

New genes on "deteriorating" Y chromosome

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The Y-chromosome of fruit flies consists not just of "gene-junk", but also functioning genes. Credit: Markus Riedl

The Y chromosome, which is found only in males, is difficult to decode even with the latest sequencing technologies. Among evolutionary biologists, the question as to which genes lie on the male sex chromosome and where they came from is therefore hotly debated. Using an innovative analysis method, a team of population geneticists from Vetmeduni Vienna have now made a crucial breakthrough. They

were able to show that genetic material in fruit flies is often transferred to the Y chromosome from the other chromosomes. Although this transfer largely occurs as a result of "accidents," the researchers now demonstrate that some of these transfers create functional genes on the Y chromosome. The findings, published in *PNAS*, will supply new momentum for the research of the male chromosome in other species.

Y [chromosomes](#), which are only inherited paternally, evolved from "normal" chromosomes known as autosomes. As males only possess one Y chromosome, there is no counterpart for recombination, the direct exchange of genetic material. This makes the deletion of harmful mutations on the Y chromosome more difficult than in other chromosomes. As a result, genes on the Y chromosome usually undergo a process of degeneration. Earlier studies with fruit flies have shown that new genes can be transferred onto the Y chromosome, although the rate was estimated as very low (1 transfer in 10 million years).

Researchers from the Institute of Population Genetics at Vetmeduni Vienna, using a new and highly specific analysis method, could now provide fresh momentum to help decode the evolutionary dynamics of the Y chromosome. Their study shows that ten times more new genes are transferred onto the Y chromosome in fruit flies than had been previously thought. Some of these new genes even appear to have taken on important functions.

The Y chromosome has been a tough nut to crack in genome research. As it possesses only few functional genes, and these are embedded in repetitive DNA that is difficult to analyse, finding these genes is a challenge. "Only seven functional genes have been identified on the Y chromosome of *Drosophila melanogaster*. But we suspect that the number of functional genes as well as the actual transfer rate must be higher," says first author Ray Tobler. "We therefore developed a new analysis method that allows us to efficiently search for gene transfers

onto the Y chromosome, so-called GeTYs."

The researchers' trick consisted in sequencing the genome of males and females from a so-called inbred strain of [fruit flies](#). These differ only in the Y chromosome sequence. "The key to our results was to search for variants in the males that do not exist among the females," says Tobler. "That means we worked without any known Y chromosome sequences that would usually be used for a comparison. This allowed us to trace the transferred genes back to so-called retrocopies, which are created when the RNA transcript of a gene is inserted into the Y chromosome."

All previously described gene transfers onto the Y chromosome involved the transfer of a piece of the chromosome and not an RNA transcript. "The high number of validated [gene transfers](#) allowed us to statistically show that there were differences between *Drosophila* species," explains senior author Christian Schlötterer. "We only found genes originating from an RNA transcript in the closely related *D. mauritiana* and *D. simulans*, which suggests that the transfer mechanisms are species-specific."

A special surprise for the research team was that four of the 25 newly transferred genes on the Y chromosome have already assumed an important function there. "As these new genes can be found in all individuals of a species, the question arises as to which functions these new Y-linked genes could have," says Tobler. Until now, it has been still completely unclear if and how long these new genes can withstand the deterioration of the Y chromosome. As the new analysis method does not require a reference genome for the Y chromosome, it offers enormous potential to study the dynamics of new [genes](#) on the Y chromosome in many different species. "I expect many more exciting findings," concludes Christian Schlötterer.

More information: Ray Tobler et al. High rate of translocation-based

gene birth on the *Drosophila* Y chromosome, *Proceedings of the National Academy of Sciences* (2017). DOI: [10.1073/pnas.1706502114](https://doi.org/10.1073/pnas.1706502114)

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