

Sex and the single chromosome

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Two different fruit fly species can be as distantly related as humans and rats. That makes these insects good model systems for examining sex chromosomes in the process of evolution. Image credits: Doris Bachtrog

Is there value to sex? For higher organisms, absolutely. Animals, plants and fungi that reproduce only by cloning are scarce as hen's teeth, suggesting the gene shuffling of sex pays handsome dividends.

The most obvious consequences of sex can be seen in the so-called sex chromosomes. These mismatched sets of chromosomes determine the sex of individual organisms. Case in point: the human X and Y. “Several hundred million years ago they were actually from an identical pair. Now our X has over a thousand genes while the Y has only a few handfals,” says Doris Bachtrog, a Berkeley professor of integrative biology.

Bachtrog studies how sex chromosomes evolve these differences. Her approach is to compare related species whose sex chromosomes emerged at different times. “We can see the chronological order of how they differentiate,” Bachtrog says. “We want to see what types of changes occur, and how quickly this happens.” In doing so, she hopes to

understand the evolutionary significance of sex itself.

One universal effect of sex is the degeneration of the unmatched chromosome. Bachtrog's comparisons of various fruit fly species demonstrate that Y chromosomes accumulate specific defects over time. These include repeated stretches of DNA, as well as epigenetic modifications (changes that don't alter the gene sequence) that affect how frequently genes are transcribed. Many of these so-called epigenetic modifications silence otherwise functional genes.

Isolation is the likely cause of the Y's decline. "The Y is completely sheltered from recombination; it has no partner with which it can exchange genes," Bachtrog says. In other words, any mutations a Y sustains are guaranteed to be passed to the next generation. Bachtrog is using her observations of progressive changes in the Y to develop and refine mathematical models of how degeneration proceeds.

Until recently, scientists believed that the X suffered no such gene loss indignities, and that recombination allowed it to remain similar to its autosomal ancestor. Skeptical, Bachtrog set out to test this assumption. She measured the rate of background adaptive mutations occurring within the genomes of fruit fly species, and compared those to the rate of adaptive mutations occurring on their X chromosomes. She found that young X chromosomes evolve about ten times faster than the rest of the genome.

"If you think about it, it makes sense. The X chromosome is faced with a lot of novel changes. It used to be an autosome, and all of a sudden, it's an X that spends more of its time in females," Bachtrog says. "Some signatures we're looking at suggest the X is adapting so quickly because it's now completely changing its genome content; it's become a good place to put female-specific genes."

If the X has become a repository for genes that might benefit females, Bachtrog reasoned, the Y is likely a haven for male-beneficial genes. To explore this idea, Bachtrog analyzed where genes expressed more frequently in males were more likely to be located. She found that such male-linked genes tend to be absent on the X.

Another curious consequence of sex is a phenomenon called dosage compensation. For example, because female humans have two X chromosomes, they shut down one X to maintain gene expression parity with males. [Fruit flies](#), however, take a different tack, with males hypertranscribing the genes on their one Xs. Bachtrog can see dosage compensation in the process of emerging among fruit fly species with younger [sex chromosomes](#). She is now investigating how male X chromosomes attract the changes promoting hypertranscription.

In recent years, Bachtrog has expanded her research to include [animals](#) with more exotic chromosomal arrangements. These include species where females possess the unique [sex](#) chromosome. Such ZW species may possess an additional quirk: a lack of dosage compensation. Bachtrog is studying emus and chickens, vipers and insects called tephritids to determine whether dosage compensation is truly absent in ZW species, and what evolutionary forces shape these odd sexual systems.

Such comparative species studies are only now becoming possible thanks to recent advances in sequencing technologies. While Bachtrog spent years piecing together the genome of a single [species](#) of fruit fly, her students can do the same within months. Says Bachtrog, “Sometimes the amount of data is overwhelming, but it’s great to have.”

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