2022 ancient DNA symposium

Date: November 29, 2022

Time: 09:00-16:30

Venue: Large auditorium, Swedish museum of natural history, Frescativägen 40, Stockholm

Online attendance: Link distributed separately

Organizers: SciLifeLab Ancient DNA unit, SciLifeLab National Genomics Infrastructure and the Centre for Palaeogenetics

Program

09:00 Introduction to ancient DNA research
Anders Götherström, Dept. of Archaeology and classical studies, Stockholm University, Sweden

09:10 *Keynote presentation:* Changing social dynamics in the light of ancient DNA: the Southwest Asian evidence
Mehmet Somel, Dept. of Biological Sciences, Middle East Technical University, Turkey

09:50 Infectious diseases in the Viking Age town of Sigtuna
Zoé Pochon, Dept. of Archaeology and classical studies, Stockholm University, Sweden

10:10 African history – inferences from modern and ancient DNA
Carina Schlebusch, Dept. of Organismal Biology, Uppsala University, Sweden

10:30 Coffee and consultations

11:00 The European arrival and admixture history of ancient cats
Greger Larson, School of Archaeology, Oxford University, UK

11:20 When Archaeology ask Questions with Key Genetic Signature!
Caroline Ahlström Arcini, the Archaeologists, National Historical Museums, Sweden

11:40 Ancient DNA from foragers, farmers and herders in Neolithic Scandinavia
Helena Malmström, Dept. of Organismal Biology, Uppsala University, Sweden

12:00 Lunch

13:20 Keynote presentation: Reconstructing past animal communities from ancient environmental DNA
Pete Heintzman, Dept. of Geological Sciences, Stockholm University

14:00 Infrastructure support for ancient DNA research
Tom Martin, SciLifeLab National Genomics Infrastructure, Sweden
Magnus Lundgren, SciLifeLab Ancient DNA unit, Sweden

14:20 Phylogenetic history of collared lemmings (*Dicrostonyx* sp.)
Edana Lord, Dept. of Zoology, Stockholm University, Sweden

14:40 Coffee and consultations

15:10 Herbarium evolutionary holo-genomics using historic herbarium specimens
Michael Martin, Dept. of Natural History, University Museum, Norwegian University of Science and Technology (NTNU), Norway

15:30 Using deep-time genomic data to understand evolutionary processes in mammoths
Camilo Chacon-Duque, Department of Archaeology and Classical Studies, Stockholm University, Sweden

15:50 Optical duplicates, optical duplicates: the effect of loading concentration on library complexity
Marianne Dehasque, Dept. of Zoology, Stockholm University, Sweden

16:10 Identifying admixture and ghost lineages in (ancient) genomes
Tom van der Valk, Dept. of Bioinformatics and Genetics, Swedish museum of natural history

16:30 End of symposium

Abstracts

Changing social dynamics in the light of ancient DNA: the Southwest Asian evidence
Mehmet Somel, Dept. of Biological Sciences, Middle East Technical University, Turkey

I will present patterns of changing social dynamics through the Holocene in Southwest Asia revealed by ancient genomics. One focus will be genetic kinship inferred among co-buried individuals, which informs on traditions related to co-residence and/or burial rites. Our analyses show that the role of genetic kinship in co-burials appears to have varied among Neolithic communities in Anatolia from the early (c.9000-7000 BCE) to the later Neolithic (c.7000-6000 BCE), which likely further changed in the post-Neolithic period. Another focus involves the dynamics of regional population movement and admixture, inferred from f-statistics and qpAdm modelling. Our work suggests an expanding range of population movement from the mid-Holocene onward. This expansion is also paralleled by a relative increase in male sex bias in population movement.

Infectious diseases in the Viking Age town of Sigtuna
Zoé Pochon, Dept. of Archaeology and classical studies, Stockholm University, Sweden

Infectious diseases are not easy to detect in paleopathology. Long-lasting diseases such as tuberculosis, leprosy, syphilis and brucellosis can leave visible traces on the bones, but this is not the case for most infectious diseases. With the development of next-generation sequencing techniques, it is now possible to recover the genomes of ancient humans but also of the pathogens they were hosting at the time of death such as Yersinia pestis, the agent of plague.
Here we focus on the impact of urbanization on infectious disease with the Viking Age town of Sigtuna as a case study. Over 40 individuals were subjected to shotgun sequencing and screening for potential pathogens using a new ancient metagenomics workflow developed in collaboration with the National Bioinformatics Infrastructure Sweden (NBIS), called aMeta and available on GitHub. Preliminary results are promising, with the detection of a foodborne pathogen and two pathogens causing sexually transmitted infections. This is an ongoing project and we would appreciate feedback on preliminary results and suggestions for downstream analysis and verification steps.

