

The fickle Y chromosome

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The male sex chromosome, long dismissed as the underachieving runt of the genome, has now been fully sequenced in a common chimpanzee. And comparison with its human counterpart — the only other Y chromosome to have been sequenced in such detail — reveals a rate of change that puts the rest of the genome to shame.

The common chimp (*Pan troglodytes*) and human Y chromosomes are “horrendously different from each other”, says David Page of the Whitehead Institute for Biomedical Research in Cambridge, Massachusetts, who led the work. “It looks like there’s been a dramatic renovation or reinvention of the Y chromosome in the chimpanzee and human lineages.”

Sex chromosomes evolved some 200 million–300 million years ago, but the chimpanzee and human lineages diverged only 6 million–7 million years ago. Comparisons of the chimp and human genomes suggested that not much has changed between the species since¹.

But those analyses excluded the Y chromosome, much of the genetic sequence of which is made up of palindromes and elaborate mirrored sets of bases that elude standard whole-genome sequencing techniques. Portions of the chimp Y chromosome were sequenced a few years ago^{2,3}, but the full landscape is only now available, after Page and his team precisely sequenced large segments of the chromosome, then stitched them together. They report their findings in a paper published online in *Nature* on 13 January⁴.

As the earlier studies had suggested, many of the stark changes between the chimp and human Y chromosomes are due to gene loss in the chimp and gene gain in the human. Page’s team found that the chimp Y chromosome has only two-thirds as many distinct genes or gene families as the human Y chromosome and only 47% as many protein-coding elements as humans. The remainder of the chimp and human genomes are thought to differ in gene number by less than 1%.

Even more striking than the gene loss is the rearrangement of large portions of the chromosome. More than 30% of the chimp Y chromosome lacks an alignable counterpart on the human Y chromosome, and vice versa, whereas this is true for less than 2% of the remainder of the genome.

Even the portions that do line up have



His sex chromosome is an evolutionary hotspot.

undergone erratic relocation. In the only other chromosome to have been sequenced to the same degree of completeness in both species, chromosome 21, the authors found much less rearrangement.

“If you’re marching along the human chromosome 21, you might as well be marching along the chimp chromosome 21. It’s like an unbroken piece of glass,” says Page. “But the relationship between the human and chimp Y chromosomes has been blown to pieces.”

The rapid evolution of the Y chromosome is not a total surprise, because the Y chromosome has no partner during cell division and so largely avoids the exchange of DNA that occurs between partnered chromosomes and keeps modifications in check. “It’s expected that they are going to be more different than the rest of the genome, but the extent of it is pretty amazing,” says geneticist Christine Disteche at the University of Washington in Seattle.

The Y chromosome is also prone to change because most of its characterized genes are involved in producing sperm, which are at the frontline of reproductive fitness, particularly in chimps; receptive females will often mate with many males in one session, so the male with the most virile sperm has the highest likelihood of success.

“The Y is full of surprises,” Page says. “When we sequenced the chimp genome people thought we’d understand why we have language and write poetry. But one of the most dramatic differences turns out to be sperm production.” ■

Lizzie Buchen

1. The Chimpanzee Sequencing and Analysis Consortium. *Nature* **437**, 69–87 (2005).
2. Hughes, J. F. *et al.* *Nature* **437**, 100–103 (2005).
3. Kuroki, Y. *et al.* *Nature Genet.* **38**, 158–167 (2006).
4. Hughes, J. F. *et al.* *Nature* doi:10.1038/nature08700 (2010).