Archeology, dna, and genetics research proposal samples

Science, Genetics



In archeology, DNA analysis has been used as an investigatory tool since it became available to scientists. DNA analysis, which is a constantly developing and expanding field, has allowed archaeologists to precisely and decisively obtain results that solve archaeological, historical, and evolutionary questions. Unfortunately, the methods for using ancient DNA analysis tend to destroy the samples from which the DNA is extracted. The results can easily be affected by external contamination as well, which requires the research to be conducted within specialized laboratories by trained laboratory personnel (Arandjelovic, Guschanski, Schubert, Harris, Thalmann, Siedel, & Vigilant, 2009).

DNA analysis has been described as " the greatest archaeological excavation of our time (Mulligan, 2006, p. 370)." It can be used to research and understand how modern humans evolved from prehistoric ancestors and it can trace the migratory patterns of people over continents and oceans. It can even be used to identify individual people or determine the genetic roots of a plant or animal. Two scientific breakthroughs have made such discoveries possible. The Polymerase Chain Reaction, or PCR, is a technique for making identical copies of a fragment of DNA. Since ancient DNA molecules are fragile and few in number, this has proven a crucial tool for the study of ancient DNA (Arandjelovic et al., 2009). The second scientific breakthrough is the recovery of DNA from preserved samples of tissue, teeth, and bone (Mulligan, 2006). Since these sources of genetic information are relatively plentiful, they have contributed to a great deal of the body of knowledge that comprises archeology today.

In molecular biology, the polymerase chain reaction can be used to amplify a

single or multiple strands of DNA. It has frequently been used to generate millions of clones of a sequence of DNA. The majority of PCR methods employ a technique known as thermal cycling. In thermal cycling, the sample is alternately heated and cooled through a defined set of temperature steps (Arandjelovic et al., 2009).

The advent of PCR has created an entirely new field within archeology. As of 1991, most of the research on ancient DNA has dealt with the sequencing and amplification of mitochondrial DNA, which is distinct from nuclear DNA. Since mitochondrial DNA exists in abundant copies within the cell, it tends to have a higher recovery rate than the DNA copies extracted from the cell nucleus. When nuclear DNA has been heavily degraded or reduced below the detection level over centuries of decay, mitochondrial DNA can be analyzed instead. Mitochondrial DNA also has the advantage of being able to be sequenced directly after the amplification product is obtained (Arandjelovic et al., 2009). This is very useful to archaeologists, because it minimizes the damage that occurs during the amplification process. In addition, mitochondrial DNA has uniparental inheritance, which means that it is inherited entirely from the maternal lineage. This is useful in population studies or in the identification of missing persons. Mitochondrial genomes have been used to identify ancient DNA samples as old as 2500 years (Arandjelovic et al., 2009).

PCR has opened up many opportunities within the field of molecular archeology. Especially when only trace samples of DNA are available, the polymerase chain reaction has proven a most convenient forensic tool. When animal or human remains are heavily damaged and conventional methods of profiling are unavailable, mitochondrial DNA typing has yielded successful identification results (Mulligan, 2006).

One of the greatest discoveries of modern archeology has been the tracing of human origins back to our ancestral land. The analysis of ancient DNA has shown that Neanderthals and other early human species originated on the African continent and migrated to other continents over thousands of years (Linz, Balloux, Moodley, Manica, Liu, Roumagnac, & Achtman, 2007).". Modern humans, which comprise the species homo sapiens, are thought to have a single origin in Africa. This conclusion came about by comparing the skulls and DNA samples of human remains around the world, as well as by combining the analysis of genetic variations across individuals. More than 6, 000 skulls from over 100 human populations were analyzed by a team of archaeologists and forensic scientists from the University of Cambridge in England. The leader of the group, Andrea Manica, was unequivocal about their findings, saying " We have combined our genetic data with new measurements of a large sample of skulls to show definitely that modern humans originated from a single area (Linz et al., 2007, p. 918)."

The theory that humans originated in Africa has become known as the " out of Africa" theory. This theory postulates that early humans began migrating to other continents and colonizing them as recently as fifty thousand years ago, after spreading out from Africa. Other studies have confirmed that genetic differences in ancient human populations may correlate to their distance from Africa. The distance from Africa was found to determine as much as 25 percent in the variation in features seen in human skulls (Linz et al., 2007). Researchers ensured the quality of their results by making sure that the DNA analysis underwent the same theoretical framework as the analysis for the skulls. This made the results of both studies comparable. Both the study of the skull and the analysis of genetic information indicated that humans originated in Africa and spread out across the continents around 50, 000 years ago (Linz et al., 2007).

