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We Are Not Our Ancestors: Evidence for Discontinuity between Prehistoric and Modern Europeans

Ellen Levy-Coffman

The model of European genetic ancestry has recently shifted away from the Neolithic diffusion model towards an emphasis on autochthonous Paleolithic origins. However, this new paradigm utilizes genetic reconstructions based primarily on contemporary populations and, furthermore, is often promoted without regard to the findings of ancient DNA studies. These ancient DNA studies indicate that contemporary European ancestry is not a living fossil of the Paleolithic maternal deme; rather, demographic events during the Neolithic and post-Neolithic periods appear to have had substantial impact on the European genetic record. In addition, evolutionary processes, including genetic drift, adaptive selection and disease susceptibility, may have altered the patterns of maternal lineage frequency and distribution in existing populations. As a result, the genetic history of Europe has undergone significant transformation over time, resulting in genetic discontinuity between modern-day Europeans and their ancient maternal forbearers.

Introduction

The genetic model currently presented by many population geneticists emphasizes the autochthonous Paleolithic ancestry of contemporary Europeans. This paradigm is based on the perspective that contemporary Europeans descend primarily from their hunter-gatherer forbearers who lived in the same region until approximately 10,000 years ago, when the beginning of settled agriculture began. This Paleolithic ancestry is seen as remaining relatively unaffected by later gene flow, including any large-scale movements of farmers out of the Middle East during the Neolithic era. These agriculturalists are in fact presented as outsiders who left only limited genetic traces among contemporary Europeans, who instead derive most of their ancestry from indigenous hunter-gatherers groups that adopted Levantine agricultural practices through a cultural diffusion process.

In an effort to lend support to this genetic model, the distribution and frequency of both mitochondrial DNA (mtDNA) and Y chromosome haplogroups among modern European populations are utilized in reconstructing ancient population histories. The Basque, lone speakers in of a non-Indo-European language living in the Pyrenees Mountains of Spain and France, are often presented as the best example of a contemporary European group that retains the strongest and most undiluted genetic ancestry derived from Europe's Paleolithic inhabitants.

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Address for correspondence: Ellen Coffman, Ellenlevy66@yahoo.com

Thus, the picture presented by this model is one of substantial genetic continuity between modern groups and the Paleolithic hunter-gatherers who inhabited the same region thousands of years ago.

Yet the DNA evidence suggests a more complex picture than a direct and undisturbed genetic link between contemporary Europeans and their Paleolithic forbearers. A significant and as of yet unexplained genetic discontinuity exists between present and past populations. Since the recent advent of techniques allowing the extraction of DNA from ancient remains ("aDNA"), in particular mtDNA, the actual genetic background of the ancient maternal inhabitants of Europe can now be compared to their contemporary counterparts. Rather than using contemporary European DNA to reconstruct the genetic histories of populations from the past, this new technique allows researchers to determine to what extent later European populations truly do retain the genetic legacy of the earlier group.

In contrast to the Paleolithic paradigm, these studies indicate an unexpected and significant genetic discontinuity exists between contemporary Europeans and their Paleolithic predecessors. They also suggest that the exclusive use of contemporary DNA samples in the reconstruction of earlier population histories has created a misleading picture of the European genetic legacy.

Various demographic and evolutionary mechanisms may have led to this genetic break with the past, including the strong likelihood of genetic contributions from migratory peoples that occurred during the Neolithic, and into the Bronze and Iron Ages. This gene flow may have been so significant that genetic signals from the earlier inhabitants of Europe have been all but

obliterated, even amounting to wholesale population replacement. Founder effects, genetic drift and bottlenecks also have had a dramatic impact. In addition, Darwinian principles of natural selection and resistance against disease may have changed the face of Europe over time, causing certain genetic groups to disappear while others have come to dominate the genetic landscape. These events, either alone or in combination, have resulted in a striking genetic discontinuity between past and present populations.

As a result, contemporary Europeans should not be viewed as descending entirely or even significantly from either Neolithic farmers or the indigenous Paleolithic inhabitants of Europe. Rather, Europeans appear to be an entirely new and modern genetic mix formed as a result of a number of demographic and evolutionary events over time, including the continual movement of peoples across the European continent over the millennia.

The Popular Paradigm of Paleolithic Ancestry: Evidence from Central Europe and the First Farmers

One recent and highly publicized DNA study illustrates not only how the idea of autochthonous Paleolithic origins of modern-day Europeans has been readily adopted by many population geneticists, but also how the researchers fail to address contradictory evidence that appears to conflict with that Paleolithic paradigm.

