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Bonobo Genome Assembly Provides Clues to Primate Evolution

May 05, 2021 | staff reporter

NEW YORK – A team from the US, Italy, and Estonia has tapped into a high-quality bonobo genome assembly for new clues into the evolution of humans and other primates.

The study "highlights the extent to, and rapidity at which, hominid genomes can differ and provides insights into incomplete lineage sorting and its relevance to gene evolution and the genetic relationship among living hominids," co-senior and co-corresponding authors Evan Eichler, a genome scientist at the University of Washington, and Mario Ventura, a biologist at the University of Bari, and their colleagues wrote in a paper published in *Nature* on Wednesday.

For their analyses, the researchers put together a new, high-quality, 3 billion base genome assembly for the bonobo (*Pan paniscus*), or "pygmy chimpanzee." They used DNA from a female bonobo, long-read Pacific Biosciences sequencing data, Illumina short-read data, optical mapping, and other methods. An earlier bonobo genome based on short-read data was described in a 2012 paper in *Nature*.

"High-quality hominid genomes are a critical resource for understanding the genetic differences that make us human as well as the diversification of the *Pan* lineage over the past two million years of evolution," they explained, noting that the bonobo "represents the last of the great ape genomes to be sequenced using long-read sequencing technologies."

The assembly made it possible to get a better look at the great ape's genome architecture, gene content, and structural variant patterns, they explained, including nearly 22,400 predicted protein-coding genes, more than 9,000 non-coding genes, and over 87 Mb of sequence marked by segmental duplications, structural variants patterns, and more.

With insights from the bonobo assembly and an available chimp assembly, meanwhile, the investigators genotyped more than two dozen other great ape genomes.

But the new genome assembly also served as a resource for comparing the bonobo to the closely related and recently diverged chimpanzee (*P. troglodytes*) species and to humans. For example, the team tracked down more than 5,500 fixed structural variant differences between the lineage leading to bonobos and the one leading to chimps, along with nearly 2,000 gene families that appear to have shrunk or ballooned in the bonobo genome relative to the human genome.

Likewise, the investigators identified incomplete lineage sorting (ILS) events that have left parts of the bonobo genome more genetically similar to the human genome than to sequences from the closely related chimp. After mapping those ILS events more fully across all three genomes, they estimated that more than 5 percent of the human genome is closer to either the bonobo or the chimp — up from 3.3 percent or so estimated in the past.

Stretching back still further into our shared evolutionary history, to gorillas and orangutans, the team suggested at least 36.5 percent of the human genome may be marked by ILS. Subsequent analyses suggested that ILS tends to include intergenic parts of the genome rather than protein-coding genes, along with exons from specific types of genes with higher-than-usual levels of amino acid replacement over time.

"Our genome-wide exon analyses specifically show that only a subset of clustered ILS exons are driving this effect," the authors explained, "and that these genes are enriched in glycoprotein and EGF-like calcium signaling functions owing to the action of either relaxed selection or positive selection of genes in these pathways."

In an email, Eichler noted that the clustered distribution of ILS segments "was surprising to us and suggests selection on a subset of this," though he cautioned that "more work needs to be done to determine the functional significance."

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