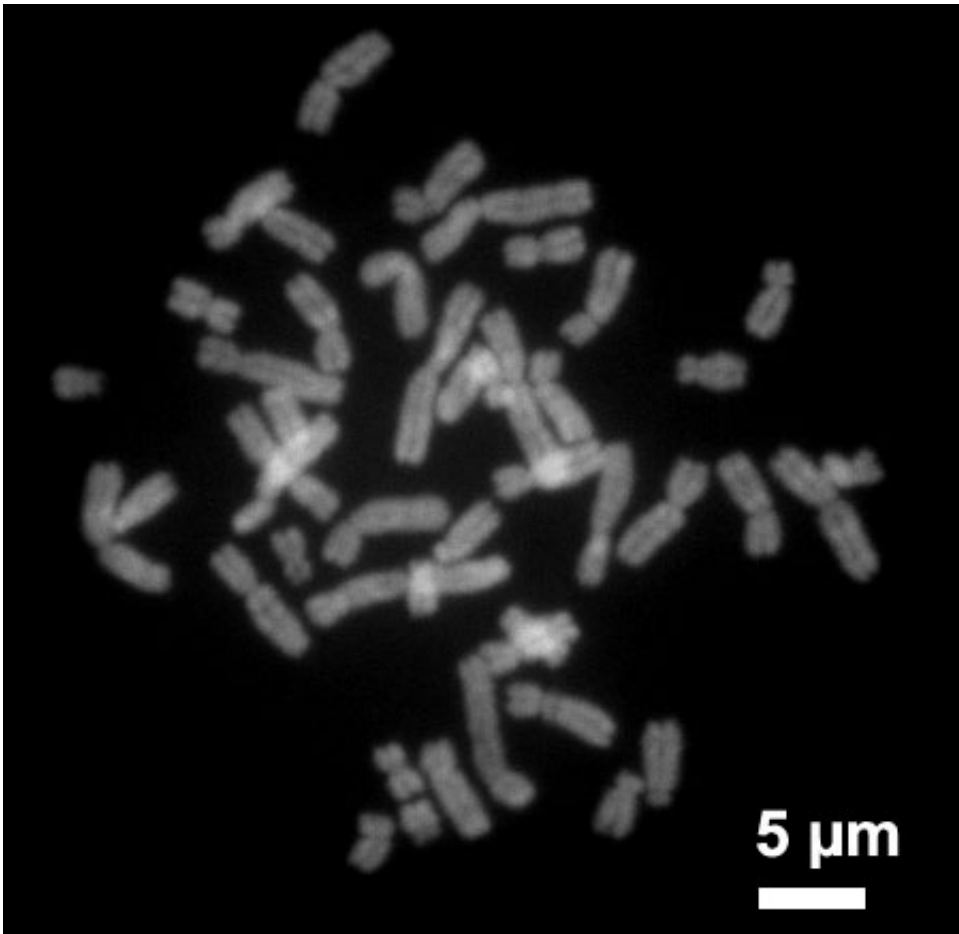


# Study dispels theories of Y chromosome's demise

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Human chromosomes during metaphase. Credit: Steffen Dietzel/Wikipedia

A comparison of Y chromosomes in eight African and eight European men dispels the common notion that the Y's genes are mostly

unimportant and that the chromosome is destined to dwindle and disappear.

"The Y chromosome has lost 90 percent of the genes it once shared with the X chromosome, and some scientists have speculated that the Y chromosome will disappear in less than 5 million years," said evolutionary biologist Melissa A. Wilson Sayres, a Miller Postdoctoral Fellow in the Department of Integrative Biology at the University of California, Berkeley, and lead author of the new analysis.

Some mammals have already lost their Y chromosome, though they still have males and females and reproduce normally. And last month, researchers reported shuffling some genes in mice to create Y-less males that could produce normal offspring, leading some commentators to wonder whether the chromosome is superfluous.

"Our study demonstrates that the genes that have been maintained, and those that migrated from the X to the Y, are important, and the human Y is going to stick around for a long while," she said.