Anthropologist Joachim Burger and graduate student Wolfgang Haak of Johannes Gutenburg University in Mainz, Germany, along with their research team, successfully extracted mtDNA from twenty-four ancient remains buried at sixteen early farming sites in Germany, Austria and Hungary. (Haak 2006) All the human remains were dated to 7500-7000 years ago based on associated cultural finds connected with the Linear Pottery Culture (Linearbandkeramik or "LBK") that is regarded as marking the onset of the early farming cultures of Central Europe. The study sought to answer the crucial question of whether Europeans descended from the first European farmers of the Neolithic period 7500 years ago, or from Paleolithic hunter-gatherers who were present in Europe since 40,000 years ago.

Out of twenty-four samples, seven were in mtDNA haplogroup H or V, five were T, four K, one J and one U3. These haplogroups, widespread in Europe, the Middle East and Central Asia, and apparently lacking "temporal or geographic discrimination," did not yield enough useful clues to allow the researchers to address their Paleolithic versus Neolithic ancestry question. Thus, the researchers focused on the remaining six results.

Those six samples were found to be within haplogroup N1a, a distinctive branch of haplogroup N that is rare among Europeans today. Among the ancient samples, however, N1a was found at a high frequency ranging from 8% to 42%, or 150 times more frequent than among modern-day Europeans. It was also widespread, appearing in sites in both Hungary and Germany.

Based on this evidence alone, the authors of the study concluded that the early Neolithic agriculturalists had "limited success in leaving a genetic mark on the female lineages of modern Europeans." They further concluded that because modern Europeans do not appear to be descended from the first farmers, they must therefore be direct descendants of indigenous Paleolithic hunter-gatherers.

However, the possibility that either genetic drift or post-Neolithic migrations caused N1a to disappear from modern European lineages requires adequate examination. Haak and Burger dismissed the first possibility and failed to address the second. Using computer simulations intended to reconstruct the impact of genetic drift over the past 7500 years, the researchers found that drift alone could not account for the disappearance of N1a lineages among contemporary Europeans. This scenario, however, did not address other factors such as susceptibility to disease which may have had an impact on mtDNA groups over time and could have affected the longevity of the N1a lineage, particularly when coupled with the effects of genetic drift.

Nor did the authors address the possibility of a post-Neolithic replacement scenario, noting only that "[a]rchaeological evidence for such an event is as of yet scant." Yet large-scale movement of peoples throughout Europe is recorded in both the archaeological record and numerous historical accounts. Given the lack of genetic continuity between modern Europeans and Paleolithic samples as evidenced by other aDNA studies, impact from post-Neolithic migrations is not only reasonable but highly likely.

Haak mentions only a single additional ancient DNA study in a footnote, addressing such evidence only in the most cursory fashion. That study involved the DNA testing of a 2500 year-old skeleton belonging to the Scytho-Siberian population of the Altai Republic in Central Asia. (Ricaut 2004) The mtDNA results indicated that this individual also belonged to haplogroup N1a. The researchers noted the absence of N1a among 490 modern Central Asian DNA results, but found N1a in low frequency in nearby Iran and southeastern India. They concluded that changes in the genetic structure of Central Asian populations must have occurred in the past 2500 years, probably as a result of Asian population movements into the west.

This conclusion was supported by the finding that in India, N1a was absent from the Dravidic-speaking population, but present in low frequency among five Indo-Aryan speaking individuals, the majority belonging to an upper Brahman caste. This suggested that the Bronze Age migrations of the Indo-Aryans and Indo-Iranians out of the Central Asian steppe and into southeastern regions of Eurasia could be responsible for the appearance of N1a among these contemporary populations. Given that the Scythians were an Indo-Iranian speaking people believed to have originated in the western Altai, there appeared to be a possible N1a connection between the ancient Scytho-Siberian peoples and contemporary Indo-Iranian derived populations of India and Iran.

Yet this explanation, while reasonable, did not explain the disappearance of N1a entirely from a region believed to be the homeland of the Scythian peoples, and when linked with Haak's older N1a findings from Europe, the mystery of N1a's disappearance only deepened. Furthermore, both the European N1a remains and the Central Asian skeleton clearly belonged to the Eurasian rather than the south Asian/African branch of N1a. It became less likely, then, that they were N1a "outsiders" migrating from outside Eurasia. This fact led the Scytho-Siberian researchers to suggest that more recent migrations within the last 2500 years impacted the genetic ancestry of modern Central Asians; Haak, on the other hand, ignored the possibility of post-Neolithic migrations impacting the genetic structure of modern Europeans. He also failed to address the fact that modern-day Middle Eastern and Caucasian groups living in regions from whence the Neolithic agriculturalists would have originated possess a high frequency of the south Asian/African branch of N1a, while Europeans fall almost exclusively within the Eurasian branch. Yet none of Haak's skeletal remains fall within the Asian/African branch, making the supposed link between N1a and the Neolithic agriculturalists less likely. (Haak 2006, supplementary material).

