Inside the Most Complete Ape Genomes Ever Assembled

Scientists just unveiled complete telomere-to-telomere genomes for six ape species—revealing what truly makes us human and rewriting the story of evolution

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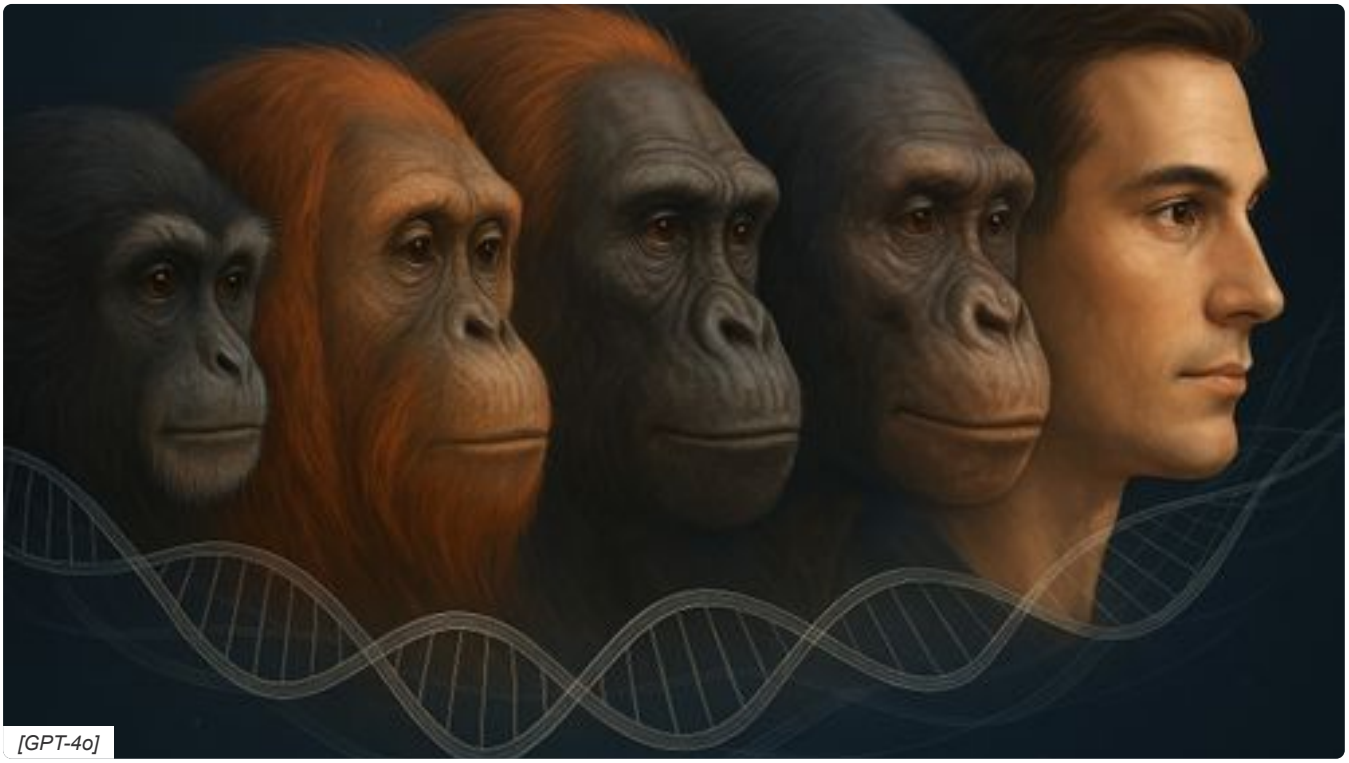

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It took over 120 scientists, more than 40 labs, and years of collaboration, but the result is stunning: near-perfect telomere-to-telomere (T2T) genome assemblies for the siamang, Sumatran orangutan, Bornean orangutan, gorilla, bonobo, and chimpanzee. These long-read, gapless genomes now match the resolution of the most complete human references—and for the first time, we can examine evolutionary changes base pair by base pair. The data for the completed genomes was published recently in ***Nature***.

