

Gorilla genome sequenced

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Male silverback Gorilla in SF zoo. Image: Wikipedia.

The assembly of the gorilla genome was announced today, March 7, by a multi-national group of researchers. The gorilla is the last genus of the living great apes to have its genome decoded. While confirming that our closest relative is the chimpanzee, the team showed that much of the human genome more closely resembles the gorilla than it does the chimpanzee genome.

This is the first time scientists have been able to compare the genomes of all four living great apes: humans, chimpanzees, gorillas and orangutans. This study provides a new perspective on human origins and is an important resource for research into human evolution and biology, as well as for gorilla biology and conservation.



Researchers at the Wellcome Trust Sanger Institute in the United Kingdom lead the study, with contributions from several other institutions, including the University of Washington.

"The gorilla genome is important because it sheds light on the time when our ancestors diverged from our closest evolutionary cousins. It also lets us explore the similarities and differences between our genes and those of the gorilla, the largest living primate," said Aylwyn Scally, first author from the Wellcome Trust Sanger Institute. "Using DNA from Kamilah, a female western lowland gorilla, we assembled a gorilla genome sequence and compared it with the genomes of the other great apes. We also sampled DNA sequences from other gorillas in order to explore genetic differences between gorilla species."

The team searched more than 11,000 genes in human, chimpanzee and gorilla for genetic changes important in evolution. Humans and chimpanzees are genetically closest to each other over most of the genome, but the team found many places where this is not the case. Some 15 percent of the human genome is closer to the gorilla genome than it is to chimpanzee, and 15 percent of the chimpanzee genome is closer to the gorilla than human.

"This finding suggests a quite rapid process of speciation and explains why it is so difficult to differentiate the three species in a very important period of our evolution," said Dr. Tomas Marques, who worked on this project when he was a postdoctoral student at the University of Washington and Howard Hughes Medical Institute genome science lab of Dr. Evan Eichler. "We noticed that the reconstruction of human evolution is more complex than we had anticipated."

The researchers also called attention to fact that ear shape is one of the few external physical traits in which humans look more like gorilla than chimps.



In all three species, genes relating to sensory perception, hearing and brain development showed accelerated evolution, particularly so in humans and gorillas.

"Our most significant findings reveal not only differences between the species reflecting millions of years of evolutionary divergence, but also similarities in parallel changes over time since their common ancestor," said Dr. Chris Tyler-Smith, senior author from the Wellcome Trust Sanger Institute. "We found that gorillas share many parallel genetic changes with humans including the evolution of our hearing. Scientists had suggested that the rapid evolution of human hearing genes was linked to the evolution of language. Our results cast doubt on this, as hearing genes have evolved in gorillas at a similar rate to those in humans."

"The study of certain categories of genes, such as those linked to hearing perception, shows that they seemed to evolved more rapidly in humans and gorillas in parallel," said Marques. The finding is new and unusual, he said, because some of these genes had been previously linked to aspects of behavior thought to be intrinsically human.

The research team's observations were consistent with studies showing a major role for adaptive modifications in sensory perception and brain development in hominines (great apes and human-like creatures), such as a genetic basis for blood flow modifications allowing for larger brains.

In gorilla evolution, one of the most significantly accelerated genes, the researchers, said may be related to the leathery texture of gorilla knuckle pads. Gorillas often drag their knuckles along the ground when they walk.

Other genes comparatively examined were those possibly related to growth hormones, sperm function, and defense against viruses.



The researchers also mentioned that, in several cases, a genetic variation thought to cause disease in humans was associated with a normal state in gorillas. Striking examples, they said, were variants linked to dementia and dangerously enlarged hearts in humans.

This paper also illuminates the timing of splits between species. Although we commonly think of species diverging at a single point in time, this does not always reflect reality: species can separate over a long period of time.

The team found that divergence of gorillas from humans and chimpanzees occurred around ten million years ago. The split between eastern and western gorillas was much more recent, in the last million years or so, and was gradual, although they are now genetically distinct. This split is comparable in some ways to the split between chimpanzees and bonobos, or modern humans and Neanderthals.

The researchers also noticed greater genetic diversity in the western versus the eastern lowland genetic samples. This finding is consistent with the smaller population of eastern lowland gorillas, which is merely one-tenth of the 200,000 estimated living western lowland gorillas.

"Because it manifests in genetic diversity, this disparity must have existed for many millennia, and cannot have resulted solely from the current pressure of human activity in central Africa or recent outbreaks of the Ebola virus," the authors surmised.

In the Middle Miocene epoch many kinds of great apes lived throughout Europe, Asia and Africa.

Since then, the authors note, the story of great ape evolution has told of fragmentation and extinction. People are the only remnant of the genus Homo. All of the other human-like primates vanished.



Gorillas survive today in just a few isolated and endangered populations in the equatorial forests of central Africa. They are severely threatened and their numbers are diminishing. This research not only provides new information about human evolution, but also highlights the importance of protecting and conserving the full diversity of these remarkable species, the researchers said.

"As well as teaching us about human evolution," the authors wrote, "the study of great apes connects us to a time when our existence was more tenuous, and in so doing, teaches us the importance of protecting and conserving these remarkable le species."

"Our research completes the genetic picture for overall comparisons of the great apes," said Dr. Richard Durbin, senior author from the Wellcome Trust Sanger Institute, "After decades of debate, our genetic interpretations are now consistent with the fossil record and provide a way for paleontologists and geneticists to work within the same framework.

"Our data are the last genetic piece we can gather for this puzzle. There are no other living great ape genera to study."

The UW's role in this project was to use previous knowledge of the great ape genomes to assess the quality of the gorilla genome assembly, especially in the more complex regions that are difficult to annotate. The UW researchers also applied their experience in studying the composition of structural variations in the gorilla genome to other great apes and humans, and to comparing the rates of variations among gorillas and humans.

More information: Scally et al. 'Insights into hominid evolution from the gorilla genome sequence' *Nature*, March 8 <u>DOI:10.1038/nature10842</u>



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