In 2009, a group of scientists under Dr. Sarah Tishkoff of the University of Pennsylvania, published the results of a comprehensive study of the genetic diversity of the people of Africa in the journal Science. The study indicated that the most ancient branch of the human species has experienced the least amount of mixing, as previously expected. It is apparently a genetic cluster, which includes the Bushmen and other peoples who speak Khoisan languages. This group of people comprises the one branch that is closest to the common ancestor of all modern humanity (Tishkoff, Reed, Friedlaender, Ehret, Ranciaro, Froment, & Williams, 2009).

In 1987, a paper titled " Mitochondrial DNA and Human Evolution" was published in the scientific research journal Nature. This paper demonstrated that the mitochondrial genome, inherited through mothers and daughters, is derived from an African ancestor who existed 100, 000 to 200, 000 years ago. This ancestor has become known as Mitochondrial Eve, named after the biblical Eve from whom, according to Christian theology, all humans are descended. All modern people are related to Mitochondrial Eve through a direct and unbroken female line. She most likely lived in East Africa, where anatomically modern humans, or Homo sapiens sapiens, were evolving separately from other human populations such as Neanderthals (Walker, Smith, & Smith, 1987).

Page 6

In the decades following the publication of the paper, more data from genetic samples from both modern and ancient human remains have supported the out of Africa theory. Alongside the fossil record, mitochondrial DNA analysis has helped reconstruct the migratory patterns of humans over tens of thousands of years (Walker et al., 1987).

In 2006, scientists retrieved a sample of mitochondrial DNA from the tooth of a Neanderthal that lived more than one hundred thousand years ago. At that time, it was the oldest human-type DNA ever found (Orlando, Darlu,

Toussaint, Bonjean, Otte, & Hanni, 2006). The discovery led to the revelation that Neanderthals, distant cousins of modern humans, were more genetically diverse than once thought. However, their diversity began to decline around 35, 000 years ago, perhaps due to changes in the environment or to disease. This corresponds to the time that humans began populating Europe (Orlando et al., 2006).

The scientists, based in Lyon, France, decoded the DNA into 123 distinct DNA " letters," or base-pair groups. They then compared the base-pair groups to other known Neanderthal DNA sequences from remains that were between 29, 000 and 42, 000 years old. Writing about the findings in the journal Current Biology, the leader of the team, Dr. Catherine Hanni, stated that " more Neanderthal sequences than the six presently available and longer than 100 base-pairs are needed to fully understand the extent of the past diversity of Neanderthals (Orlando et al., 2006, p 401)."

The study of DNA from the nucleus of the cell, or nuclear DNA, has revealed that there may have been periods of interbreeding with other human subspecies. Around two percent of the modern human genome is thought to be shared with Neanderthals. However, this is true only for people with non-African ancestry. Neanderthal DNA does not appear to be present in the lineage of people of strictly African descent. In addition, Neanderthal DNA has shown to be a risk factor for developing certain diseases, such as type-2 diabetes, and is also associated with depression and addictive behavior (Orlando et al., 2006; Walker et al., 1987).

The study of ancient DNA has been applied to a vast variety of specimens. Skeletal material, preserved plant remains, mummified tissues, ice cores, and Holocene plankton are just a few of the candidates for study by DNA analysis, and have led to significant discoveries about the ancient world (Mulligan, 2006). This is of great interest to scientists because understanding how the world was in the past brings us to a greater understanding of our present. Questions about how people lived, adapted to their surroundings, moved around, and evolved culturally are starting to be answered thanks to modern technological developments.

For example, DNA analysis has united the field of archeology with historical linguistics. Ancient DNA evidence provides clues to the origins of Indo-European languages, which span the eastern hemisphere and include more than four hundred distinct languages and dialects. Though scholars have studied Indo-European languages since the mid-17th century, most of what is known today has been discovered with the help of DNA analysis (Haak, Lazaridis, Patterson, Rohland, Mallick, Llamas, & Reich, 2015). Researchers have determined that an ancestral language, called Proto-Indo-European (PIE), was first spoken over six thousand years ago by pastoral herders who lived on the steppes near the Black Sea. One study has indicated that these herders may be the ancestors of most Europeans alive today. Another study on genetics used ancient DNA to show that massive migration from the steppes may have been a source of the Germanic languages in Europe. One study, published in the journal Nature, suggested that there were not one, but two waves of mass migration of farmers (Haak et al., 2015). The

one, but two waves of mass migration of farmers (Haak et al., 2015). The first wave of migration brought an influx of people from a region of Turkey known as Anatolia. About four thousand years later, another influx of people brought people to central Europe from the steppes of Russia. These people brought with them the ancestral Proto-Indo-European language which spread across the European continent and gave rise to modern European languages.