African history – inferences from modern and ancient DNA
Carina Schlebusch, Dept. of Organismal Biology, Uppsala University, Sweden

Genetics helped to establish Africa as the birthplace of anatomically modern humans. However, the history of human populations in Africa is complex and includes various demographic events that influenced patterns of genetic variation across the continent. Through genetic studies of modern-day, and most recently, ancient African genetic variation, it became evident that deep African history is captured by connections among African hunter-gatherers, displaying the deepest population divergences. Furthermore, it was shown that Holocene population expansions and large-scale migrations, linked to food-production

and technological innovation, had a significant influence on the distribution of current-day Africans. These later population movements disrupted pre-existing population distributions and complicate inferences regarding deep human history. With the increased availability of full genomic data from diverse sets of modern-day and prehistoric Africans we have more power to infer human demography and the next few years will be exciting for investigating African genetic history.

The European arrival and admixture history of ancient cats
Greger Larson, School of Archaeology, Oxford University, UK

While the global population of domestic cats exceeds 400 million individuals, the European wildcat exists only in a fraction of its former distribution. This is especially the case in Britain where the critically endangered *F. silvestris* is restricted to small populations in Scotland. To establish the timing and consequences of the domestic cats' arrival into Europe, we combined radiocarbon dating with analyses of zooarchaeological, genetic, historical and iconographic evidence. Our results show that domestic cats derived from *F. lybica* spread to northern Europe during the Iron Age and that successive populations of domestic cats with unique mitochondrial signatures were subsequently introduced during the Roman and Viking Ages. The analysis of 14 ancient nuclear genomes and 31 modern genomes demonstrates that, despite cohabitating for at least 2,000 years on the European mainland and in Britain, domestic cats possess less than 10% of their ancestry from European wild cats, and ancient European wild cats possess little to no ancestry from domestic cats. This strong reproductive isolation, however, is now being eroded as a result of anthropogenic activities. Using genomic data derived from museum samples we reconstructed the decline and severely hybridised nature of the Scottish wildcat population over the last 70 years. Our analyses also revealed the domestic ancestry present in modern wildcats is over-represented in immune function genes, likely implying that introgression in this region is currently providing a protective function against diseases associated with the introduction of domestic cats. Combined, these studies reveal how the introduction of domestic cats can be used to track human dispersal and how human-mediated hybridisation poses a severe threat to wild populations.

When Archaeology ask Questions with Key Genetic Signature!
Caroline Ahlström Arcini, the Archaeologists, National Historical Museums, Sweden

Geneticists have developed methods to retrieve genetic data from osteological materials. For archaeologists and osteologists, the opportunity to participate and ask questions has so far only been possible if you have had contacts with researchers in the field. In 2019 a new opportunity arose when SciLifeLab opened as a part of the national resource for ancient DNA. So, what are the questions that archaeologists and osteologists are interested in? During my nearly 40 years as an osteologist, a standing question has been to what degree individuals buried at a site is genetically related and what kinship structures was at hand. My presentation today will share with you the results of some kinship studies and what knowledge is added to the understanding of ancient societies.

Ancient DNA from foragers, farmers and herders in Neolithic Scandinavia
Helena Malmström, Dept. of Organismal Biology, Uppsala University, Sweden

The Neolithic archaeological record in Scandinavia show interesting material culture variation. The first study using genome wide ancient DNA data to investigate how well this variation correlates with genetic variation was published only 10 years ago. Since then, we have been able to paint the broad strokes on the archaeogenetic landscape of Neolithic Scandinavia. This lecture will summarize what we this far have learned about the people of the Funnel Beaker Culture, the Pitted Ware Culture and the Battle Axe culture.

Reconstructing past animal communities from ancient environmental DNA
Pete Heintzman, Dept. of Geological Sciences, Stockholm University

Animals are key ecosystem components but are often neglected in sediment-based palaeoecological reconstructions. Inferences on animal presence and activity are instead usually obtained indirectly from proxies that can only be used to infer broad taxonomic groups. Ancient environmental DNA (aeDNA) offers an alternative approach that is taxonomically unconstrained and can allow for robust identifications to the species and, in some cases, population level. However, animal-based reconstructions are challenging, in part due to low biomass on the landscape resulting in low aeDNA input. With the increasing sensitivity of aeDNA methods, both in terms of improved recovery and computational processing, our ability to reconstruct animal communities by capturing a greater proportion of taxa is allowing for more nuanced ecological reconstructions. In this talk, I showcase recent examples and advances. Although challenges remain, particularly regarding false negatives, these are likely to be resolved with emerging and increasingly sensitive methods. Together with its recently demonstrated potential for population-level reconstructions, aeDNA is poised to revolutionize our understanding of Quaternary animal palaeoecology.

