



## Abstracts

### **Ancient DNA and Human Evolution** Public Symposium • Friday, April 29, 2016

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(CARTA)



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#### ***The Landscape of Archaic Ancestry in Modern Humans*** **Sriram Sankararaman, University of California, Los Angeles**

One of the major discoveries to come out of the analyses of the genomes of archaic humans such as Neanderthals and Denisovans is that of admixture between these archaic and present-day human populations. We now know that non-African populations today trace about 2% of their ancestry to a population related to the Neanderthals while populations from Australia and New Guinea carry additional ancestry related to the Denisovans.

By viewing these admixtures as natural, genome-scale perturbations, we can begin to understand the impact of archaic ancestry on human biology as well as the genetic changes that were likely important for the modern human phenotype. I will describe methods that enable us to map the locations of archaic ancestry in present day humans and the applications of these methods to understand the impact of Neandertal and Denisovan ancestry in present-day humans.

#### ***Prehistoric Human Biology as Inferred from Dental Calculus*** **Christina Warinner, University of Oklahoma**

Advanced molecular methods have revealed a startling fact - that our bodies are not merely ourselves. Microorganisms comprise more than half of our cells, contain 99% of our genes, and perform vital functions in digestion, immunity, and homeostasis. Yet while we have made great strides in revealing the diversity, variation, and evolution of the human genome, we know surprisingly little about the microbial portion of ourselves, our microbiome. Recently, it has been discovered that dental calculus (calcified dental plaque) entraps a rich biomolecular record of the oral microbiome that preserves for thousands of years. Analysis of ancient dental calculus has revealed a wide range of commensal and pathogenic bacterial taxa, bacteriophages, human and dietary biomolecules, and an abundance of DNA "dark matter" from unknown and uncharacterized organisms. This talk explores how emerging ancient dental calculus research is changing the way we investigate the human past and how this is leading to a deeper understanding of human biology and evolution.

#### ***The Oldest Human DNA Sequences*** **Matthias Meyer, Max Planck Institute for Evolutionary Anthropology**

Over the past years, genomic sequences have been retrieved from hundreds of ancient human remains as well as those of extinct human relatives such as Neanderthals and Denisovans. However, due to constraints on DNA preservation in non-permafrost environments, genetic analyses of human fossils are usually limited to the Late Pleistocene (the last 125,000 years). Recently, improvements to DNA extraction and library preparation techniques have enabled the reconstruction of a mitochondrial genome sequence as well as small amounts of nuclear DNA sequences from the 400,000 year-old hominin remains from Sima de los Huesos, a cave site located within the archeological complex of Atapuerca, Northern Spain. The DNA found in these fossils is even more degraded and sparse than in any other ancient human fossil that has produced genetic data to date. I will discuss the technical difficulties one has to face when working with poorly preserved ancient material and the insights this work provides into human evolution in the Middle Pleistocene.

#### ***Neandertal and Denisovan Genomes and What They Tell Us*** **Kay Prüfer, Max Planck Institute for Evolutionary Anthropology**

The study of the genomes of our closest extinct relatives allows for insights into the recent evolutionary history of anatomically fully modern humans. The discovery of ancient remains with extraordinary preservation from a cave in the Altai mountains allowed us to generate high coverage genome sequences from two archaic human individuals, a Neandertal and a Denisovan. The analysis of these genomes reveals that they are more closely related to one another than they are to modern humans. At least two events of gene flow from these groups into modern humans have been found; Neandertal admixture has been detected into all out-of-Africa populations while Denisovan gene flow likely occurred to the common ancestors of Australians and New Guineans. The high quality sequences of a Denisovan and a Neandertal individual also allows for analyses of gene flow into the archaic lineages. We found evidence for two gene flow events into Denisovans: Gene flow from

Neandertals and gene flow from an hitherto unknown group of archaic humans. Taken together, these results show that gene flow was not uncommon among human groups in the Pleistocene.