The international team behind this milestone was led by Evan Eichler of the University of Washington and Howard Hughes Medical Institute; Kateryna Makova of Penn State University; and Adam Phillippy of the NIH's National Human Genome Research Institute. Postdoctoral researcher DongAhn Yoo served as the study's lead author.

"These ape genomes will enable us to reconstruct the evolutionary history of every base pair in our genome," Eichler said.

By resolving regions of structural complexity—including highly repetitive DNA and gene-rich zones like centromeres and segmental duplications—the researchers eliminated the long-standing bias in favor of the human genome. That's a big deal. Until now, comparative studies often assumed human assemblies were more accurate, skewing our understanding of what makes us biologically distinct.

A Genetic Mirror: Pangenomes, Brain Genes, and Evolutionary Detours

A gorilla in Uganda poses for a portrait. [Evan E. Eichler/UW Genome Sciences]

The team didn't stop with individual genome assemblies. They created a 10-way pangenome that integrates all six ape species alongside four diverse human genomes. This comparative framework includes an interspecies graph, enabling scientists to identify lineage-specific innovations, gene duplications, and evolutionary hotspots.

One particularly revelatory area: the brain.

Researchers discovered a Human Ancestor Quickly Evolving Region (HAQER) containing a gene involved in vocal learning—analogous to one found in the song centers of birds. It's expressed in a motor cortex area linked to speech. This region includes a human-specific regulatory sequence that may help explain our unique capacity for language.

In total, they uncovered more than twice as many HAQERs as previously documented. These fast-mutating segments often house species-specific genes, particularly in humans, and are enriched in repetitive DNA—once considered genomic "junk."

The new data also spotlights the major histocompatibility complex (MHC), a hyper-variable immune-related gene cluster. The ape genomes reveal ancient, species-specific differences in this region, which may explain why some diseases affect only humans.

Beyond immune and neurological genes, the researchers found adaptations related to diet, metabolism, and sensory perception—such as the ability to detect bitter compounds and metabolize lipids and iron. These adaptations reflect ecological pressures and divergent lifestyles across ape lineages.

The Power of Segmental Duplications and Chromosomal Architecture

Among the most impactful findings are in segmental duplications—duplicated sequences that are hotspots for genetic innovation. Using long-read sequencing, scientists were able to fully map these regions for the first time, shedding light on their role in genome evolution and disease.

"Segmental duplication rearrangements may be a greater source than previously realized of interspecies differences and potential gene neofunctionalization," the researchers noted.

These duplications often occur on acrocentric chromosomes—those with off-center centromeres, such as chromosomes 13, 14, 15, 21, and 22. Orangutans, for example, have more acrocentric chromosomes and more segmental duplications than any other ape.

Another surprise: bonobos, which split from chimpanzees roughly 1.8 million years ago, have centromeres that are structurally smaller yet still functionally robust. These streamlined designs could inspire synthetic biology applications, such as artificial chromosomes for gene therapy.

With this new genomic clarity, scientists are also able to trace rapidly evolving DNA sequences and explore how segmental duplications differ across species. In humans, some of these duplicated genes are associated with traits like brain expansion. In other apes, they may underlie behaviors and physiological traits we have yet to understand.

What's Next? From Genomic Gaps to Biomedical Gold

Despite the milestone, the work isn't done. About 15 additional ape species and subspecies still lack complete reference genomes. The team also aims to close the final stubborn gaps in the existing

A younger Western Lowland gorilla joins an older gorilla from its tribe in sitting on a large branch. [Leila R. Gray/UW Medicine]

assemblies and to refine gene annotation processes that still prioritize the human genome at the expense of others.

“We are discovering hundreds of protein-coding genes embedded in these segmental duplications that are unique to each ape species,” Eichler said. “Some of these have already been shown to contribute to changes that make us uniquely human, such as a bigger brain.”

These new ape references are more than evolutionary curiosities. They’re launching pads for biomedical discovery, potentially linking the architecture of our genomes to conditions like intellectual disability, developmental delay, and neurodivergent traits.

As these annotated genomes continue to guide research, one thing is clear: the story of human uniqueness is far from over—and our evolutionary cousins are helping us write the next chapter.