DNA and archeology

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Introduction

Recent developments in the science of genetics have had a profound impact on our knowledge about prehistory. The study of DNA has contributed to our knowledge in two important ways: by studying genetic diversity in modern populations patterns of prehistoric movements and gene flows can be discerned; and, by studying ancient DNA from archaeological specimens of pre-modern hominids we can arrive at important new knowledge about the earliest stages of human evolution and the evolutionary relations between us and our closest relatives in the genus *Homo*. As development advances of better techniques for retrieving, sampling and analyzing ancient and modern DNA we are likely to acquire much new knowledge about our earliest history.

DNA

The molecule called DNA is located in the cells of all living things. It controls the genetically determined part of human development through its complex sequence of base pairs that encode for the production of particular proteins. In human cells, DNA is found in the cell nucleus and in the mitochondria. Each cell contains only two copies of the chromosomal DNA, but many copies of the mitochondrial DNA. In the nucleus, DNA is found in the two chromosomes. This DNA is inherited from both parents and is recombined so that it is neither identical to the mothers or fathers DNA.

Mitochondrial DNA however, in the vast majority of cases is only inherited from the mother and is identical to the mother's Mitochondrial DNA, except for mutations. It is the tracking of mutations through time that allows the construction of phylogenetic trees of human populations.

When a mutation occurs in the transmission of DNA from parents to child it exchanges one base pair in the DNA sequence with another. This new sequence is in turn transmitted to the child's own offspring. Such a sequence that varies from its predecessor by a single nucleotide polymorphism is called a haplotype, and the group of people descended from the first ancestor with this mutation and who share this particular new sequence is called a haplogroup. Since Mitochondrial DNA is inherited directly from mother to daughter, determining the mitochondrial DNA haplotype makes it possible for us to determine whether two individuals are descended from the same woman. This is the technique that leads to the proclamation of the discovery of 'Mitochondrial Eve', a woman from which all living people on the earth are descended. It is also possible to track ancestry through the male line by using Y chromosomal DNA, as this is inherited from father to son. Haplotype analysis of DNA shows group ancestry of human populations and can be correlated with geographic data so as to track human migrations in prehistory.

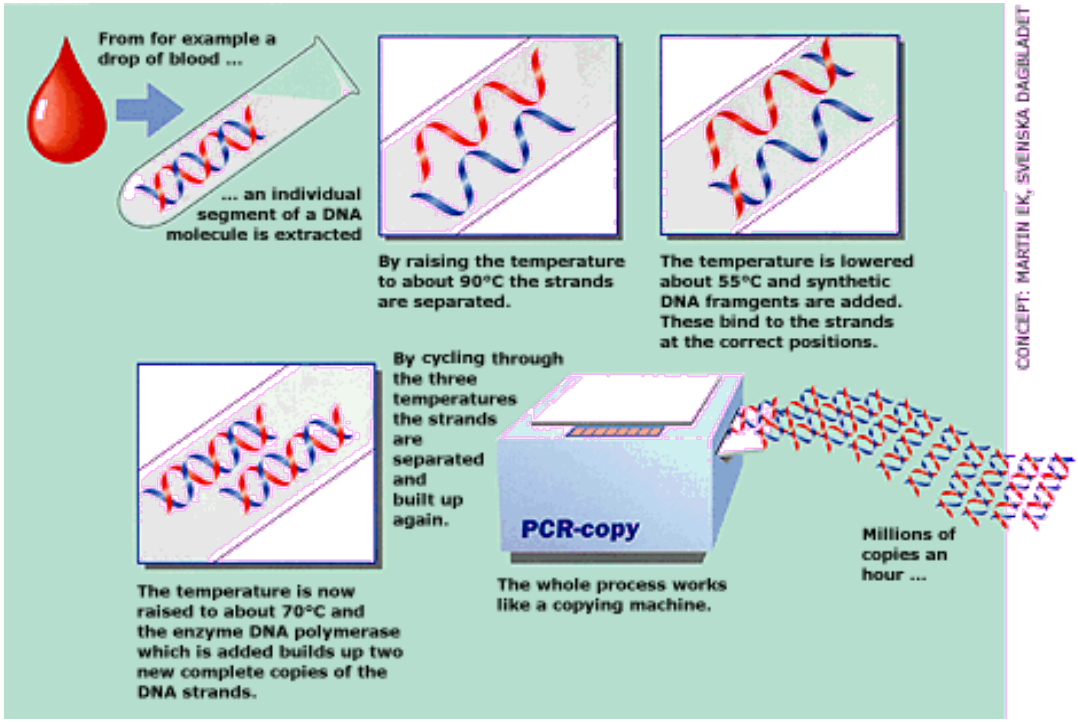
A gene is a sequence of base pairs on the DNA that code for the production of a single protein. Specific genes can be shown to correlate with specific somatic conditions or with specific abilities or physical traits. Knowledge about which genes an individual has can tell us about specific genetically determined physical traits that the individual exhibits. Or more precisely, since the presence of the