Haak also failed to examine other available aDNA research on ancient Paleolithic remains from Europe. Had he done so, he would have discovered that haplogroup N, the ancestral root of N1a, was present among Paleolithic Europeans. Two Cro-Magnon individuals were tested from the Paglicci cave of Southern Italy, a site dated to 23,000 years ago, during the Upper Paleolithic era. (Caramelli 2003) The first individual belonged to haplogroup HV or pre-HV, two groups noted to be "rare in general but with comparatively high frequencies among today's Near Easterners." The second individual belonged to macrohaplogroup N, which contains haplogroups W, X, I, N1a, N1b, N1c and R. However, a single change in the CRS at HVRI nucleotide position 16223 suggested classification in haplogroup N* rather than in any of its subgroups. N* is found today in low frequency in the Middle East and Central Asia, including among Iranian, Pakistani and Indian populations. (Quintana-Murci 2004) It is rare in modern-day Europeans. Thus, not only did the study suggest that Paleolithic Europeans displayed a closer genetic link with modern-day Middle Easterners than they did with contemporary Europeans, but it also established that the root of N, obviously ancestral to Eurasian group N1a, was already present in Europe during the Paleolithic period.

Nor did Haak address the extensive Strontium isotope data derived from the same sites presented in his study. Strontium isotopes from human teeth and bones provide a geochemical signature of the place of birth and the place of death of the individual. Thus, it can be used as a direct measurement of migration, tracking the movement of groups between different geological zones. Three archaeological studies containing Strontium isotope data on the LBK sites generated similar results. (Bentley 2003; Bentley 2002; Price 2001). They suggested that while many of the LBK inhabitants moved to these sites from some distance away, it was also evident that some of the individuals were of local origin. This raised the question, however, of whether the local individuals represent sedentary farmers or local foragers/herders. One study suggested that the non-locals were hunter-gatherers (Bentley 2003) while the other study suggested they were immigrant farmers (Price 2001).

Archaeologists were also able to differentiate between the locals and immigrants by examining burial goods. In particular, many locals were buried with shoe-last adzes, particularly at sites like Flomborn in Germany, where the isotopic data also indicate that almost all the adze burials accompanied individuals of local origin. (Bentley 2002) Haak noted in his study that the Flomborn individual was indeed buried with a shoe-last adze. However, many of the other human remains he tested were not and thus, it should not be assumed that they were also of local origin. Rather, Haak appeared to be examining a mixture of locals and immigrants. His samples should therefore not be deemed exclusively "Neolithic" or "Paleolithic" in origin.

Intermixture between Paleolithic and Neolithic peoples is further supported by the fact that burial orientation also correlated with place of origin. (Price 2001) This intermixture becomes especially apparent at Schwetzingen, at site also tested by Haak and representing the later phase of the LBK, when the process of contact between the farmers and hunter-gatherers appears to become more complex. At Schwetzingen, all but two of the immigrant burials are oriented in directions from north to east. At Flomborn,

4 of the 5 west-facing burials were of immigrants. Yet in Haak's samples, the Flomborn and Derenburg individuals were buried in an East-West direction, while in Halberstadt, the burial orientation was West-East. One study suggested that immigrant brides may have been incorporated into the community and given a local identity, including burial in a northeastern direction. (Bentley 2003)

The failure of Haak's genetic study to incorporate important archaeological data along with other ancient DNA results leaves the question of N1a's ultimate origins unanswered. Nor is the mystery of N1a's disappearance among Europeans today adequately addressed. The idea that N1a represents a Neolithic farming lineage that failed to impart a genetic legacy is not supported by the evidence. Based on the limited N1a findings, Haak made a sweeping generalization that the Neolithic farmers overall failed to have a significant genetic impact on Europe. But the evidence suggests a much more complex picture, even the possibility that N1a may represent a Paleolithic European lineage that has mysteriously diminished over time.

The Basque: Reflections of a Paleolithic Past?

