that region for at least 3,000 years. This overall pattern may be associated with a number of factors including trade networks, an Algonquian language (and likely cultural) expansion, the emergence of agriculture, and/or patrilocal mating patterns. Genetic drift has also likely been a significant force, and together with a major population crash after European contact, has caused the loss of many haplotypes. The present study, limited by sample sizes and examination of only one genetic marker, underscores the need for additional genetic analysis of prehistoric populations in the Northeast to help differentiate between these varied causes and to further illuminate the pattern of genetic change in the Northeast.

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