gene does not necessarily imply that the trait it codes for has been fully developed, knowing about the presence of specific genes, suggests traits that an individual is at risk of developing or passing. This kind of genetic analysis is not in wide use in archeology, because it can only tell something about an individual and very little about the group to which he belonged. However, in some cases such as when a gene is shown to correlate to faculties or traits considered important to all humans the presence or absence of a single gene may be considered important. For example the gene FOXP2 has been shown to correlate with the development of speech in modern humans, and if we assume that it had the same correlation in the far past, its presence in for example early hominids can be seen as suggestive of their having a speech faculty similar to that of modern humans (Krause et al. 2007). Analysis of specific genes can also be used to determine particular somatic conditions in historical humans that are of particular interest – Tutankhamen, for example (Hawass et al. 2010).

The Technology: Polymerase Chain Reaction

The prerequisite for being able to analyze ancient DNA was the development of the technology called Polymerase Chain Reaction. This is a technology that makes it possible to detect even very small trace amounts of DNA by amplifying particularly interesting sequences. This is done by placing a sequence of DNA to be copied, called a primer, in a buffer solution containing the polymerase enzyme and base nucleotides, which is then passed through a cycle of temperature changes, causing the enzyme to produce copies of the primer out of the free nucleotide bases. The first phase called denaturation the DNA helix is decomposed into single strands. In the annealing phase the primer sequence hybridizes to its recognition sites. And in the final elongation phase the polymerase reacts to

form copies of the primer sequence. The cycle is then repeated to multiply the primer sequence to enable sampling (Hummel 2003).



DNA specific formation processes and their influence on research design

When using DNA as archeological data a number of formation processes that are specific for DNA must be taken into account. These include issues of degradation of the DNA itself occurring over time, and also contamination by DNA from other organisms.

Even under the best conditions of preservation, which for DNA means a dry, cold environment with a stable Ph value, DNA degrades over time. The DNA strands simply break into short pieces, making it difficult to reconstruct complete sequences. Another process, cytosine deamination, chemically degrades the basepairs changing the C and G bases into T and A respectively. This process further complicates the picture resulting from PCR.

While degradation can usually be off set by making more samples, contamination with foreign

DNA is the most serious factor. Contamination may originate from humans handling the remains before arrival at the laboratory, but commonly contamination comes from microbial infections of the human remains. As the PCR process amplifies the sample the error from contamination is also multiplied – and as more PCR cycles are added the error becomes statistically greater. In some sample of Neanderthal DNA, contamination by microbes has accounted for as much as 95% of the sample. The best way to avoid this is to minimize the risk of contamination by human DNA as this is the most difficult to discern as foreign in the resulting sample. The way in which the level of contamination is determined is by identifying loci in the DNA to be sampled that are rare in modern human populations (Pääbo et al. (2010) use loci that differ from 99% of modern human populations) and determine how great a percentage of the sampled DNA carries that rare allele, discounting those instances where a difference may be due only to cytosine deamination. Other ways to detect contamination is to start by determining the sex of the sample and establish how much of the DNA is of the wrong sex, and to genotype all of the researchers who will be handling the sample's so that their DNA can be recognized in the result and discarded.

Because of the extremely complex nature of using ancient DNA, the limited sampling material is normally destroyed in the process, and the fact that it is also a very costly procedure that often does not produce useable results, DNA studies should only be applied where it is both crucial to answering particular research questions and where the sampling material is judged to be of sufficient quality to produce a result (Mulligan 2006).