The group most often presented as the best representatives of the genetic descendants of Europe's pre-Neolithic hunter-gatherer past is the Basque. In this case, the idea of relatively undiluted Paleolithic ancestry is an understandable one, given that the Basque, who today inhabit the Pyrenees Mountains of Spain and France, remain the only Western European group that continue to speak a non-Indo-European language (Euskara) with no known living European relatives. The Basque are generally depicted not only genetic vestiges of the most ancient inhabitants of Europe, but also one of the oldest human isolates, receiving no significant genetic contributions from outsiders. This isolation is supposedly reflected by Basque cultural/linguistic uniqueness, a high level of endogamy, and geographic protection among the Pyrenees Mountains.

Because of this uniqueness, the Basque have been the subject of numerous genetic studies, allowing researchers to investigate whether the perception of the Basque as representatives of the indigenous Paleolithic gene pool is in fact a valid one. Furthermore, numerous aDNA studies have also been performed on both Neolithic and historic Basque remains, providing the opportunity for researchers to compare modern-day Basque with their Neolithic counterparts. Thus far, however, only a few researchers have performed such a comparison. Those few have reached conclusions that conflict with the idea that the Basque population represents a "living fossil" of the first European settlers of the Paleolithic. (Alonso 2005)

One such study compared the mtDNA variability of a historical Basque population (VI-VII c. AD) recovered from the necropolis of Aldaieta (Nanclares de Gamboa, Araba, Basque Country) with remains tested from three prehistoric sites in Basque Country dating to 4000-5000 Years Before Present ("YBP"). (Alzualde 2005) These populations were then compared with modern-day Basque. The results were stunning.

The researchers discovered that the mtDNA of the historical Basque population falls within the range of present-day populations along Europe's Atlantic coast, known as "the Atlantic fringe," while the prehistoric Basque populations were clearly differentiated. In particular, notable frequency differences were found among the haplogroup K, V, H and J results. For example, haplogroup K was found at a high frequency among the prehistoric groups (16% - 23%), but is nearly absent from present-day Basque, and in contemporary Europeans, it occurs between 3.6% to 7.7%.

Haplogroup H, hypothesized to have been present in Europe since at least the late Paleolithic and the most common haplogroup among present-day Europeans (approximately 50%) and Basque (62%), was also found at a high frequency of 48% among the historical remains at Aldaieta, but at lower frequencies at the prehistoric sites (37% at SJAPL and Rico Ramos, 44% at Longar). This variation suggests that there was heterogeneity between the various prehistoric communities themselves, with some communities having a higher frequency of certain haplogroups than others. (Alzualde 2005)

Haplogroup V, on the other hand, is believed to have originated in the vicinity of Basque County, possibly during the late Paleolithic, and is found today in its highest frequency among present-day Basque groups (10.2%). Yet the researchers found no V among the prehistoric remains. The complete absence of prehistoric V in a region where V is believed to have originated disrupts the theory of V's Paleolithic origins among southwestern Europeans. It also casts doubt on the belief that V's presence among modern-day Basque represents genetic continuity from their Paleolithic ancestors (Izgirre 1999).

Further aDNA evidence supports the view that V either did not originate in this region or does not represent the vestiges of an autochthonous Paleolithic lineage. It has been theorized that the Spanish and the Basque may share genetic affinities as relics of indigenous Paleolithic Europeans. The ancient Iberians, a group inhabiting the Iberian Peninsula including present-day Spain (and

ultimately giving their name to this region) from at least the Bronze Age, also spoke a non-Indo-European language, though unrelated to the Basque tongue. Yet in a study examining the mtDNA diversity of the Pre-Roman ancient Iberians dating from 6th century BC, the researchers also found an absence of haplogroup V, leading to the conclusion that "this lineage was not especially prevalent in the ancient populations with non Indo-European languages from the Iberian Peninsula" (Sampietro 2005). The researchers instead suggested that the high frequency of V among contemporary Basque was more likely the result of genetic drift or admixture with later arrivals of haplogroup V migrants. In particular, the impact of later immigrants to the region, including the Romans, Visigoths and Vandals, may have exerted a cumulative genetic impact on the peoples of the Iberian Peninsula. Most importantly, they strongly suggested that both Iberians and Basque were composed of a "complex mosaic of pre-Roman peoples" that emerged not from Paleolithic hunter-gatherers, but from later Bronze and Iron Age local communities.

Similar conclusions were reached when analyzing the mtDNA haplogroup J results among the Basque. Haplogroup J is considered a main lineage of Neolithic expansion out of the Middle East. Although absent from the prehistoric Basque site of Longar, it is present at the other two, displaying values of approximately 16%. Even at Aldaieta, haplogroup J reached a frequency of 14.7%, yet in present-day Basque, J is present at a low frequency of 2.4%. Thus, this Neolithic lineage was clearly present in high frequency among both the prehistoric and historic Basque groups. Yet the low frequency of haplogroup J among contemporary Basque has led researchers to incorrectly conclude that the early Basque population was genetically unaffected by this Neolithic lineage. In reality, haplogroup J appears to have suffered a severe reduction in frequency among the Basque over time.

