CSIR-CCMB

Global primate genome study reveals their evolution and applications for human health

Genomes of 233 primate species reveal key features of primate evolution, human disease, and biodiversity conservation

- **The Institute of Evolutionary Biology, Pompeu Fabra University, Illumina, and Baylor College of Medicine have led a series of global new studies on primate genomes which is published in the journal Science. CSIR-CCMB, Hyderabad is the Indian collaborator and analyzed the Indian primate species.**
- **After generating the most complete catalog of genomic information for primates ever produced, the studies have compared the genomes of 809 individuals from 233 species of primates, in research that involved researchers from 24 countries.**
- **The studies have helped to create a new AI algorithm with clinical applications to identify new insights into the genetic causes of human disease using the primate genomic data.**
- **These studies have discovered DNA sequence variants and developed phylogenies for primate species that will provide new data on primate and human evolution, as well as primate biodiversity.**

Hyderabad, 2nd June, 2023: Two new investigations carried out by global scientists combines genome sequencing of over 800 individuals from 233 primate species, covering nearly half of all existing primate species on Earth, with the study of fossil remains and multiplying by four-fold the number of primate genomes available to date. This also included 83 samples from 19 major primate species in India also studied by scientists at CSIR-Centre for Cellular and Molecular Biology (CCMB). The study provides new information about primates' genetic diversity and evolution, which is important for understanding and conserving the biodiversity of the closest species to our own.

By comparing the genomes of 809 non-human primate individuals from 233 species to the human genome, the research has identified 4.3 million common missense mutations. Such mutations affect the composition of amino acids and can alter the function of proteins, leading to many human diseases.

Only rare missense mutations can raise the risk of disease

One of the current limitations of human and clinical genetics is the inability to detect, among hundreds of thousands of mutations, those that cause diseases. Currently, the genetic causes of many common diseases, such as diabetes and heart diseases, are unknown, both due to the lack of genetic information and the large number of genetic factors involved. Some diseases are thought to originate when a set of mutations with "mild" effects act together to cause a disease of polygenic origin, like diabetes or cancer.

“We have identified 6% of the 4.3 million missense mutations, and these are abundant in primates. We consider these to be ‘potentially benign’ in human diseases, given that their presence is tolerated in these animals,” states Kyle Farh, Vice President of Artificial Intelligence at Illumina.

The identification of disease-causing mutations has been achieved thanks to the PrimateAI-3D deep learning algorithm. PrimateAI-3D is an artificial intelligence (AI) algorithm developed by Illumina, the world’s leading company in DNA sequencing. The algorithm is a kind of ChatGPT for genetics that uses genome sequence instead of human language.

New insights into primate evolution and human uniqueness

The publication of this unique dataset includes the most complete catalog of primate genomic information produced so far, covering nearly half of all existing primate species on Earth. It contains information of primates from Asia, America, Africa, and Madagascar. This catalog has enabled the investigators to compare genomes, to improve the understanding of the evolutionary history of primates and it has provided key insights into what makes us human.

According to Govindhaswamy Umapathy, Senior Principal Scientist at CSIR-CCMB whose group, include Shivakumara Manu and Mihir Trivedi contributed to this research from India, said, "Primates have a great genetic diversity that varies between the different geographical regions and taxonomies. The study of this diversity is crucial for human evolutionary studies, human disease, and for their future conservation". Further, he added that the study also found that the western hoolock gibbon, the only ape from India and lion-tailed macaques from northeastern India and Western Ghats, respectively had low genetic diversity among the global primates examined in this study. He stated that these primates need to be given highest priority both *in situ* and *ex situ* conservation efforts in India.

These studies have also indicated that the genetics of primates does not always correspond with their current classification system. The research found several cases in which relationships among primate species are best described as complex and network-like rather than simple branching trees.

“These studies provide clues as to which species are in most dire need of conservation efforts, and could help to identify the most effective strategies to preserve them,” Vinay Kumar Nandicoori Director, CSIR-CCMB.

Lastly, the new genomic catalog has halved the number of genomic signatures that were believed to be exclusively human. This observation facilitates the identification of those mutations not shared with primates that might be the foundation of the characteristics that make us human.

Reference articles:

Kuderna, L.F.K et al., '***A global catalog of whole-genome diversity from 233 primate species***' Science. May, 2023. DOI: 10.1126/science.abn7829

Gao, H, et al., '***The landscape of tolerated genetic variation in humans and primates***' Science. May 2023. DOI: 10.1126/science.abn8197

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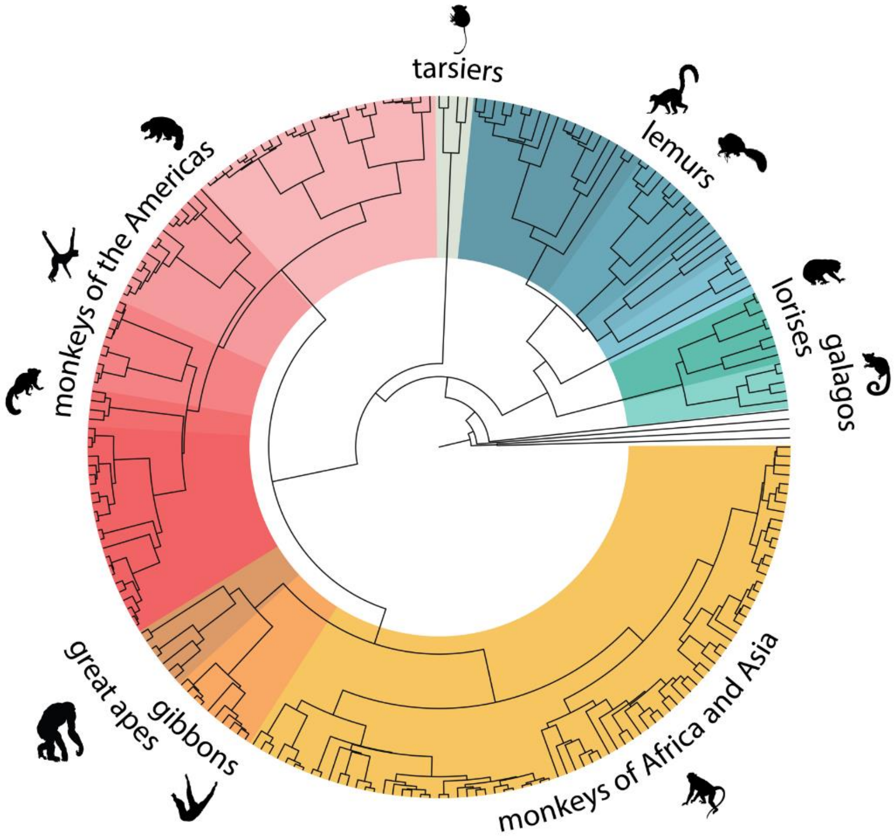
Supplementary images:

Download the images here: https://bit.ly/3N0jbIV

Image 1:



Primates phylogenetic tree. Credit: Lukas Kuderna

Western Hoolock Gibbon (*Hoolock hoolock*)

Female (Biswas) Male (Kaushik)

Western hoolock gibbon, the only ape from Northeast India

Sub-adult male lion-tailed macaque from Western Ghats