

Male sex chromosome losing genes by rapid evolution, study reveals

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A wallaby is a marsupial. Credit: Kateryna Makova, Penn State

Scientists have long suspected that the sex chromosome that only males carry is deteriorating and could disappear entirely within a few million years, but until now, no one has understood the evolutionary processes that control this chromosome's demise.

Now, a pair of Penn State scientists has discovered that this sex chromosome, the Y chromosome, has evolved at a much more rapid pace than its partner chromosome, the X chromosome, which both males

and females carry. This rapid [evolution](#) of the Y chromosome has led to a dramatic loss of genes on the Y chromosome at a rate that, if maintained, eventually could lead to the Y chromosome's complete disappearance. The research team, which includes Associate Professor of Biology Kateryna Makova, the team's leader, and National Science Foundation Graduate Research Fellow Melissa Wilson, will publish its results in the 17 July 2009 issue of the journal [PLoS Genetics](#).



A giraffe is a eutherian mammal. Credit: Kateryna Makova, Penn State

"There are three classes of mammals," said Makova, "egg-laying mammals, like the platypus and the echidna; marsupials, like the opossum and the wallaby; and all other mammals -- called eutherians -- which include humans, dogs, mice, and giraffes. The X and Y chromosomes of marsupials and eutherians evolved from a pair of non-sex chromosomes to become [sex chromosomes](#)."

Humans have 23 pairs of chromosomes, which are the structures that hold our DNA, but just one pair of these chromosomes are sex chromosomes, while the others are referred to as non-sex chromosomes. "In eutherian mammals, the sex chromosomes contain an additional region of DNA whereas, in the egg-laying mammals and marsupials, this

additional region of DNA is located on the non-sex chromosomes," said Makova. "At first, bits of DNA within this additional region were readily swapped between the X and Y chromosomes, but some time between 80 and 130 million years ago, the region became two completely separate entities that no longer swapped DNA. One of the regions became specifically associated with the X chromosome and the other became specifically associated with the Y chromosome."

By comparing the DNA of the X and Y chromosomes in eutherian mammals to the DNA of the non-sex chromosomes in the opossum and platypus, the team was able to go back in time to the point when the X and Y chromosomes were still swapping DNA, just like the non-sex chromosomes in the opossum and platypus. The scientists then were able to observe how the DNA of the X and Y chromosomes changed over time relative to the DNA of the non-sex chromosomes. "Our research revealed that the Y-specific DNA began to evolve rapidly at the time that the DNA region split into two entities, while the X-specific DNA maintained the same evolutionary rate as the non-sex chromosomes," said Makova.



An echidna is a monotreme. Credit: Kateryna Makova, Penn State

Once the biologists determined that the Y chromosome has been evolving more rapidly and has been losing more genes as a result, they wanted to find out why the Y chromosome has not already disappeared entirely. "Today, the human Y chromosome contains less than 200 genes, while the human X chromosome contains around 1,100 genes," said Wilson. "We know that a few of the genes on the Y chromosome are important, such as the ones involved in the formation of sperm, but we also know that most of the genes were not important for survival because they were lost, which led to the very different numbers of genes we observe between the once-identical X and Y. Although there is evidence that the Y chromosome is still degrading, some of the surviving genes on the Y chromosome may be essential, which can be inferred because these genes have been maintained for so long."

The team then decided to test the hypothesis that some of the genes on the Y chromosome are being maintained because they are essential. The team's approach was to compare the expression and function of genes on the Y chromosome with analogous genes on the X chromosome. "If the genes' expressions and/or functions were different, then it would make sense that the genes on the Y chromosome would be maintained because they are doing something that the genes on the [X chromosome](#) can't do," said Makova. "This hypothesis turned out to be correct."

Although some of the genes on the Y chromosome have been maintained, most of them have died, and the team found evidence that some others are on track to disappear, as well. "Even though some of the genes appear to be important, we still think there is a chance that the Y chromosome eventually could disappear," said Makova. "If this happens, it won't be the end of males. Instead, a new pair of non-sex chromosomes likely will start on the path to becoming sex [chromosomes](#)."

In the future, the team plans to use its newly generated data to create a

computer model that tracks the degeneration of the Y chromosome. The scientists hope to determine how long it will take for the Y chromosome to disappear. They also hope to identify the processes that are most important for degeneration of the [Y chromosome](#).

Source: Pennsylvania State University ([news](#) : [web](#))

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