Frequency differences among the mtDNA results between modern and prehistoric Basque populations led researchers to conclude that a "discontinuity" exists between prehistoric and modern-day groups (Alzualde 2005). The results also suggest that the reconstruction of the biological history of European populations based only on current DNA results is often misleading and incorrect. Using haplogroup J as an example, Alzualde explains that because "the Basque population is considered an outlier regarding the Neolithic component, it has been proposed that this region experienced a smaller genetic impact from Neolithic farmers. But if we accept that lineage J is a marker of migrations of Neolithic populations from the Near East, then the Basque Country also experienced the impact of these people, as shown by the high frequency of haplogroup J in certain ancient populations" (Alzualde 2005).

Additional aDNA evidence from Basque archaeological sites lends support to this conclusion. Alzualde more closely investigated the frequency and presence of various haplogroups, including a number of uncommon haplotypes, among the Basque of the 6th-7th centuries from the historic site of Aldaieta. (Alzualde 2006) He had also examined the mtDNA from Aldaieta in his previous study, though not with the depth of coverage present in this subsequent investigation.

The study emphasized the uncertain background of the Aldaieta population. While the remains suggested that the site was settled by "autochthonous individuals" with stable familial ties, the high percentage of weaponry and similarity of mortuary objects with Frankish cemeteries were also noted, indicating possible trade links or even temporary Frankish control of Basque territory.

Of the fifteen haplotypes from Aldaieta, nine are uncommon or unique haplotypes. The unique haplotypes are found within haplogroups T, U5, U2 and J. One of these haplotypes bore close affinities with modern-day populations in Eastern Europe, while the remainder of the unusual haplotypes were nearly absent from contemporary European populations, including the Basque.

The researchers also discovered the presence of haplogroup M1 at Aldaieta, a rare haplogroup among present-day Europeans and peoples of the Iberian Peninsula. This suggested a probable genetic relationship between the historical Basque group and Northwestern Africa, where sequences similar to those at Aldaeita have been found. This relationship predates the Moorish period, since the chronology of the material remains at Aldaeita is dated earlier than the 8th century AD, the time when occupation by the Moors began.

The researchers issued a warning to other geneticists, suggesting that hypotheses formulated solely on the basis of DNA results from modern-day populations, without accompanying aDNA evidence, can lead to inaccurate reconstructions of population histories. Geneticists who propose an undiluted Paleolithic ancestry for the Basque often do so without reference to numerous aDNA studies. As a result, they incorrectly attribute the unique and unusual genetic results of contemporary Basque as indicators of undiluted Paleolithic ancestry.

The Aldaieta study concluded with the controversial suggestion that the Basque were not only impacted by cumulative gene flow from Neolithic Near East ancestors and as well as later invaders, but may have been affected by significant post-Neolithic biological events, including genetic drift and natural selection. Given the genetic discontinuity between present and prehistoric populations, the researchers urged their

colleagues to consider the idea that "the genetic patterns of present-day populations reflect the evolutionary processes experienced by their predecessors," suggesting that these post-Neolithic processes have altered the genetic composition of the Basque and European populations as a whole.

Other genetic studies on the Basque have focused on examining blood groups, STR loci, and autosomal markers, often in an attempt to support the Paleolithic paradigm. However, in light of the aDNA studies, Basque distinctiveness can be accounted for by the processes of genetic drift, inbreeding over long periods of time and natural selective processes. For instance, a correlation was observed between increased genetic differentiation between Europeans and Basque groups still speaking the Basque language. (Perez-Miranda 2005) It has been postulated that one of the causative agents of Basque isolation over the centuries is the Basque language. Thus, the more conservative the retention of the Basque language, the more likely the particular Basque community suffered the effects of isolation and genetic drift.

Moreover, the researchers noted that the Basque are unique among European populations due to their extremely high rate of consanguinity. Basque social and cultural traditions continue to promote consanguinity. The genetic impact of such inbreeding has yet to fully explored by geneticists, but the high frequency of inherited disorders among the Basque, including Coagulation Deficiences (Factor XI) and Mutation F508 (Cystic Fibrosis Gene), support the suggestion that drift, inbreeding, and a small population size maintained over many generations, as opposed to significant retention of Paleolithic genetic ancestry, best explains the present genetic makeup of the Basque (Alonso 2005; Bauduer 2005).