***The Origins of Modern Humans in Africa***  
**Brenna Henn, Stony Brook University**

Over twenty-five years ago, geneticists sequenced mitochondrial DNA from a diverse sample of human populations and hypothesized that all humans have a common origin in Africa 200,000 years ago. The broad outlines of this hypothesis remain remarkably unaltered, but many details of our African origin continue to be elusive. After decades of advances in human genetics, we are no longer data limited - either in terms of DNA samples or genomic loci, but there is little consensus on many key issues. These include: where in Africa did the species originate? And what did the ancestral population look like physically? Additionally, is there a discordance between anatomically modern humans and behaviorally modern humans? Specifically, I will explore patterns of genetic diversity across Africa and models for modern human origins. In the past, I have argued for a geographic origin of *Homo sapiens* in southern Africa. I discuss whether genetic data is concordant with archaeological data and suggest directions for future research. In particular, phenotypic adaptation to regional environments within Africa may provide clues to the ancestral location of modern humans via tests for local adaptation to specific environmental features (e.g. ultraviolet radiation, malaria).

***A Map of Neandertal Genes in Present Day Humans***  
**Joshua Akey, University of Washington**

Anatomically modern humans overlapped in time and space with Neandertals, and genetic data has revealed that hybridization occurred. We have developed methods to identify Neandertal sequences that persist in the DNA of modern individuals, and applied it to whole-genome sequences from over 1,500 geographically diverse individuals. We leverage the catalog of surviving Neandertal sequences to show that admixture occurred multiple times in different non-African populations, characterize genomic regions that are significantly depleted of archaic sequence, and identify signatures of adaptive introgression. These data provide new insights in hominin evolutionary history and genomic regions that may harbor substrates of uniquely modern human phenotypes.

***The Phenotypic Legacy of Neandertal Interbreeding on Modern Humans***  
**Tony Capra, Vanderbilt University**

Many modern human genomes retain DNA inherited from interbreeding with archaic hominins, such as Neandertals, yet the influence of this admixture on human traits is largely unknown. We analyzed the contribution of common Neandertal variants to over 1000 electronic health record (EHR)-derived phenotypes in ~28,000 adults of European ancestry. We discovered and replicated associations of Neandertal alleles with neurological, psychiatric, immunological, and dermatological phenotypes. Neandertal alleles together explained a small, but significant fraction of the variation in risk for depression and skin lesions resulting from sun exposure (actinic keratosis), and individual Neandertal alleles were associated with specific human phenotypes, including hypercoagulation and tobacco use. Our results establish that archaic admixture influences disease risk in modern humans, provide hypotheses about the effects of hundreds of Neandertal haplotypes, and demonstrate the utility of EHR data in evolutionary analyses.

***Ancient European Population History***  
**Johannes Krause, Max Planck Institute for the Science of Human History**

Ancient DNA can reveal pre-historical events that are difficult to discern through the study of archaeological remains and modern genetic variation alone. Our research team analyzed more than 200 ancient human genomes spanning the last 10,000 years of Western Eurasian pre-history. We find direct evidence for two major genetic turnover events at the beginning and at the end of the Neolithic time period in Europe. Our data provide strong support of a major migration of early farmers spreading from Anatolia starting around 9,000 years ago bringing agriculture and domestic animals to Europe. Following their arrival, early farmers genetically admix with indigenous Europeans in the course of the coming 3,000 years. At the end of the Neolithic period, around 5,000 years ago, we find the first genetic evidence for another major migration event of people from the pontic steppe, north of the black sea, into the heartland of Europe. The newcomers practice pastoralism, are highly mobile, due to the widespread use of horses, wheels and wagons, and they may be responsible for the first spread of plague among human populations in Eurasia. We find that all modern European populations today are a genetic mixture of steppe pastoralist, early farmers and indigenous European hunter-gatherers in varying proportions. We furthermore find that due to genetic mixture and local biological adaptation, there are major changes in human phenotypes such as eye color, skin color and the ability to digest milk sugar through the course of the last 10,000 years.

## ***The Genetic History of the Americas***

**María C. Ávila-Arcos, National Autonomous University of Mexico**

Generally perceived as the carriers of the biological properties of species, genomes also harbor the demographic history of populations. By studying patterns of genetic variation of modern populations, it is possible to dissect the demographic events that occurred in their journey through time. In addition, recent advances in the field of ancient DNA have considerably broadened our knowledge of past events in the history of humans. Yet, a majority of these studies have focused on populations of European descent, leaving important gaps in our understanding of the genetic history of populations with different ancestry. In this regard, my research aims at combining both sources of genetic information, ancient and modern, in order to refine our understanding of the genetic history of understudied populations in Latin America, with an emphasis on Mexico. In this talk, I will present a review of the current state of knowledge on the genetic history of the Americas as revealed by ancient and modern DNA studies.