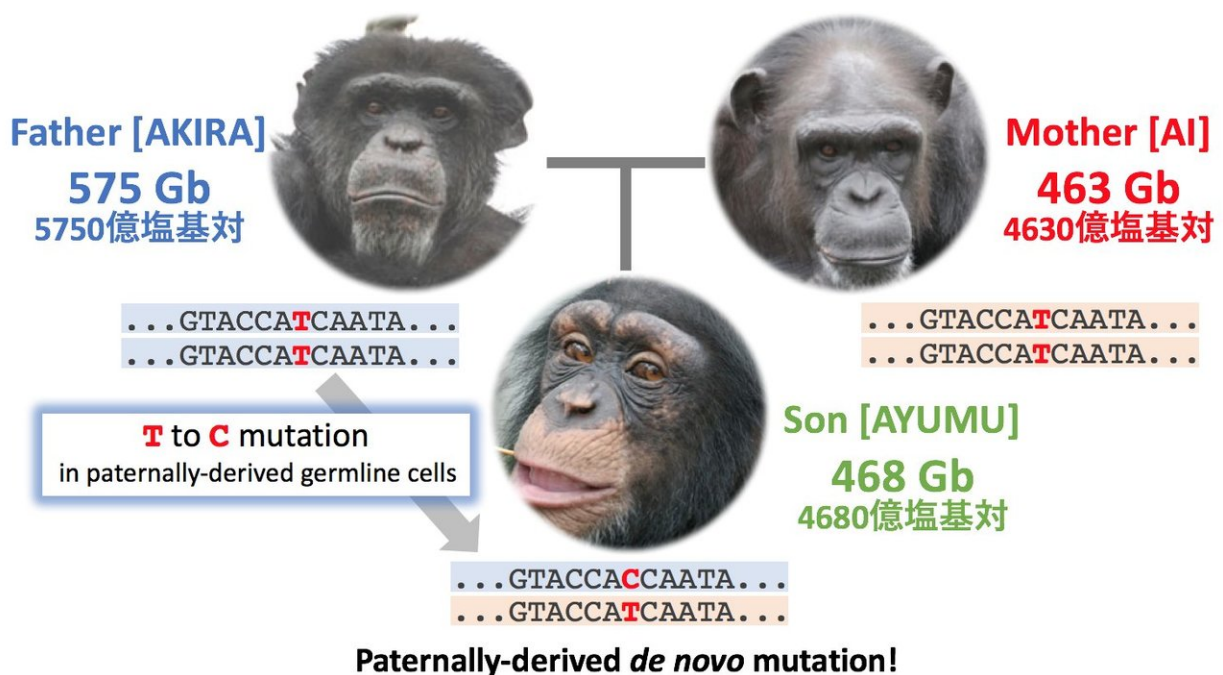


How far did you fall from the tree? Scientists estimate the mutation rate from chimpanzee parents to their offspring

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Deep sequencing reveals mutation rates: Genomic study of a chimpanzee parent-child trio has shed light on mutation rates from one generation to the next.
Credit: Kyoto University

Mutations generate genetic variation, and are a major driving force of evolution. Therefore, examining mutation rates and modes are essential to better understand the genetic basis for physiology and evolution.

One traditional method to estimate this rate is a 'phylogenetic' approach that examines the sequence divergence between two species, and the time between divergence events. However, this method entails many uncertainties, such as the extent of ancestral polymorphisms, effective population size, and generation time.

To overcome these difficulties and estimate [mutation rates](#) directly, a team of researchers including Tetsuro Matsuzawa of the Kyoto University Institute for Advanced Study (KUIAS) and Yasuhiro Go of Japan's National Institutes of Natural Sciences (NINS) conducted a study utilizing high-quality whole genome sequencing of a chimpanzee parent-offspring trio, and successfully disclosed the precise mutation rates and modes through the transmission of genomes from parents to child.

It is well known that chimpanzees are humans' closest genetic relatives. The trio of Akira, Ai, and Ayumu (father, mother, son) in this study have taken part in a wide variety of comparative cognitive research—notably in "The Ai-Project" led by Matsuzawa—dating back to 1978.

Due to the abundance of this previous cognitive research, the authors anticipate that these chimpanzees can facilitate an understanding of how genetic architecture influences phenotypic—cognitive—traits, shedding light on the development of individuality.

In this study, 574, 463, and 468 gigabases (Gb) of DNA were analyzed from the father, mother, and son. As the chimpanzee genome is roughly three-gigabases in length, the volume of this sequence data was unprecedented, enabling the scientists to decipher precise mutation trajectories occurring within one generation.

When analyzed, the rate of germline, newly-formed mutations was calculated to be 1.48 new mutations for every 100 million bases, higher

than that for humans, which has been reported as 0.96 to 1.2 new mutations.

A strong, male-biased mutation spectrum was also measured, wherein three-quarters of [mutations](#) occurred in sperm thorough spermatogenesis, as well as new structural alterations, such as changes in gene conversion and copy number. This is particularly important as the precise rates and modes of new structural alterations are still poorly understood, even in humans.

To this end, the team considers their experimental and methodological framework useful for further inquiry into the complex nature of genomic changes.

The paper "Direct estimation of de novo mutation rates in a chimpanzee parent-offspring trio by ultra-deep whole genome sequencing" appeared 1 November 2017 in *Scientific Reports*.

More information: Shoji Tatsumoto et al. Direct estimation of de novo mutation rates in a chimpanzee parent-offspring trio by ultra-deep whole genome sequencing, *Scientific Reports* (2017). [DOI: 10.1038/s41598-017-13919-7](#)

Provided by Kyoto University

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