Finally, even researchers that have found limited genetic evidence of probable Paleolithic ancestry among the Basque also acknowledge that such findings do not support the contention that contemporary Basque retain significant genetic links with indigenous Paleolithic Europeans. (Gonzalez 2006) For instance, although the Basque mtDNA lineage U8a may date to the late Paleolithic, it is rarely found today among modern-day Europeans and, furthermore, constitutes only 1% of contemporary Basque mtDNA results. Thus, U8a has diminished in frequency among populations today in a manner similar to the N1a lineage.

Etruscans: Extinction or Mutation?

Like the ancient Basque, the origin of the Etruscan people remains obscure. The Etruscans lived in central Italy from the 8th-2nd centuries BCE. Like the Basque, they spoke a non-Indo-European language, but unrelated to the Basque language. After the Romans rose to dominant Italy in the 2nd century BCE, the Etruscan language disappeared from the records. It was therefore assumed that the Etruscan population had been culturally and genetically assimilated by the Romans. But the aDNA evidence tells a different story.

Two separate aDNA studies on the Etruscans reached similar conclusions, finding essentially no genetic relationship between the ancient Etruscans and the modern-day inhabitants of Tuscany (ie, "Tuscans") (Belle 2006; Vernesi 2004). Specifically, out of twenty-eight mtDNA sequences, only six occur in any modern-day groups. The remaining twenty-one haplotypes, identified as belonging to the JT haplogroup, do not occur in any contemporary European populations, including the common Etruscan haplotypes 16126-16193 and 16126-16193-16278. These sequences, while occurring among modern-day haplogroups J2 and T, are not accompanied by substitutions at 16069 and 16294, respectively, which are inevitably present among the contemporary motifs (Vernesi 2004).

The researchers attributed this lack of genetic relationship between Etruscans and Tuscans to two possible processes – the extinction of Etruscan mtDNA lineages among modern-day Europeans, or demographic and evolutionary processes occurring in the last 2,500 years. These processes, if they occurred, were severe enough to disrupt the genetic continuity between the modern and ancient inhabitants of Tuscany.

Researchers performed a number of simulations to investigate whether certain phenomenon, such as genetic drift, migration or a higher than average mtDNA mutation rates, could have impacted the genetic continuity between Etruscans and Tuscans. (Belle 2006) None of their simulations were compatible with the DNA results. The genetic evidence did not support the conclusion that Tuscans were the modern-day descendants of the Etruscans, although the researchers noted that the skeletal remains used for their aDNA samples may not have been representative of the entire Etruscan population, but of a more elite sub-strata. Even so, they seemed to have contributed very little to the mtDNA background of modern Tuscans.

However, the researchers also found that genetic continuity could be generated if the mtDNA mutation rate was set very high (0.5 mutations per million years as opposed to commonly used lower rate of approx. 0.05 mutations per million years per nucleotide) or if gene flow from other areas was so extensive that Etruscan descendants became underrepresented in the modern Tuscan samples. They concluded, however, that the very high mtDNA mutation rates needed to reproduce genetic continuity were "implausible" and, furthermore, the only way to determine if descendants

were underrepresented in the study was to collect more modern samples over time.

Thus, the study concluded that modern-day Tuscans largely descend from non-Etruscan ancestors. Regarding the fate of the Etruscans, the suspicion voiced by the researchers was that the Etruscan lineages simply went extinct.

Evolutionary Extinction of mtDNA Lineages

If the idea of mtDNA lineage extinction is accepted as the most likely scenario leading to the weak genealogical relationship between ancient and modern-day populations, then the question arises as to what evolutionary, demographic or human processes have caused such extinctions.

For example, in the case of the Etruscans, significant historic gene flow could have diluted the Etruscan lineages to the point where they have become difficult to detect in modern-day Tuscans, particularly if the lineages were already weakened by extensive gene flow from the Romans. DNA studies examining ancient remains from periods much later in time than the Iron Age Etruscans suggest admixture with other populations may indeed be a strong contributing factor to the apparent genetic break between past and present populations. Given the later date of these aDNA remains, more genetic traces of these ancient lineages are often found among their modern-day European descendants. Still, notable differences remain.