Possible applications:

Studies of ancient DNA can be used to answer different kinds of questions. By comparing an individual sample with a specific sample group, an individual can be either matched to or exlcuded form genetic membership of a particular group. By showing that two chronologically successive populations are either genetically similar or genetically different DNA evidence can be used to argue for or against interpreting a change in the archeological record as a replacement of one group by another. If there is a question of whether a population was large and diverse or small and inbred the degree of diversity or inbreeding can be assessed. If it is to be determined whether a single individual found in a context with other individuals represented a foreign element in the group, a DNA test showing the individual to belong to a completely different haplogroup than the other individuals could suggest that he was an outsider. In group burials, DNA tests can determine kinship relations between the individuals. The sex of an individual that can not otherwise be assigned to a sex category can also be determined genetically. Analysis of DNA found in dried fecal matter can be used to determine which kinds of plants and animals an individual consumed (Mulligan 2006).

Genetic prehistory

The tools of modern genetics can be applied to the study of the prehistory from two angles. It can be applied to the genetic variation within the living human population in order to discern patterns of migrations by correlating geographical space with the phylogenetic haplogroup tree. And it can be applied directly to ancient DNA found in archeological excavations to determine the genetic makeup of ancient hominids in order to know more about their physical traits and about their genetic relation to

modern humans. Arguably the most important study of prehistory through modern genetic variability is Cavalli-Sforza's studies that provided important evidence in favor of our current understanding of human migrations out of Africa. Yet the most spectacular application of genetic methods to ancient DNA is Svante Pääbo's Neanderthal Genome Project, an attempt to map the entire Neanderthal genome - a project that can potentially solve most of the questions regarding Neanderthals and their relation to modern humans.

Prehistoric migrations

The use of genetic evidence for migrations was prompted by the observation that human blood types are not equally distributed throughout the world, and that some of this variation coincides with human ethnic or linguistic boundaries. With the development of population genetics the picture painted by using blood types as a proxy for genetic heritage could achieve much greater detail.

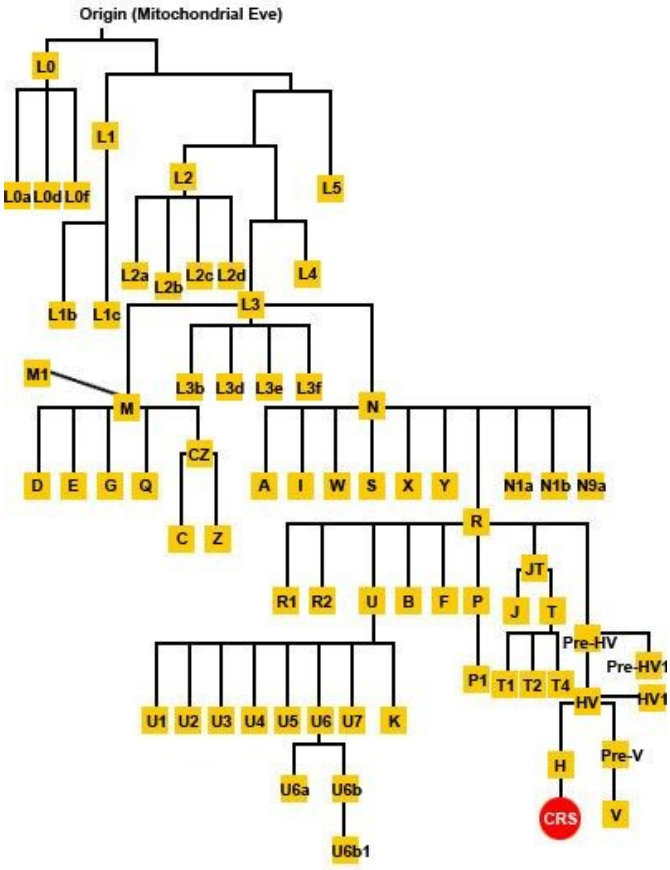


Illustration 1: Human mtDNA haplogroups since 'mitochondrial Eve' in the order of their diversion.

One of the first important insights gained from mtDNA analysis is that all modern humans share a common female ancestor in Africa about 150-200.000 years ago. The discovery this 'Mitochondrial Eve' was highly publicized and constituted

an important piece of evidence for the 'out of Africa' hypothesis of human origins. Dating based on mtDNA is not very certain and depends on the researchers' assumptions about the rate of mutations over time.