Wilson Sayres and coauthor Rasmus Nielsen, UC Berkeley professor of [integrative biology](#), show in a paper published online today (Jan. 9, 2014) in *PLOS Genetics* that patterns of variation on the Y chromosome among the 16 men are consistent with [natural selection](#) acting to maintain the gene content there, much of which has been shown to play a role in male fertility. The Y chromosome's puny size – it contains 27 unique genes versus thousands on the other [chromosomes](#) – is a sign it is lean and stripped down to essentials.

"Melissa's results are quite stunning. They show that because there is so much natural selection working on the Y chromosome, there has to be a lot more function on the chromosome than people previously thought," Nielsen said.

Variations in Y chromosomes are used to track how human populations moved around the globe, and according to Nielsen, the new research will help improve estimates of humans' evolutionary history.

"Melissa has shown that this strong negative selection – natural selection to remove deleterious genes – tends to make us think the dates are older than they actually are, which gives quite different estimates of our ancestors' history," Nielsen said.

## **Y has degraded over past 200 million**

Before about 200 million years ago, when mammals were relatively new on Earth, early versions of the [sex chromosomes](#), X and Y, were just like other pairs of chromosomes: with each generation, they swapped a few genes so that offspring were a mix of their parents' genes. Fertilized eggs that got two proto-Xs became females and eggs with a proto-X and proto-Y became males.

But for some reason, Wilson Sayres said, the gene that triggers the cascade of events that result in male features became fixed on the Y chromosome and attracted other male-specific genes, such as those that control development of the testes, sperm and semen. Many of these turned out to be harmful for females, so the X and Y stopped swapping genes and the two chromosomes began to evolve separately.

"Now the X and Y do not swap DNA over most of their lengths, which means that the Y cannot efficiently fix mistakes, so it has degraded over time," she said. "In XX females, the X still has a partner to swap with and fix mistakes, which is why we think the X hasn't also degraded."

Wilson Sayres was fascinated by the strange history of the sex chromosomes and in particular the lack of genetic variation worldwide on the Y chromosome compared to the variety seen in DNA on the non-

sex chromosomes. This variation, though used to chart human history, was poorly characterized across the entire Y chromosome.

"Y chromosomes are more similar to each other than we expect," said Wilson Sayres. "There has been some debate about whether this is because there are fewer males contributing to the next generation, or whether natural selection is acting to remove variation."

## **Did fewer males contribute genes to Y chromosome?**

The UC Berkeley researchers demonstrated that if fewer males were the only cause of the low variability, it would mean that fewer than 1 in 4 males throughout history had passed on their Y chromosome each generation. Variations in other [human chromosomes](#), including the X chromosome, make this an unlikely scenario. Instead, they showed that the low variation can be explained by intense natural selection, that is, a strong evolutionary pressure to weed out bad mutations that ended up trimming the chromosome down to its essentials.

"We show that a model of purifying selection acting on the Y chromosome to remove harmful mutations, in combination with a moderate reduction in the number of males that are passing on their Y chromosomes, can explain low Y diversity," Wilson Sayres said.

The researchers also found that all 27 genes on the Y chromosome – the 17 that humans retain after 200 million years, and 10 more recently acquired but poorly understood genes – are likely affected by natural selection. Most of the newer genes, called ampliconic [genes](#), are present in multiple copies on the chromosome and loss of one or more copies has been linked to male infertility.

"These ampliconic regions that we haven't really understood until now are evidently very important and probably should be investigated and

studied for fertility," she said.

Wilson Sayres was able to precisely measure Y variability because for the first time she compared variation on a person's Y chromosome with variation on that person's other 22 chromosomes (called autosomes), the X chromosome and the mitochondrial DNA. She used whole genome data from 16 men whose DNA had been sequenced by the Mountain View-based company Complete Genomics Inc., which has the most accurate sequences of the Y chromosome. The company was recently acquired by BGI, the Beijing Genome Institute.

Cross-population studies of variation in the Y chromosome are in their infancy, she said, noting that of the more than 36 mammalian genomes sequenced to date, complete Y chromosomes are only available for three. Most of the 1,000+ human genomes already sequenced do not have sufficiently accurate coverage of the Y to make this type of comparison among individuals, but advances in technology to better characterize DNA will facilitate future analyses of the Y chromosome, she said.

Provided by University of California - Berkeley

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