Did increased gene duplication set the stage for human evolution?

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These are two western lowland gorillas at Woodland Park Zoo, Seattle. This species is from western equatorial African. The gorilla on the left shows the knuckles-on-the-ground stance characteristic of great apes. Credit: Allison C. Gray

(PhysOrg.com) -- Roughly 10 million years ago, a major genetic change occurred in a common ancestor of gorillas, chimpanzees, and humans. Segments of DNA in its genome began to form duplicate copies at a greater rate than in the past, creating an instability that persists in the genome of modern humans and contributes to diseases like autism and schizophrenia. But that gene duplication also may be responsible for a genetic flexibility that has resulted in some uniquely human characteristics.

"Because of the architecture of the human genome, genetic material is constantly being added and deleted in certain regions," says Howard Hughes Medical Institute investigator and University of Washington geneticist Evan Eichler, who led the project that uncovered the new findings. "These are really like volcanoes in the genome, blowing out pieces of DNA." The research was published in the February 12, 2009, issue of *Nature*.

Eichler and his colleagues focused on the genomes of four different species: macaques, orangutans, chimpanzees, and humans. All are descended from a single ancestral species that lived about 25 million years ago. The line leading to macaques broke off first, so that macaques are the most distantly related to humans in evolutionary terms. Orangutans, chimpanzees, and humans share a common ancestor that lived 12-16 million years ago. Chimps and humans are descended from a common ancestral species that lived about 6 million years ago.

By comparing the DNA sequences of the four species, Eichler and his colleagues identified gene duplications in the lineages leading to these species since they shared a common ancestor. They also were able to estimate when a duplication occurred from the number of species sharing that duplication. For example, a duplication observed in orangutan, chimpanzees, and humans but not in macaques must have occurred sometime after 25 million years ago but before the orangutan lineage branched off.

Eichler's research team found an especially high rate of duplications in the ancestral species leading to chimps and humans, even though other mutational processes, such as changes in single DNA letters, were slowing down during this period. "There's a big burst of activity that happens where genomes are suddenly rearranged and changed," he says. Surprisingly, the rate of duplications slowed down again after the lineages leading to humans and to chimpanzees diverged. "You might like to think that humans are special because we have more duplications than did earlier species," he says, "but that's not the case."

These duplications have created regions of our genomes that are especially prone to large-scale reorganizations. "That architecture predisposes to recurrent deletions and duplications that are associated with autism and schizophrenia and with a whole host of other diseases," says Eichler.

Yet these regions also exhibit signs of being under positive selection, meaning that some of the rearrangements must have conferred advantages on the individuals who inherited them. Eichler thinks that uncharacterized genes or regulatory signals in the duplicated regions must have created some sort of reproductive edge. "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," he said.

An important task for future studies is to identify the genes in these regions and analyze their functions, according to Eichler. "Geneticists have to figure out the genes in these regions and how variation leads to different aspects of the human condition such as disease. Then, they can pass that information on to neuroscientists and physiologist and biochemists who can work out what these proteins are and what they do," he says. "There is the possibility that these genes might be important for language or for aspects of cognition, though much more work has to be done before we'll be able to say that for sure."

More information: *Nature*, February 12, "A burst of segmental duplications in the genome of the African great ape ancestor"

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