Geographic Structure in Human Genetic Variation

You in the Context of Human Genetic Variation

At the DNA sequence level, and at many other biological levels, humans are now the most extensively characterized species on the planet. The Human Genome Project set in motion the propulsion of the human species to the forefront of research on population genetics, which is the science of characterizing and understanding genetic variation. Prior to the human genome project, studies of DNA sequence variation in humans focused on the single DNA molecules that comprise the mtDNA (mitochondrial DNA) and the Y chromosome. Availability of the reference sequence of the human genome paved the way for broad assessment of DNA sequence variation throughout the genome. Analyses of your own personal genome data in the context of global patterns of genetic diversity within and among human populations provides a glimpse into your deep ancestry by fitting your genetic composition against geographic patterns established over the evolutionary and migration history of modern humans.

Maternal and Paternal Haplogroups

Modern humans evolved in Africa. Following their origin and relatively long evolutionary history in Africa, humans extended their range outside of the African continent migrating into Asia and Europe and ultimately colonizing the globe. As humans reproduced and migrated creating populations separated by geographic distance - but sometimes reconnecting - the single DNA molecules of the mtDNA and Y chromosome have accumulated mutations. Respectively, mutations in the mtDNA and Y chromosome shared among a subset of living individuals reflect more recent common matrilineal and patrilineal ancestry since the collective ancestry of all modern humans back to "mitochondrial Eve" and "Y-Chromosome Adam". The consequences of these biological processes are that any particular mtDNA or Y chromosome sequence, experimentally defined as a haplogroup by the accumulated mutations present, has a geographic distribution reflective of migration patterns of individuals. Conversely, humans currently living in a geographic region will exhibit a variety of maternal and paternal lineages reflective of the region's colonization history.

Analyses of mtDNA and Y chromosome variation have been conducted for studies of human migration history. In addition to providing a glimpse into our evolutionary past, these studies have produced broad measures of haplogroup diversity across the globe. For example, the image below illustrates percentages of individuals in the H1 and H3 mtDNA haplogroups throughout Europe. If your own mtDNA haplogroup is determined to be H1, your own matrilineal ancestry is likely to trace through a region where this haplogroup is most common. Considering the deeper connections between haplogroups as determined by phylogenetic relationships and current geographic distributions, patterns of human migration are reconstructed. Since mutations accumulate roughly as a function of generations, numbers of accumulated mutations within a lineage also provides an indication of its age.

The Obligatory Ancestry Prediction

Archaic Humanoid Percentages

Links to Additional Resources

Tracing Ancestry with mtDNA - PBS

mtDNA Haplogroups and human migration - Douglas Wallace

Human Family Tree - NYT

Bones, Stones, and Genes: The Origin of Modern Humans - HHMI BioInteractive

Where did my M&Ms come from? - Course Activity (ppt slides)

Reading List

- Stix (2008) Traces of a distant past. *Scientific American* 299:56-63
- Underhill and Kivisild (2007) Use of Y chromosome and mitochondrial DNA population structure in tracing human migrations. *Annual Review of Genetics* 41:539-564
- Emery et al. (2015) Estimates of continental ancestry vary widely among individuals with the same mtDNA haplogroup. *American Journal of Human Genetics* 96:183-193
- The 1000 Genomes Project Consortium (2015) A global reference for human genetic variation. *Nature* 526:68-74