Deutsches Primatenzentrum GmbH (DPZ) Leibniz-Institut für Primatenforschung Kellnerweg 4 D-37077 Göttingen http://dpz.eu



Press Release

Genomes of 233 primate species sequenced

New insights into the genetic diversity and evolution of our closest relatives and the genetic causes of human diseases

Göttingen, Germany, June 2, 2023. Researchers from 24 countries have analyzed the genomes of 809 individuals from 233 primate species, generating the most complete catalog of genomic information about our closest relatives to date. The project, which consists of a series of studies in which researchers from the German Primate Center - Leibniz Institute for Primate Research (DPZ) were also involved, provides new insights into the evolution of primates, including humans, and their diversity. In baboons, for example, hybridization and gene flow between different species occurred in the past and is still ongoing in several regions of their range. This makes baboons a good model for the evolution of early human lineages within and outside Africa. In addition, using a specially designed AI algorithm, the genomic data enable new insights into the genetic causes of human diseases (Science, Special Issue).

Primates show great genetic diversity that varies between species and geographic regions. "Studying this diversity is crucial also for understanding human evolution, the causes of human diseases, and for preserving our closest relatives," says Christian Roos, a scientist in the Primate Genetics Laboratory at the German Primate Center and one of the authors. Led by researchers from Universitat Pompeu Fabra, Spain, Baylor College of Medicine, USA, and Illumina Inc, USA, the genomes of 809 individuals from 233 primate species have been sequenced. This covers nearly half of the extant primate species and increases the number of available primate genomes fourfold.

New insights into primate evolution and the uniqueness of humans

The comparative analyses provide fundamental information on the genetic diversity and evolutionary history of primates and important insights into what distinguishes humans from other primates. The genomic data have halved the number of genomic variants thought to occur exclusively in humans. "This makes it easier to look for mutations that we do not share with other primates and that could therefore be the basis for the traits that make us human," says Dietmar Zinner, a scientist in the Cognitive Ethology Laboratory at the German Primate Center and also one of the authors.

One of the studies looks more closely at baboon evolution and finds that there have been several, previously unknown episodes of hybridization and gene flow between baboon species. "We found that baboons from western Tanzania are the first nonhuman primates to have received input from three genetic lineages," said Liye Zhang, a doctoral student at the German Primate Center and one of the lead authors of the baboon study. "These results suggest that the genetic structure of the baboon population and its history of genetic exchange between species is more complex than previously thought and show that baboons make a good model for similar processes in the evolution of early human lineages in and outside Africa," says Dietmar Zinner.

Species conservation with the help of genome data

High genetic diversity enables species to better adapt to changing environmental conditions and pathogens. Especially in small populations, there is a risk of inbreeding and thus a reduction in genetic diversity. Already, 63 percent of all primate species are threatened with extinction, and the analysis of genetic diversity provides information which species most urgently need to be protected, at least from a genetic point of view. "We found particularly low genetic diversity in the golden snubnosed monkey of China and the aye-aye in Madagascar," says Christian Roos.

Rare mutations can increase disease risk

One of the limitations in human and clinical genetics is that it is currently not possible to identify among hundreds of thousands of mutations those that cause disease. To date, the genetic causes of many common diseases, such as diabetes and heart disease, are unknown, due either to a lack of genetic information or to the large number of genetic and other factors involved. By comparing the primate genomes, 4.3 million mutations have now been identified that may alter protein function and thus lead to disease in humans. Six percent of the 4.3 million mutations identified are, however, common in primates and are therefore considered to potentially have little impact on human disease because they are tolerated in these animals. Thanks to the PrimateAI-3D deep-learning algorithm developed by Illumina Inc, disease-causing mutations can now be better identified. "It's a kind of ChatGPT for genetics that uses genome sequences instead of human language," explains Kyle Farh, vice director of the AI group at Illumina Inc, the global leader in DNA sequencing.

Original publications

- Kuderna, LFK, et al.: A global catalog of whole-genome diversity from 233 primate species. Science. May, 2023. https://doi.org/10.1126/science.abn7829
- Sørensen, E, et al.: Genome-wide coancestry reveals details of ancient and recent maledriven reticulation in baboons. Science. May 2023. https://doi.org/10.1126/science.abn8153
- Gao, H, et al.: The landscape of tolerated genetic variation in humans and primates. Science. May 2023. https://doi.org/10.1126/science.abn8197
- Wu, H, et al.: Hybrid origin of a primate, the gray snub-nosed monkey. Science. May 2023. https://doi.org/10.1126/science.abl4997
- Shao, Y, et al.: Phylogenomic analyses provide insights into primate evolution. Science. May 2023. https://doi.org/10.1126/science.abn6919

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You will also find the press release on our <u>website</u>. Please send us a reference copy or link in case of publication.

The German Primate Center GmbH (DPZ) - Leibniz Institute for Primate Research conducts biological and biomedical research on and with primates in infection research, neuroscience and primate biology. The DPZ also maintains four field stations in the tropics and is a reference and service center for all aspects of primate research. The DPZ is one of the 97 research and infrastructure facilities of the Leibniz Association.

Images with captions

Printable images: https://medien.dpz.eu/pinaccess/pinaccess.do?pinCode=Dz1T2V7TWjni



Prof. Dr. Christian Roos, scientist at the Primate Genetics Laboratory of the German Primate Center – Leibniz Institute for Primate Research. Photo: Karin Tilch



Dr. Dietmar Zinner, scientist at the Cognitive Ethology Laboratory of the German Primate Center – Leibniz Institute for Primate Research. Photo: Karin Tilch



Liye Zhang, PhD student at the Primate Genetics Laboratory of the German Primate Center – Leibniz Institute for Primate Research. Photo: Karin Tilch



Baboon in western Tanzania, here at Mahale Mountains National Park, have received genetic input from three lineages. Photo: Yvonne de Jong and Tom Butynski



Juvenile golden snubnosed monkey at Shennongjia National Park, Central China. Photo: Julia Ostner



Yellow baboon (*Papio cynocephalus*) at Ruaha National Park, Tanzania. Photo: Yvonne de Jong and Tom Butynski



Olive baboon (Papio anubis) at Lake Manyara National Park, Tanzania. Photo: Dietmar Zinner



Hamadryas baboon (Papio hamadryas) in the Eritrean highlands. Photo: Dietmar Zinner



Chacma baboon (Papio ursinus) at De Hoop Nature Reserve, South Africa. Photo: Dietmar Zinner



Guinea baboon (*Papio papio*) in the Niokolo Koba National Park, Senegal. Ludwig Ehrenreich