Ancient DNA studies of human evolution

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CONTENTS PAGE

Abstract

The recovery of ancient DNA from preserved human remains provides a unique opportunity to directly study the impact of past events on prehistoric populations. The past event which is of interest to this thesis is the development of agriculture in Europe, also termed the 'Neolithic Revolution'. This event resulted in the transition from a hunter-gatherer to agriculturist lifestyle. The adoption of farming altered many aspects of human culture, including how societies are structured, where we obtain our food from and which diseases burden human populations. This thesis aimed to assess how the arrival of farming in Europe influenced the population structure and health of both prehistoric and modern human populations through the analysis of ancient genetic data. As human remains are a valuable resource, it was also of interest to evaluate the methods used to recover degraded genetic material from preserved remains.

The affect of common sampling techniques on the recovery of DNA from skeletal remains was assessed by quantifying mitochondrial (mt) DNA in ancient human and bovid samples (n=42). The key finding is that drilling of bone and teeth under standard sampling conditions can reduce DNA amounts by over five orders of magnitude. Also, it was shown that current approaches do not sample the most DNA rich tissues (the cementum layers of tooth roots) and have failed to show where DNA survives accurately and how it degrades over time. The results promote a standardised approach to the genetic analysis of human remains.

To investigate whether a single, continuous population has existed in Central Europe since the Early Neolithic until today, a dataset of ancient mtDNA from Early Neolithic and Late Neolithic/Early Bronze Age cultures from Central Europe was compiled (n=108). Population continuity from the introduction of agriculture in the Early Neolithic until today was found to be an unlikely model of demographic history, as indicated by the results from population genetic and coalescent analyses of the ancient mtDNA, which was compared to extant populations from the same region. Internal population changes in Europe between the Early Neolithic and Late Neolithic/Early Bronze Age appear to have contributed substantially to the population structure of extant Central Europeans, as all the Late Neolithic/Early Bronze Age cultures examined (Bell Beakers, Corded Ware or Unetice) were found to be more likely ancestors of modern Central Europeans than either of the Early Neolithic cultures (LBK and Rössen) investigated. Haplogroup distributions suggest that Palaeolithic mtDNA haplogroups which were infrequent in the Early Neolithic, such as haplogroups H and U, became more frequent during the Late Neolithic. These findings alter the traditional view of which past settlement events in Europe have contributed to the current genetic structure of the continent and demonstrate that past events which are not associated with major climatic or economic

changes in human history, such as internal migrations in Europe during the Late Neolithic/Early Bronze Age, can also substantially alter population structure.

Ancient pathogen DNA was used to explore how farming affected the health of prehistoric humans. This required the identification of a genetically preserved source of human-associated microbiota. Dental calculus samples recovered from human remains, which dated between the Neolithic to Medieval period, were found to contain ancient oral microbial DNA. Preserved microbial DNA was extracted from calculus samples (n=28) and used to create PCR amplicon libraries of the 16S rDNA gene. Phylogenetic analyses revealed that the diversity of oral microbiota in early agriculturists was significantly higher than that found in modern Europeans. During the early stages of agriculture there was a dominance of bacteria involved in periodontal disease. This suggests periodontal disease was an early consequence of an agricultural diet, which was high in carbohydrates and low in processed sugar and grain. In contrast, modern Europeans have a restricted suite of bacteria, mainly associated with tooth decay. The current dominance of decay associated taxa may be because of the introduction of refined sugar and grain to the diet which occurred during the Industrial Revolution (150-200 years ago). The results highlight the impact of changes in human ecology to the co-evolved mutualism between humans and their microbiota.

By using three disparate approaches to research, this thesis has provided original insights into the lives of prehistoric humans. The results have demonstrated that accompanying the early stages of agriculture, there were changes in population structure and the presence of bacteria associated with oral diseases. The application of ancient DNA techniques to examine dental calculus, enabled for the first time, the direct analysis of the past state of human associated microbiota, which will have multiple implications for the study of prehistoric human lifestyle. This thesis also contributed more generally to the field of ancient DNA by highlighting that commonly used sampling techniques were detrimental to the recovery of DNA from skeletal remains. By combining three different approaches to ancient DNA research, this thesis has provided original insights into the lives of prehistoric humans, which advances the fields of archaeology, anthropology, modern and ancient population genetics, forensics and medical research.

Declaration

This work contains no material which has been accepted for the award of any other degree or diploma in any university or other tertiary institution to Christina Jane Adler and, to the best of my knowledge and belief, contains no material previously published or written by another person, except where due reference has been made in the text.

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The published works in my thesis include:

Adler, C J, Haak, W, Donlon, D & Cooper, A 2011. Survival and recovery of DNA from ancient teeth and bones. *Journal of Archaeological Science,* 38**,** 956-964.

Jones, C. 2011. Researchers to drill for hobbit history. *Nature*, doi:10.1038/news.2011.702.

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