Infrastructure support for ancient DNA research
Tom Martin, SciLifeLab National Genomics Infrastructure, Sweden
Magnus Lundgren, SciLifeLab Ancient DNA unit, Sweden

Analysis of ancient DNA is a powerful source of information about the past, especially when combined with other analyses. Ancient DNA can provide information about *e.g.*, human migrations, societal structure and culture, development of agriculture, history of pathogens, evolution, biodiversity and ecology. Swedish national research infrastructures can provide extensive support for ancient DNA analysis. The SciLifeLab Ancient DNA unit supports project discussions, sampling, DNA extraction and sequencing library preparation. The SciLifeLab National Genomics Infrastructure (NGI) provide a range of high throughput Next Generation DNA and RNA sequencing technologies, as well as genotyping. Data processing and downstream data analysis is supported by the Ancient DNA unit and the SciLifeLab Bioinformatics platform, the National Bioinformatics Infrastructure Sweden (NBIS).

Phylogenetic history of collared lemmings (*Dicrostonyx* sp.)
Edana Lord, Dept. of Zoology, Stockholm University, Sweden

Collared lemmings (*Dicrostonyx* sp.) are key Arctic species present today in the tundra of Siberia and North America. Mitogenome data suggests that the Eurasian and North American collared lemmings diverged around 200 thousand years ago. The extent of post-speciation gene flow that may have occurred between collared lemmings is currently unknown, however a recent study identified conflicting mitochondrial and nuclear phylogenies within the North American collared lemming species. Here, we generated ten modern and eleven ancient genomes from the Eurasian collared lemming (*D. torquatus*), along with eight modern genomes from three North American *Dicrostonyx* species (*D. groenlandicus*, *D. hudsonius*, and *D. richardsonii*). We additionally sequenced the genome of a circa 300-thousand-year-old mummified Siberian *Dicrostonyx*. We aimed to evaluate the phylogenetic relationships between the collared lemming species, and furthermore to examine whether post-speciation gene flow occurred during the Late Pleistocene.

Herbarium evolutionary holo-genomics using historic herbarium specimens
Michael Martin, Dept. of Natural History, University Museum, Norwegian University of Science and Technology (NTNU), Norway

The world's herbaria contain millions of specimens stretching back to historical times. Their excellent preservation and meticulous curation have been used for studies of taxonomy, morphology, phenology, parasitology, and more. Recent advances in DNA sequencing mean these collections can also be viewed as repositories of genomic information. I present several case studies in which genomic information from historical herbarium collections has been used to elucidate the evolutionary consequences of the introductions of plants and pathogens to Europe.

Using deep-time genomic data to understand evolutionary processes in mammoths
Camilo Chacon-Duque, Department of Archaeology and Classical Studies, Stockholm University, Sweden

Temporal genomic datasets spanning tens of thousands of years have improved our understanding of diverse evolutionary processes. However, most processes shaping current-day biodiversity stretch back into the Early Pleistocene – about 2.6 million years ago (Mya). Last year my colleagues at CPG reported, for the first time, the retrieval of DNA from >1Mya (mammoth) specimens (van der Valk *et al*. 2021) and used this information to explore how mammoth species relate to each other and how they adapted to changing environments. To further characterise mammoth evolution, we are expanding this “deep-time” dataset by recovering genomic data from specimens collected across Eurasia and north America, ranging from 100 Kya to >1Mya. I will describe this ongoing work, which will allow us to gain more insight into both the transition from steppe to woolly mammoths and the hybrid nature of north American mammoths. These findings, along with the associated technical and analytical improvements, will serve as a benchmark to study other ancient taxa.

Optical duplicates, optical duplicates: the effect of loading concentration on library complexity
Marianne Dehasque, Dept. of Zoology, Stockholm University, Sweden

Optimizing sequencing efficiency remains a major challenge in palaeogenomic research. Sequencing duplicates are a major concern, as they can drastically inflate sequencing costs. Many methods aim to minimize duplicates originating from PCR amplification. An often-overlooked source of duplicates, however, are optical duplicates. These duplicates arise from underloading an Illumina flow cell and can severely affect sequencing efficiency. Overloading the flow cell, on the other hand, is associated with lower sequencing quality. To test the effect of loading concentration on final sequencing efficiency of ancient DNA, we sequenced the same library pool at four different loading concentrations. We found a positive correlation between loading concentration and sequencing efficiency, even when accounting for DNA quality. We therefore advise to use loading concentrations of 400 to 500 pM in future ancient DNA sequencing runs using Illumina S4 sequencing technology.

Identifying admixture and ghost lineages in (ancient) genomes

Tom van der Valk, Dept. of Bioinformatics and Genetics, Swedish museum of natural history

The sequencing of ancient DNA has reshaped our knowledge on human history. Inspired by the findings of ancient admixture events between hominids, researchers have developed sophisticated methods to infer past events using genomic data. However, it remains challenging to identify potential gene-flow events from unknown origins. In this talk I will discuss several recent studies on humans, chimpanzees and woolly mammoths, in which ancient admixture events from unknown origin (“ghost admixture'') were identified and modelled using only modern genomic data."