Evolutionary biologists glimpse early stages of Y-chromosome degeneration

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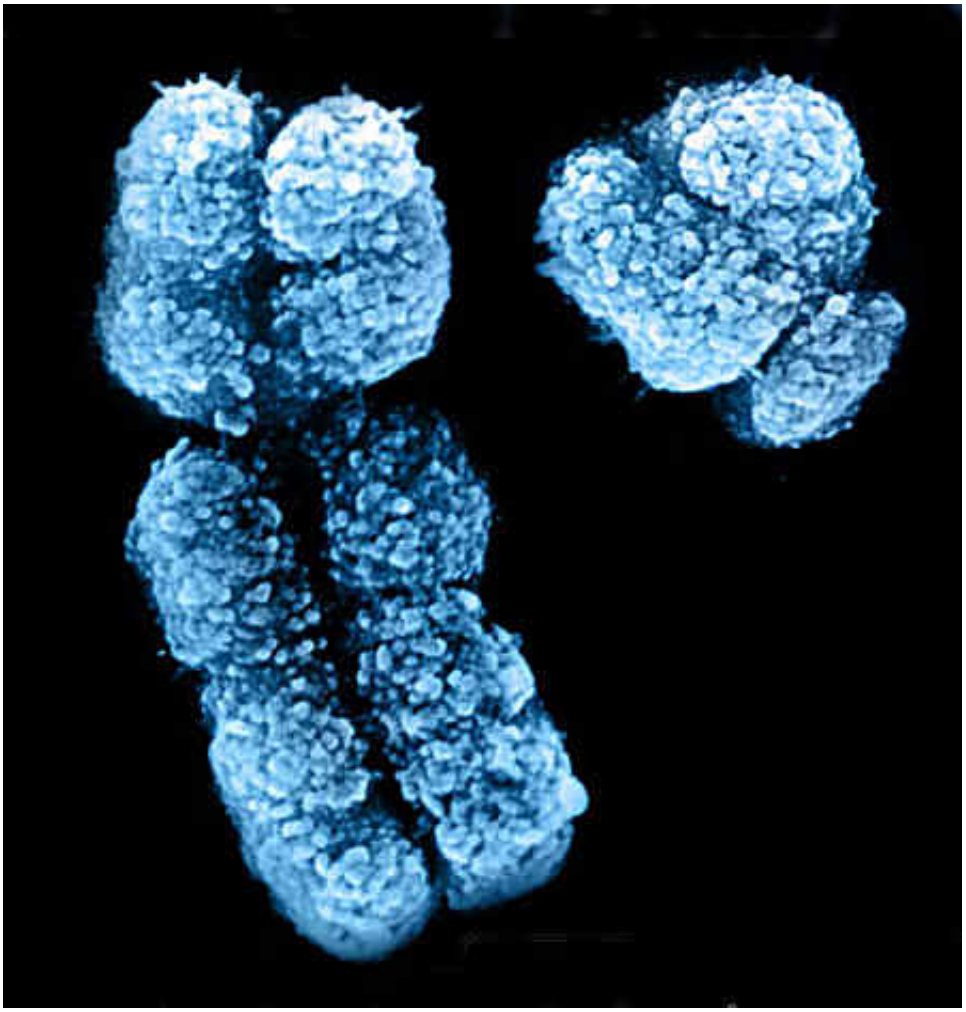

In many species, the possession of X and Y chromosomes determines

whether an individual develops into a male or female. In humans, for example, individuals who inherit their father's Y chromosome become male (XY), and individuals who inherit their father's X chromosome become female (XX).

This system of sex determination has evolved independently multiple times and a striking feature of its evolution is that Y chromosomes have degenerated genetically, losing many genes over time. What is not well understood, however, is what happens to the Y chromosome during the earliest stages of this evolution, or the time scales over which degeneration occurs.

Now, University of Toronto (U of T) researchers have found a way to shed light on the early stages of degeneration, by investigating the process in plants.

"In humans, the Y chromosome has undergone extensive gene loss over its roughly200-million-year evolutionary history, and now retains only about three per cent of its ancestral genes. We know very little about the early stages of the process, however, because it happened so long ago," said U of T Department of Ecology & Evolutionary Biology (EEB) professor Spencer Barrett, co-investigator of a study published today in *Proceedings of the National Academy of Sciences*. "The most well-studied Y chromosomes, including those in humans and other animal species, began degenerating hundreds of millions of years ago. Not so with plants."

"The emergence of separate sexes in plants is a relatively recent evolutionary innovation, making them ideal for this study," said Barret. "Only about six per cent of flowering plants have males and females. The remainder are hermaphrodites."

The scientists used a plant species with separate sexes whose X and Y

chromosomes probably first evolved around 15 million years ago at the most, making them relatively young compared to those in animals.

"We tested for Y-chromosome degeneration in Rumex hastatulus, an annual plant from the southern USA commonly known as heartwing sorrel. We found that genes on the Y chromosomes have already started to undergo genetic degeneration, despite their relatively recent origin," said Josh Hough, a PhD candidate in U of T's Department of Ecology & Evolutionary Biology and lead author of the study. "Importantly, our results indicate that the extent of this degeneration depends on how long ago the genes on the sex chromosomes stopped recombining with each other."

The theory of sex chromosome evolution holds that Y-chromosome degeneration occurs as a result of X and Y chromosomes failing to recombine their genes during reproduction. Recombination is a key genetic process in which chromosomes pair and exchange their DNA sequences, and it occurs between all other chromosomes in the genome, including the X chromosome, which recombines in females. This genetic mixing has become suppressed between the X and Y chromosomes, however, probably because they contain genes that affect 'femaleness' and 'maleness', and combining these genes onto a single chromosome can cause infertility problems.

"Suppressing recombination between the X and Y makes sense because it prevents genes that determine female-specific traits from occurring on the Y chromosome," said Hough. But without recombination natural selection becomes less efficient, and harmful mutations cannot be removed from the Y chromosome. As a result, genes on the Y chromosome eventually become impaired in function or lost entirely."

The researchers crossed multiple male and female plants and then traced the inheritance of genes by sequencing the DNA in parents and their

offspring. This allowed them to find which genes were located on the sex chromosomes because they segregate differently than genes on other chromosomes. Computer-assisted analyses of the genetic sequences enabled the scientists to then test for gene loss, loss of gene function, the accumulation of mutations, and other harmful changes on the sex chromosomes.

Suppressed recombination between X and Y chromosomes occurred much more recently in plants than in animals, so the scientists were able to get a unique glimpse of what happens during the very earliest stages of Y-chromosome degeneration.

"In addition to being much younger than in animals, the sex chromosomes in Rumex hastatulus are particularly interesting because of the recent emergence of a new sex chromosome system, in which some males carry a second, even younger, Y chromosome," says Hough. "This allowed us to compare the two Y chromosomes and assess the time scales over which genes are deteriorating."

"The genes on the second Y chromosome are very new arrivals, having arisen within a single species", says EEB professor Stephen Wright, another investigator on the study. "This gave us a key time point to understand the chronology of Y-chromosome evolution. Remarkably, even these genes were already showing early signs of degeneration."

More information: Genetic degeneration of old and young Y chromosomes in the flowering plant Rumex hastatulus, *PNAS*, www.pnas.org/cgi/doi/10.1073/pnas.1319227111

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