Dr. J. Sparkuhl Endowment Fund

Insights from ancient genomes

Thursday, October 7, 1:00pm - 2:15pm

The genomes of archaic and ancient modern humans offer a unique window into their histories. However, the sequencing and analysis of DNA from ancient humans is complicated by DNA degradation, chemical modifications and contamination. Recent technological advances have made it possible to retrieve and sequence DNA from bones and other remains found at archaeological excavations, and we have been able to reconstruct the genomes of several Neandertals. We have also identified, based on their genome sequences, a previously unknown extinct Asian hominin group related to Neandertals, who we call "Denisovans". The genomes of our extinct Neandertal and Denisovan relatives offer a unique opportunity to learn about the similarities and differences between us. We have used these archaic genome sequences to identify genetic changes that are unique to modern humans and not shared with Neandertals or Denisovans, and ongoing work aims to determine whether any of these genetic variants might underlie traits characteristic of modern humans. Further, we have also shown that the ancestors of some of us interbred with both Neandertals and Denisovans such that all people outside of Africa carry approximately 2% Neandertal DNA, and some populations, largely in Oceania, also carry DNA inherited from Denisovans. This introgressed DNA has been shown to have both positive and negative outcomes for present-day carriers: underlying apparently adaptive phenotypes such as high altitude adaptation, as well as influencing immunity and disease risk. In recent work we have identified Neandertal haplotypes that are likely of archaic origin and determined the likely functional consequences of these haplotypes using public genome, gene expression, and phenotype datasets.



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