Other genetic evidence in favor of the recent African origin of all modern humans is the fact that there is as much genetic variation within African populations as there is in all human populations outside of Africa. This conclusion has been repeatedly established by studies of mtDNA, as well as Y and X chromosomal DNA. In all studies there is a strong negative correlation between the geographical distance of a population from Africa and its genetic diversity. The migration has been dated to around 65,000 years ago. DNA evidence also suggests that the out of Africa migration did not follow a path through the Levant as often assumed, but rather a southerly route into Asia. This has been supported by archeological finds in the Red Sea region dating to around 125,000 BP (Hofreiter 2010:547).

The first colonization of Europe and the arctic has been dated to around 40,000 years ago. Y-chromosomal DNA evidence suggests a date for the arrival of humans in the Americas anywhere between 10-20,000 years ago. This suggests human occupation of the Americas prior to the rise of the Clovis lithic culture, but makes the earliest proposed dates of human arrivals some 25,000 years ago. The dating of DNA on fossilized human excrement in the 5 Mile Point caves of southern Oregon to 12,300 BP (Gilbert, Jenkins et al. 2008); using mtDNA, scholars showed that these pre-Clovis humans belong to the A2 and B2 haplogroups common in Native American populations today. Studying the DNA evidence for the peopling of Oceania it has been concluded that Polynesian

populations have ancestry both in Asia and in Melanesia. The number of haplotypes of Melanesian origin is larger when testing Y-chromosomal DNA than when testing mtDNA, which could be suggestive of intermarriage between Asian and Melanesian groups following a matrilocal system, similar to that found in many Polynesian groups today. Genetic diversity of Polynesian populations decreases along an axis from west to east, further suggesting that the area was populated by eastward movement (Hofreiter 2010:549).