One such study examined the genetic legacy of the ancient Cumanians, believed to have migrated from Central Asia during the 13th century, among their alleged Hungarian descendants. After analyzing the aDNA of eleven medieval Cumanian samples, the researchers found that some of the lineages could not detected among modern-day Hungarians. (Bogacsi-Szabo 2005) One Cumanian sequence belonged to haplogroup D, found in modern Buryats of Central Asia, but absent from Hungarians (and rare in Europeans, found primarily among European Russians at 1.86%). Some of the samples were assigned to haplogroup U, but again no identical sequences could be found among Hungarians. One of the skeletal samples was identified as a Cumanian chieftain based on burial goods. His sequence belonged to U3, present in Greeks and some Balkan groups, but absent from the contemporary Hungarian sample. A few of the sequences, originally thought to belong to haplogroup F, but possibly belonging to haplogroup T, are found among Hungarians today. Still, the notable lack of genetic continuity between Cumanians and Hungarians led the researchers to suggest that a dilution of the Cumanian lineages had occurred, possibly due to admixture with "more westerly genetic elements." Yet the study failed to cite evidence supporting this contention, other than relying on "legends" of Cumanian nomads carrying off local women from raided territories (Bogacsi-Szabo 2005).

In another study examining aDNA from an early Danish Christian cemetery dating to 1000-1250 AD, two rare haplotypes were found among the ten samples. One belonged to haplogroup U7, absent from modern-day Scandinavians, but found in contemporary groups in the Middle East, India and western Siberia. The other unusual sequence belonged to haplogroup I, occurring in only 2% of modern-day Scandinavians. These results suggested that individuals living in this area of Denmark as recently as 1,000 years ago "comprised individuals with genetic links with populations that were much farther away." Furthermore, there was a surprising diversity among the aDNA results and a lack of direct maternal or sibling relationship between the subjects, indicating that "the ancient Danes were not just members of a tightly knit local population; some probably originated from far way" (Rudbeck 2005).

The researchers speculated that the Scandinavian population had either not yet become stable, or in the alternative, that the close proximity of the site to a nearby port town had brought in significantly more immigrants than might be found in groups residing in the surrounding countryside. This suggests the possibility of increased genetic continuity of ancient lineages among populations living far from ancient urban centers where admixture and cumulative gene flow with other incoming populations may have been more significant. It also indicates that ancient populations were not genetically static or homogenous, but incorporated immigrants from regions far away.

This fact was further emphasized in a study of Anglo-Saxon remains from a number of archaeological sites in England (Topf 2005). Researchers found that while the early skeletal remains, including those from early Saxon sites, showed close genetic links with northern European populations such as Estonia, Norway and Finland, later Saxon sites dated a mere few hundred years later were closer not only to southern European populations (Germany & Spain) but to Middle Eastern groups as well (Topf 2005).

The idea of dilution of genetic lineages was also proposed in the ancient Iberian study in which the researchers noted the likely cumulative genetic impact exerted by the invading Romans, Visigoths and Vandals on the peoples of the Iberian Peninsula. (Sampietro 2005) However, while redistribution of mtDNA lineages by migration and dilution may be one factor impacting genetic continuity, evolutionary processes may be another contributing factor. This controversial idea was proposed by Alzualde in his studies on the

Basque (Alzualde 2005). What these evolutionary processes might be, however, was not explored further.

There are numerous studies suggesting that mtDNA genetic variation may be associated with adaptive selective, as well as linked with complex diseases and disorders. (Ruiz-Pesini 2004; Moilanen 2003) Some researchers have argued that the human genome evolution has been shaped over time primarily by infectious disease and that mtDNA has played a central role in the selection process due to its control of cellular metabolism (Samuels 2006).

Mitochondrial haplogroup J has been associated with Leber's hereditary optic neuropathy, a rare disease that causes blindness in young people (Man 2004). It has also been associated with possible protection against Parkinson Disease, but increased susceptibility to multiple sclerosis (Ruiz-Pesini 2004; Ross 2003). Furthermore, Haplogroup J has been linked to increased longevity (De Benedictis 1999; Coskun PE 2003). Haplogroups K and T have been associated with protection against Alzheimer's disease (Ruiz-Pesini 2004). Haplogroup K has also been linked with a lower risk of Parkinson's Disease (Ghezzi 2005). Haplogroup U has been linked to increased risk of occipital stroke, and sub-clade U5 specifically to migrainous stroke (Finnila 2000). Haplogroup H has been linked to increased survival rates after recovery from sepsis (Baudouin 2006).

However, a number of other studies have failed to substantiate links between various mtDNA haplogroups and selective disease resistance or adaptive advantages (Houshmand 2004; Yao 2002; Rose 2001). Thus, the role that mitochondrial function and variability may play in adaptive selection, particularly disease resistance, remains unclear.

Other researchers have asserted that specific mtDNA replacement mutations allowed our prehistoric ancestors to adapt to more northern climates as they migrated out of Africa (Ruiz-Pesini 2004). These same mutations are allegedly influencing our health today. These secondary health effects became either deleterious or adaptive in terms of evolutionary selection.

