

Tiny differences in our genes help shed light on the big picture of human history

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(PhysOrg.com) -- By examining very small differences in people's genes, scientists from Cornell University have developed a new tool for identifying big events in human history and pinpointing the origins of specific gene mutations. This research, published in the May issue of the journal *GENETICS*, helps shed light on times when the human population moved close to extinction and helps scientists close in on gene mutations that make some demographic groups more likely to develop diseases such as cancer, heart disease, diabetes, among others.

"We know that many diseases are caused by a combination of genetic and environmental factors," said Kirk E. Lohmueller, one of the researchers involved in the work from Cornell University. "To find the genes that contribute to disease, it's very helpful to know the demographic history of the population being studied. Accurate estimates of population events help inform the search for mutations that might have been helpful and necessary for survival at the time, but no longer necessary and potentially harmful today."

In their work, Lohmueller and colleagues confirmed the existence of a major decline in European populations (called a "bottleneck") 32,500-47,500 years ago. They used computer simulations to model the expected correlation among segments of DNA containing very small genetic mutations that only involve a single letter of the [genetic code](#) (called "single nucleotide polymorphisms" or SNPs). Prior to this development, methods used to identify major population events relied on the frequency patterns of individual SNPs, while ignoring the patterns of

specific groups of SNPs. This work shows that looking at groups of SNPs helps us better understand what happened long before there was a human historical record.

"When we think of the past, we often think in terms of the historical or geological records," said Mark Johnston, Editor-in-Chief of the journal *GENETICS*. "What makes this development so amazing is that it helps align these records with an emerging biological record based on our DNA. This technique can be applied to any species, making it possible for us to learn and compare the biological histories of all living creatures."

Source: Federation of American Societies for Experimental Biology
([news](#) : [web](#))

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