The genetic analysis relied on DNA samples recovered from the skeletons of 69 ancient individuals, ranging in age from 3, 000 to 8, 000 years. These skeletons were compared to each other and to modern European groups in an attempt to reconstruct the lineages of modern Europeans. The researchers found similarities between 5, 000 year old remains discovered north of the Black sea and 4, 500 year old remains of people who lived near Leipzig in central Germany. The ancient Germanic people were part of the Corded Ware culture, which refers to a type of pottery that was common throughout northern Europe during that archaeological era. The people who lived north of the Black Sea are thought to belong to a culture known as the Yamnaya (Haak et al., 2015).

The researchers discovered that although the Corded Ware people and the Yamnaya people were seperated by half a millenium and by over a thousand miles, they had at least 75 percent of their ancestry in common, making them at least close cousins. In addition, the remains of the Corded Ware people were very dissimilar to the bones of people living in that part of Germany only a thousand years earlier, indicating that a period of westward migration occurred sometime between 4, 500 and 5, 500 years ago. One archaeologist involved in the study, losif Lazaridis, described the migratory period as almost " a total replacement event (Haak et al., 2015, n. p.)." In fact, the more recent remains were found to have a completely different genetic signature from the specimens one thousand years older. This provides evidence that a movement of people from the steppes north of the Black Sea came to dominate the genetic landscape of north and central Europe. This evidence corroborates the so-called " steppe hypothesis" posed by linguists, which postulates that Proto-Indo-European has its origins in the pastoral regions to the north of the Caspian and Black Seas (Haak et al., 2015). However, there are more questions to be answered and more work needs to be done in order to come to definitive conclusions about the origins of Indo-European languages. For example, it may be that the steppe north of the Black Sea is not the ultimate source of Proto-Indo-European, but is just one of several routes taken by the tongue to Europe, India, and Iran. In order to determine if that is the case, more genetic data must be gathered and analyzed.

In addition to studying mitochondrial DNA, geneticists have begun to analyze DNA from the Y chromosome. Y chromosome DNA is passed along from father to son, unlike mitochondrial DNA which is passed from mothers to daughters. Genetic mutations cause changes in the Y chromosome, which makes Y chromosomal analysis useful as a clock for tracing the history of

human populations. If the assumption is made that all living people share a common male ancestor as they do a common female ancestor, then analysis of the Y chromosome should shed light on when he lived. Some estimates place this patrilineal ancestor, known as Y chromosomal Adam, between 200, 000 and 300, 000 years in the past (Mendez, Krahn, Schrack, Veeramah, Woerner, & Hammer, 2013). This means that the most recent common female ancestor and the most recent common male ancestor did not live at the same time.

Y chromosome analysis has led to a greater understanding of human evolutionary history. With modern genetic techniques, a new branch of archeology has been founded called archaeogenetics (Mendez et al., 2013). This can involve not only the analysis of ancient DNA, but also the analysis of DNA from current human populations. The study of archaeogenetics has applications in the study of the legacy of human interaction with the ecosystems of the world, which are collectively known as the biosphere. This means that archaeogenetics has assisted researchers in understanding how early humans shaped modern physical landscapes, giving us a window into a past that was once obscure (Renfrew, 2001).

In addition to human DNA, the analysis of non-human DNA has opened up the possibility of learning about ancient cultural practices. For example, the analysis of cattle DNA can allow us to learn about human domestication of animals. The domestication of animals and crops is a cumulative process marked by genetic and morphological changes. Before genetic analysis became a widely available tool, it was thought that the domestication of cattle first occurred in the Near East (Zeder, Emshwiller, Smith, & Bradley,

2006). Archaeologists thought that domesticated cattle in Africa, India, and Europe all descended from a progenitor in the Near East. These cows are thought to have given rise to populations of distinct breeds over generations of breeding.

Now, DNA analysis has shown that Indian cattle parted from an ancestral line that originated sometime between 117, 000 and 275, 000 years ago. Between 22, 000 and 26, 000 years ago, the line split again, giving rise to modern European and African cattle (Zeder et al., 2006). These results were surprising to archaeologists and contradicted the previous theory about a Near Eastern ancestor, because cows in the Near East did not become domesticated until only as recently as 9, 000 years ago. Since cattle in Africa and Europe were genetically distinct long before then, they could not have descended from a Near Eastern progenitor. This means that African and European cattle must have been domesticated independently (Zeder et al., 2006).

