Great Ape DNA Is Providing New Clues About Human Evolution

Genetic variation is what distinguishes humans from non-humans.

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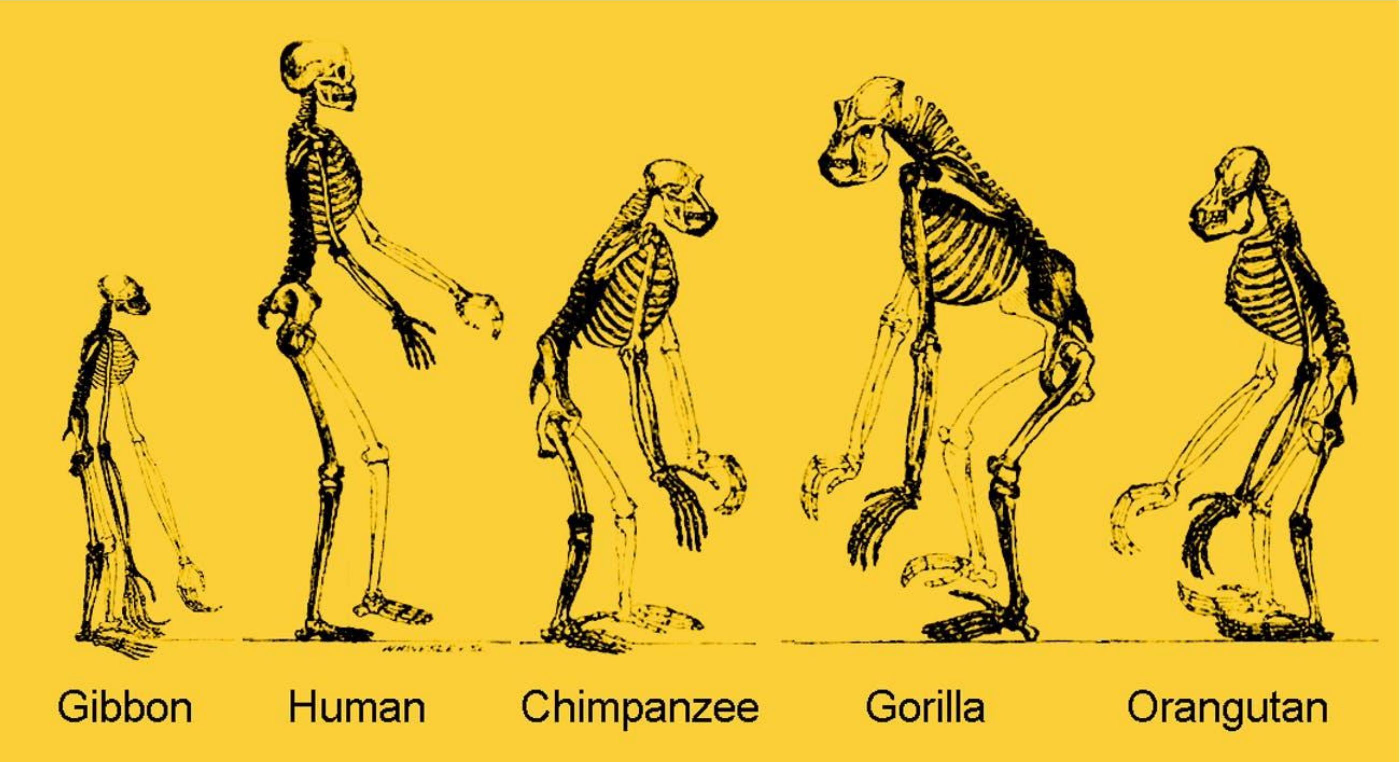
An enduring mission of science is to pinpoint exactly what distinguishes humans from other apes in order to understand our ancestral lineage. This week, we came one step closer to answering that paramount evolutionary question because of a breakthrough published in Science.

Scientists announced Thursday they have successfully sequenced the most complete assemblies of great ape genomes to date, an accomplishment that will drive greater study of the similarities we share with our hominid family.



We're not so different, but scientists have learned more about ape genetics, setting the stage for even more research.

Like chimpanzees, gorillas, and orangutans, humans are a kind of ape. We share with these other apes a common ancestor that lived between 8 and 6 million years ago. Comparing the genetics between the two groups brings us closer to understanding this shared origin.

The scientists sequenced and assembled a chimpanzee and an orangutan genome to a high depth using single-molecule, real-time sequence data. They sequenced more than 500,000 full-length genes from each species, a process that they explain significantly improved the contiguity of chimpanzee and orangutan genomes.

A high-resolution comparative analysis of great ape genomes is providing new information on primate, including human evolution.

These newly assembled genomes, the study authors explain in a statement released Thursday, “present a better resource for novel gene discovery and high-resolution comparative genomics amongst the greatest apes.”

Genetic variation is what distinguishes humans from non-humans — and while humans are different than apes, our evolutionary kinship means that our biological blueprints are still entwined.

Study co-author and professor of genome sciences at the University of Washington, Evan Eichler, Ph.D., tells *Inverse* that having ape genomes that are more complete allows researchers to examine genetic variants that likely affect gene expression — and in turn, parse out that distinguishment.

“We are beginning to get a sense of how some regions of our genome have been radically restructured, specifically in the human lineage,” says Eichler.

Scientists assembled a nearly complete chimp genome.

Eichler and his team compared the new ape genomes with previously assembled human genomes and a gorilla genome to understand how the genome has been restructured.

This process revealed the human genome has 17,000 DNA variants, which represent changes in gene regulation. The majority of these variants were enriched in genes associated with brain morphology: SRGAP2C, ARGAP11B, TBC1D3, and NOTCH2NL.

Eichler cites this as “good evidence” that the genes “specify cortical expansion” and says his team’s next goal is to finish the 1,000 gaps in the great ape genome. He says that “finding genetic hotspots of mutation that affect critical genes during brain development that are shared among all humans but different when compared to all other apes is particularly exciting.”

Previous to this research, scientists were working with ape genomes with a “humanizing” bias by filling in the thousands of gaps in ape genomes with human genome assemblies. This, they explain in the statement, potentially hid whatever structural and expressional differences between human and ape DNA *by* human DNA — in turn, minimizing the chance of understanding the differences between the two.

“Here we filled in almost all of the missing gaps with the correct ape sequence and discovered the full-length transcripts in each ape,” explains Eichler. “Therefore, no need for human fillers.”

The genetic variants that exist between apes and humans highlighted in this study are in line with the argument that ape genomes underwent an expansion of segmental duplication more than 10 million years ago. When the full genome of apes is eventually completed, we might get a look at an even deeper past.