A burst of genetic changes occurred around 10 million years ago in one of the ancestors we share with chimpanzees, with consequences that are being felt today.

A new analysis of the genomes of macaques, orang-utans, chimpanzees and humans shows that DNA segments in this ancestor began to make duplicate copies at a very high rate around ten million years ago, even though other mutation processes such as single letter changes were slowing.

“There’s a big burst of activity that happens where genomes are suddenly rearranged and changed,” says Evan Eichler, of the Howard Hughes Medical Institute.

He adds, “Because of the architecture of the human genome, genetic material is constantly being added and deleted in certain regions. These are really like volcanoes in the genome, blowing out pieces of DNA.”

In *Nature* the team reports that this duplication spree occurred before humans and chimpanzees diverged, around 6 million years ago, but after the divergence from orang-utans 12-16 million years ago and the divergence from macaques prior to that. (See also this week’s news feature on human evolution: *The other strand.*

“In light of the importance of segmental duplications in contributing to copy-number changes associated with neurocognitive disease and disease susceptibility, we predict that this apparent acceleration has had a profound impact on the reproductive success, adaptability and evolution of ancestral hominid populations,” they write.

But although this rash of duplications may be associated with autism and other medical conditions they also appear to be under positive selection. “I believe that the negative selection of these duplications is being outweighed by the selective advantage of having these newly minted genes, but that’s still unproven,” says Eichler.

In another press release, paper authors Tomas Marques-Bonet and Jeffrey Kidd highlight just how much this paper poses as many questions as it answers:

“It is unclear why, but the common ancestor of humans, chimps and gorillas had an unusual activity of duplication,” Kidd added. “Moreover, we don’t yet know the functions of most of the genes that were affected by these duplications.”

... “There is still no final answer as to why chimps and humans are different,” Marques-Bonet and Kidd said. “Maybe segmental duplications that are specific to humans are another layer to explore, or maybe the distinction between human and chimps is not found in these genetic differences.

“What is certain is that genetic differences contribute significantly to what makes a human and chimp different, and we know that these regions of our genetic code are changing much more rapidly than most others. The next challenge will be making sense of all these differences and the genes that are affected by them.”
Gene copies played key role in human-ape split: study - AFP
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