In 2013, an international team of scientists recovered ancient DNA from human remains dated at 5, 500 years to create a reconstruction of the genetic history of Europe. The reconstruction showed that a series of migrations, including the ones from the steppes north of the Black and Caspian Seas into central Europe, changed the population landscape over the course of thousands of years, as evidenced by the genetic lineages carried by as much as 45 percent of Europeans (Brotherton, Haak, Templeton, Brandt, Soubrier, Adler, & Genographic Consortium, 2013). The detailing of the genetic history begins with the early Bronze age and goes up to the modern era.

Page 12

The record of maternal genetic inheritance was named Haplogroup H. It revealed that the first European farmers came about as a result of wholesale migration from Turkey and the Near East, where the first farming techniques were discovered. These migrants arrived in Germany around 7, 500 years ago. However, they were genetically replaced around 4, 500 years ago, during a major genetic transition. The team found that modern Europeans can trace their lineage to a genetic foundation in the Mid-Neolithic period. The lineage was further modified by cultures arriving from Eastern Europe and Iberia during the Late-Neolithic. Professor Alan Cooper, who co-founded the study, stated that " these well-dated ancient genetic sequences provide a unique opportunity to investigate the demographic history of Europe. We can not only estimate population sizes but also accurately determine the evolutionary rate of the sequences, providing a far more accurate timescale of significant events in recent human evolution (Brotherton et al., 2013, n. p.)."

In addition to answering questions about our heritage, ancient DNA analysis can potentially address a number of other anthropological questions. For example, the genetic analysis of coprolites, or fossilized fecal matter, has shed light on ancient human dietary patterns, and it may be used as an alternate source of human DNA (Fox, 2008). For example, coprolites discovered in the Paisley Caves, a system of four caves in south-central Oregon, revealed DNA samples radiocarbon dated to 14, 300 years ago. DNA analysis showed the samples had genetic ties to Asia or Siberia, providing evidence for a migration over an ice bridge (Fox, 2008).

Fossilized feces have revealed the dietary preferances of early human

ancestors. It is known that Neanderthals, as an example, ate both meat and vegetables. Fifty thousand year old coprolites from Neanderthals extracted from an ancient hearth in Southern Spain showed that although their diet was meat-heavy, they also ate nuts, berries, and other vegetables (Sistiaga, Mallol, Galván, & Summons, 2014). To study the coprolites, researchers pulverized the dried fecal matter and searched for chemical compounds that are created by bacteria during the metabolism of meat and vegetables. This showed that Neanderthal digestion worked similarly to the way digestion works in modern humans. In addition to providing clues to the diet of early human species, the genetic analysis of coprolites also suggested which kinds of plant life dotted the landscape of ancient Spain (Sistiaga et al., 2014). Genetic analysis of Neanderthal coprolites could potentially answer questions about what led to their extinction. Since their diet appeared to be meat-centric, they may have been forced to compete for resources with other early human species that were more omnivorous. The coprolites also showed that Neanderthals' bodies were ridden with parasites, such as hookworms and pinworms (Sistiaga et al., 2014). These parasites were very similar to the ones that affected other early people as well as modern humans.

The polymerase chain reaction has given scientists the ability to create billions of clones of a specific sequence of DNA from only a few molecules, leading to rapid expansion in the field of ancient DNA analysis. Unfortunately, PCR amplification is a process that is prone to contamination from outside sources, and is also often very costly (Mulligan, 2006). Since PCR amplification involves creating new, synthetic DNA samples from a small

Page 14

number of progenitor molecules, contamination by exogenous DNA in an initial PCR cycle can result in the contaminant DNA being exclusively amplified. Especially since modern DNA is less likely to be damaged or exist in trace amounts, there is a risk that it will be preferentially amplified over the endogenous DNA (Mulligan, 2006).

Despite its shortcomings, ancient DNA analysis has provided many valuable insights into what life was like for the humans of the past. Used in conjunction with other methods and sources of historic data, such as the fossil record and radiocarbon dating, it has allowed scientists to create a more complete picture of the prehistoric world. Not only can we now know where our early ancestors came from, we can also find out what they ate and what diseases and parasites they carried, where they moved and settled, when they began to domesticate animals, and how their languages developed. The body of knowledge that ancient DNA analysis has given rise to has the potential to answer important questions about how our modern world came to be.

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