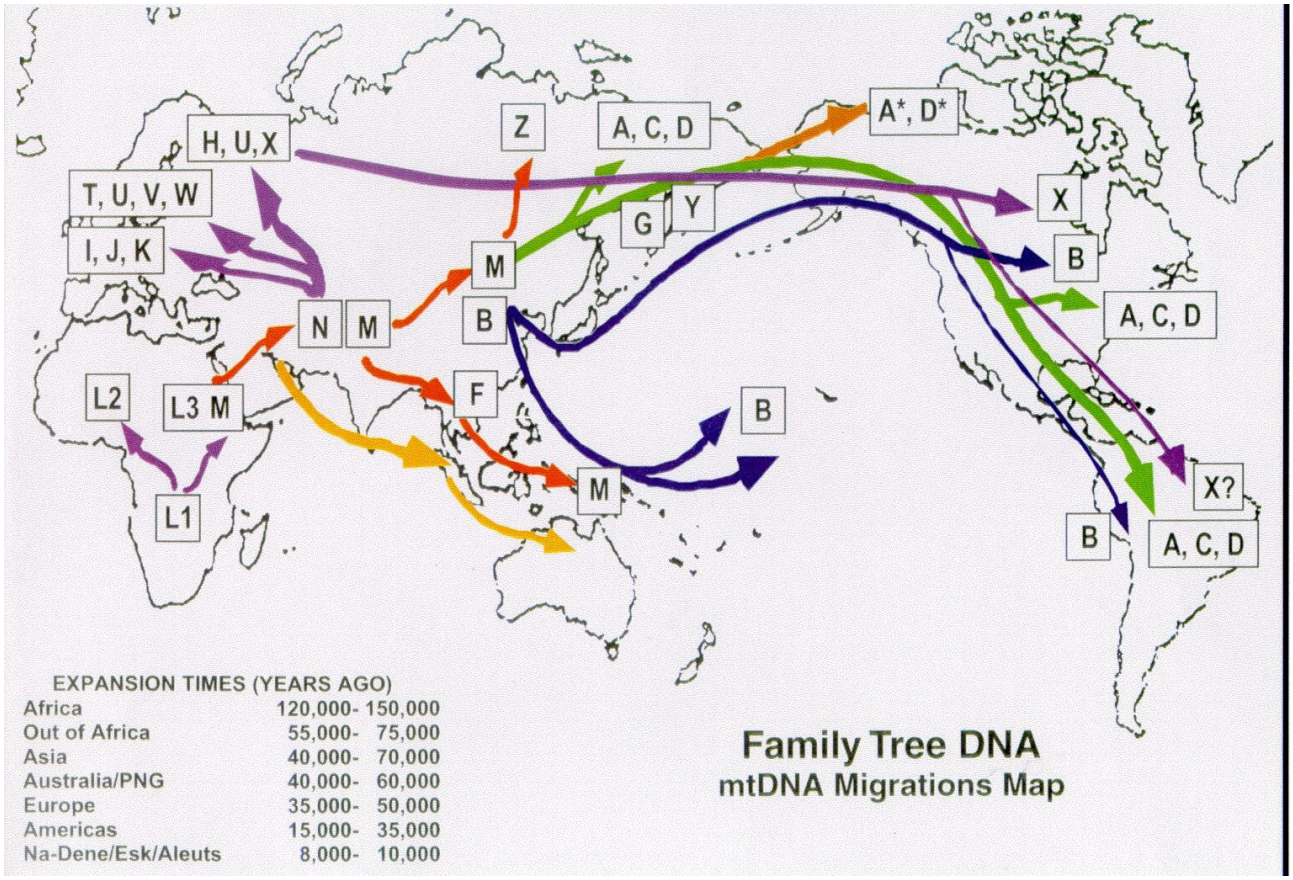


Illustration 2:Correlation of haplogroups with geography showing the probable path of human migrations out of Africa.

Studying the genetic diversity of modern populations of domesticated plants and animals can also give clues about the earliest patterns of domestication. The domestication of wheat, rye, barley and the cow

goes back to the fertile crescent of the Middle East around 11,000 BP. DNA evidence also show a separate domestication of the Indian Zebu cattle around the same time. DNA evidence suggests that in accordance with common wisdom the earliest domesticate animal was the dog, around 15,000 years ago in China.

The study of Neanderthal DNA

The study of the Neanderthals occupies a special place in paleo-anthropology. The fact that Neanderthals were the first pre-modern hominid to be discovered in 188x, and that they are the single closest relative of modern man, means that speculation and theorizing about Neanderthals and their relation to ourselves has had more than a century to flourish. Where they our forefathers? Did they have relations with early modern humans? Which kind? If they weren't like us how did they differ? In which ways where they similar?

An example of the kind of debate sparked by Neanderthal remains is the Binford-Bordes debate in which Bordes argued that the typology of different tools of the Mousterian complex of the middle paleolithic showed cultural variation among Neanderthals. Binford argued that rather the typology reflected different tool functions.

Illustration 3:A replica Neanderthal skull performing Hamlet with Swedish geneticist Svante Pääbo.

The question went to the core of the question of Neanderthal cognitive functions (Trigger 2006:xx).

The Neanderthal occupy a special place in paleo-anthropology in a further sense: they are the only pre-modern human species from which it has been possible to extract DNA. The first tests of Neanderthal mitochondrial DNA extracted from the specimen found in 1856 were published in 1997. Krings et al. concluded that Neanderthal DNA fell outside of the variation in modern humans, suggesting that Neanderthals were a completely different branch of the genus homo and had gone extinct without passing DNA on to modern humans. This conclusion was further corroborated in a 2004 study, which indicated that Neanderthal contributions to modern human DNA had to be less than 5% (Serre et al.). Technical developments in the past ten years has made it a feasible undertaking to sequence the entire Neanderthal genome, and geneticist Svante Pääbo from the Max Planck Institute of Evolutionary Anthropology has begun this task. In 2010 his team published a draft genome sequence of 4 billion nucleotides from three Neanderthal individuals found in Vindija cave in Croatia. The study concluded that, contrary to earlier reports, there are traces of Neanderthal DNA in modern humans on the order of 1- 4% of the genome. However, this contribution is only found in populations outside of Africa, the Neanderthal contribution is of equal size in both European, Asian and Oceanian populations. This leads the researchers to suggest a scenario of gene flow into modern humans after their having left Africa, but before their further diversification. Just as important, the project only found 78 nucleotide pairs in which all humans have an innovative variant relative to the Neanderthals - this suggests that what separates us as a species from the Neanderthals can be summed up in a very limited number of mutations.

In these ways DNA studies can shed new light on how our homo sapiens forefathers related to their Neanderthal cousins, and by extension the degree to which they perceived Neanderthals as either different from or equal to themselves.

Conclusions:

The study of ancient DNA has revolutionized archaeology by providing acces to kinds of information that were previously completely outside of the scope of Archaeology – information about individuals and their ancestry and group memberships and even more importantly about human evolution. Svante Pääbo happily states that "this is the golden age of Genetics" and as the field advances, doubtles many more possibilities for application in Archaeology will become possible.

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