In this scenario, lineages that encounter new environments for which their mitochondria were maladapted would be eliminated by selection, as would any lineages that developed deleterious mutations. These extinctions would leave no traces in the phylogeny. Similarly, selection would cause lineages with mutations that were positively adaptive to become more numerous. Those adaptive mutations can still be observed in the internal nodes of the phylogeny where their position indicates that they have been highly conserved.

The researchers suggested that mtDNA haplogroup variation was primarily influenced by climatic selective pressures. Specifically, changes in mtDNA amino acid variants permitted certain ancient European mtDNA lineages to adapt to colder climates, particularly among haplogroups H, I + N1b, J, and X. These haplogroups had higher replacement mutation values among their internal branches and higher retention of the altered amino acids when compared to those in mtDNA haplogroup L, the most common haplogroup in Africa, indicating the influence of adaptive selection among the European lineages. According to the researchers, this arctic selection resulted in "the regional enrichment of specific mtDNA lineages (haplogroups)" in Europe (Ruiz-Pesini 2004).

However, it should be noted that haplogroups I, N1b, and X occur at much lower frequencies among European populations today than haplogroups H and J, in conflict with the idea that these particular lineages have equivalent survival advantages based on climatic adaptation. Additionally, the theory of climatic selective pressure shaping the mtDNA genome continues to remain a matter of debate among geneticists.

In another study, researchers suggested that differences observed between the mtDNA groups utilized in the Ruiz-Pesini study were merely the result of comparing "region-specific haplogroups of different diversity levels: e.g., the "old" paragroup L in Africans vs. "young" Arctic haplogroups" (Kivisild 2006). Although Kivisild's study did not detect lineage-specific positive selection, evidence of site-specific positive selection was found within mitochondrion-encoded rRNA. This selection appeared to involve the replacement of two specific amino acids, threonine and valine, with two other acids, alanine and isoleucine. This pattern led Kivisild to suggest that diet rather than climate could be one important selective factor impacting mtDNA population histories. According to Kivisild, [t]hreonine and valine, essential amino acids that must be taken in the diet, are abundant in meats, fish, peanuts, lentils, and cottage cheese, but deficient in most grains."

Given the significant modification in diet that European populations underwent during the Neolithic era as they transitioned from hunter-gatherer subsistence to an agricultural grain-based diet, Kivisild's theory of potential selective pressure based on dietary factors warrants further investigation.

Ancient mtDNA variants advantageous in one climate or dietary environment may have been maladaptive in a different environment, contributing to the rise of modern bioenergetic disorders such as obesity, hypertension, diabetes and cardiovascular disease. (Mishmar 2002) However, whether due to dietary factors, climate adaptation, disease resistance or a

combination of selective pressures, these studies suggest that natural selection may have played a role in determining which mtDNA lineages survived over time.

Conclusion: Why We Are Not Our Ancestors

The ancient DNA studies present a picture of genetic break or "discontinuity" between ancient and modern-day European maternal histories. This evidence indicates that modern-day mtDNA haplogroup frequencies and distributions should not be considered living fossils of Europe's Paleolithic past.

Currently, the genetic picture presented by the aDNA studies is based exclusively on mitochondrial DNA results. This form of DNA, unlike that of the Y chromosome, is generally preserved in a form that allows for testing of ancient remains. However, the Y chromosome genetic picture of Europe may also have undergone significant change similar to that impacting the ancient maternal lineages. The ancient DNA results provide a cautionary framework for geneticists in their reconstruction of the distribution and frequency of ancient European Y chromosome lineages. Modern-day Europeans cannot accurately be used as genetic proxies for their prehistoric counterparts.

These findings stand in stark contrast to the model presented by many DNA studies of an undisturbed genetic link between contemporary and Paleolithic European groups. Yet evidence of such genetic continuity is sparse, even among populations such as the Basque. More problematically, it contradicts the findings of the ancient DNA studies. These studies indicate that populations have indeed changed dramatically over time, with some ancient lineages suffering reductions and even extinctions from the European gene pool.

Extinction appears to be the fate suffered by the Etruscans maternal lineages. Many other ancient groups appear to have suffered a similar fate, the continuity of their genetic lineages extinguished for future generations. Only the archaeological record remains a testament to their existence. Certain genetic lineages, like mtDNA haplogroup H, came to dominate the genetic landscape over time. The contemporary European genetic picture is thus a reflection of these complex demographic and evolutionary processes, changing and adapting until it is no longer a mere reflection of its genetic past, but a new and constantly evolving population.

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