

Genomics study provides insight into the evolution of unique human traits

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Today, researchers from the University of Colorado Health Sciences Center, along with colleagues from Stanford University, report the results of a large-scale, genome-wide study to investigate gene copy number differences among ten primate species, including humans.

The study provides an overview of genes and gene families that have undergone major copy number expansions and contractions in different primate lineages spanning approximately 60 million years of evolutionary time. In the report, which appears online in *Genome Research* (www.genome.org), the scientists speculate how unique, lineage-specific gene copy number expansions and contractions in humans may underlie traits such as endurance running, higher cognitive function, and susceptibility genetic disease.

Primates first appeared on earth approximately 90 million years ago, and today, about 300 different species of primates exist. “One of the main genomic driving forces in primate evolution is gene duplication,” explains Dr. James Sikela, Professor at the University of Colorado. “To our knowledge, this study is the most comprehensive assessment of gene copy number variation across human and non-human primate species so far.”

To survey the differences in gene copy number among these species, Sikela and colleagues used DNA microarrays containing over 24,000 human genes to perform comparative genomic hybridization experiments. They compared DNA samples from humans to those of

nine other primate species: chimpanzee, gorilla, bonobo, orangutan, gibbon, macaque, baboon, marmoset, and lemur. This allowed them to identify specific genes and gene families that, through evolutionary time, have undergone lineage-specific copy number gains and losses.

The authors of the report suggest that “many of the genes identified are likely to be important to lineage-specific traits found in humans and in the other primate lineages surveyed.” To illustrate this potential, the scientists highlighted several gene families that exhibited striking lineage-specific differences. In particular, the human lineage-specific copy number expansion of a gene called *AQP7* could explain why humans have evolved the capacity for endurance running. *AQP7*, or aquaporin 7, plays a role in transporting water and glycerol across membranes. Therefore, it may facilitate the mobilization of glycogen (energy) stores during long periods of intense exercise; it may also play a role in dissipating excess heat through sweating.

The scientists also found dramatic gene copy number differences potentially associated with cognition, reproduction, immune function, and susceptibility to genetic disease.

Source: Cold Spring Harbor Laboratory

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