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International Team Catalogs Ape Genetic Diversity

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By a GenomeWeb staff reporter

NEW YORK (GenomeWeb News) – In a study appearing online today in *Nature*, an international team led by investigators in the US and Spain described the information it gleaned by sequencing the genomes of 79 captive and wild-born apes selected from the main great ape species and sub-species.

Using these sequences, the researchers were able to look at the genetic variation present at the species, subspecies, and population levels. That diversity also helped them to delve into the details of great ape relationships with one another, ape population histories, and inbreeding patterns, in the case of apes born in the wild.

"This comprehensive catalogue of great ape genome diversity provides a framework for understanding evolution and a resource for more effective management of wild and captive great ape populations," the University of Washington's Evan Eichler and co-senior author Tomas Marques-Bonet, with the Scientific Research Council-Pompeu Fabra University Evolutionary Biology Institute in Barcelona, and their colleagues wrote.

For the study, researchers used Illumina's HiSeq 2000 to sequence 79 individuals from six great ape species and seven sub-species, generating 25-fold mean coverage of each individual's genome. In analyzing the genomes, they also added in genome sequence data for nine humans, including three individuals from Africa and six non-Africans.

After multiple analysis and validation steps, the team found some 88.8 million high-quality SNPs in the ape genomes, along with 84 million substitutions that have become fixed in one ape group or another.

Across the ape lineages, the researchers also tracked down nearly 2,000 loss-of-function variants. They noted that there does not seem to have been any obvious shifts in the likelihood of gene loss from the human genome compared to those of apes from other lineages.

"The [more than] 80 million SNPs we identified in this study may now be used to characterize patterns of genetic differentiation among great apes in sanctuaries and zoos," the study's authors concluded, "and, thus, are of great importance for the conservation of these endangered species with regard to their original range."

The variant set not only served as a source of apparent ancestry-informative markers, but it also helped researchers understand genetic diversity in general within each ape population — patterns that provide clues to the complex population histories and population sizes within various branches of the ape tree over time.

In chimpanzees, for instance, the team found that western chimp populations and chimps from Nigeria and Cameroon tended to cluster together genetically, while chimps living in central and eastern parts of Africa formed another group with closer-than-usual genetic ties.

Similarly, within the western lowland gorilla group, researchers noticed genetic distinctions between the gorillas from the Congo relative to those from Cameroon — genetic differentiation that apparently arose after the split between the western and eastern lowland gorilla species.

The genetic data also hint that inbreeding is a common occurrence in wild ape populations across the board. In the eastern gorilla population, though, inbreeding patterns were particularly pronounced.

Eastern lowland gorillas appear prone to low genetic diversity, too, as do bonobos, western chimpanzees, and non-African human populations. At the other extreme, genetic diversity was especially high in populations of western lowland gorillas, chimpanzees from central Africa, and in both Sumatran and Bornean orangutans.

"Humans, western chimpanzees, and eastern gorillas all show a remarkable dearth of genetic diversity when compared to other great apes," the study's authors wrote, noting that "sequencing of 79 great ape genomes identifies more than double the number of SNPs obtained from the recent sequencing of more than a thousand diverse humans — a reflection of the unique out-of-Africa origin and nested phylogeny of our species."

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