Whitehead Institute

CHIMP AND HUMAN Y CHROMOSOMES EVOLVING FASTER THAN EXPECTED

X and Y chromosomes from a human male karyotype.

Image: National Human Genome Research Institute (NHGRI)

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TAGS: PAGE LAB EVOLUTION + DEVELOPMENT

CAMBRIDGE, Mass. – Contrary to a widely held scientific theory that the mammalian Y chromosome is slowly decaying or stagnating, new evidence suggests that in fact the Y is actually evolving quite rapidly through continuous, wholesale renovation.

By conducting the first comprehensive interspecies comparison of Y chromosomes, Whitehead Institute researchers have found considerable differences in the genetic sequences of the human and chimpanzee Ys—an indication that these chromosomes have evolved more quickly than the rest of their respective genomes over the 6 million years since they emerged from a common ancestor. The findings are published online this week in the journal *Nature*.

“The region of the Y that is evolving the fastest is the part that plays a role in sperm production,” say Jennifer Hughes, first author on the *Nature* paper and a postdoctoral researcher in Whitehead Institute Director David Page’s lab. “The rest of the Y is evolving more like the rest of the genome, only a little bit faster.”

The chimp Y chromosome is only the second Y chromosome to be comprehensively sequenced. The original chimp genome sequencing completed in 2005 largely excluded the Y chromosome because its hundreds of repetitive sections typically confound standard sequencing techniques. Working closely with the Genome Center at Washington University, the Page lab managed to painstakingly sequence the chimp Y chromosome, allowing for comparison with the human Y, which the Page lab and the Genome Center at Washington University had sequenced successfully back in 2003.

The results overturned the expectation that the chimp and human Y chromosomes would be highly similar. Instead, they differ remarkably in their structure and gene content. The chimp Y, for example, has lost one third to one half of the human Y chromosome genes--a significant change in a relatively short period of time. Page points out that this is not all about gene decay or loss. He likens the Y chromosome changes to a home undergoing continual renovation.

“People are living in the house, but there’s always some room that’s being demolished and reconstructed,” says Page, who is also a Howard Hughes Medical Institute investigator. “And this is not the norm for the genome as a whole.”

Wes Warren, Assistant Director of the Washington University Genome Center, agrees. “This work clearly shows that the Y is pretty ingenious at using different tools than the rest of the genome to maintain diversity of genes,” he says. “These findings demonstrate that our knowledge of the Y chromosome is still advancing.”

Hughes and Page theorize that the divergent evolution of the chimp and human Y chromosomes may be due to several factors, including traits specific to Y chromosomes and differences in mating behaviors.

Because multiple male chimpanzees may mate with a single female in rapid succession, the males’ sperm wind up in heated reproductive competition. If a given male produces more sperm, that male would theoretically be more likely to impregnate the female, thereby passing on his superior sperm production genes, some of which may be residing on the Y chromosome, to the next generation.

Because selective pressure to pass on advantageous sperm production genes is so high, those genes may also drag along detrimental genetic traits to the next generation. Such transmission is allowed to occur because, unlike other chromosomes, the Y has no partner with which to swap genes during cell division. Swapping genes between chromosomal partners can eventually associate positive gene versions with each other and eliminate detrimental gene versions. Without this ability, the Y

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In chimps, this potent combination of intense selective pressure on sperm production genes and the inability to swap genes may have fueled the Y chromosome's rapid evolution. Disadvantages from a less-than-ideal gene version or even the deletion of a section of the chromosome may have been outweighed by the advantage of improved sperm production, resulting in a Y chromosome with far fewer genes than its human counterpart.

To determine whether this rapid rate of evolution affects Y chromosomes beyond those of chimps and humans, the Page lab and the Washington University Genome Center are now sequencing and examining the Y chromosomes of several other mammals.

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Written by Nicole Giese Rura

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David Page's primary affiliation is with Whitehead Institute for Biomedical Research, where his laboratory is located and all his research is conducted. He is also a Howard Hughes Medical Institute investigator and a professor of biology at Massachusetts Institute of Technology.

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"Chimpanzee and human Y chromosomes are remarkably divergent in structure and gene content"

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