Highlight: The Degenerating Y Chromosome: Under the Pressures of Sex and Selection

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Sexual reproduction is a big hit among living things. It spins out offspring with large genetic variability, allowing nature a wide scope for selection. The process is metabolically costly and reduces the overall number of progeny. Nonetheless, the vast majority of multicellular life forms—animals, plants, and fungi—and even many single-celled creatures would not do it any other way.

When a sexual reproductive system evolves, two homologous chromosomes, containing the sex determining genes, lose the ability to recombine, according to the current theory. This ensures that the female-determining genes remain separate from those that determine maleness. These protosex chromosomes start evolving independently. In the case of an XY sex determination system, as found in humans and Drosophila, the Y chromosome begins to degenerate. The majority of its genes will, in time, stop working.

The process of gene loss is, however, not random. The more important and more highly expressed genes are retained longer than their counterparts, biologists from the University of California at Berkeley report in Genome Biology and Evolution. Though the neo-Y is degenerating at a breakneck pace on an evolutionary timescale, selection pressures are still felt (Genome Biology and Evolution 10.1093/gbe/evr103).

“Gene loss is happening so fast, I wouldn’t have thought there was any scope for selection to weed out genes that are more or less costly to lose,” said Vera Kaiser, lead author and postdoctoral scientist. “I wasn’t expecting to find a pattern, to be honest.”

Kaiser et al. chose Drosophila miranda to study because its Y-chromosome, or one part of it, is still quite young in evolutionary terms. “The miranda system is wonderful for looking at because it is at a midpoint [in Y chromosome evolution],” said Brian Charlesworth, an evolutionary biologist at the University of Edinburgh. “It’s only about halfway down the toilet.”

About 1.5 Ma, one of the autosomes fused to the existing Y chromosome. Today’s Y chromosome is in two parts: the old Y and the neo-Y. (The X chromosome, likewise, has a new and an old part.) On the neo-Y, about 40% of the genes have degenerated, with more being lost all the time.

Y chromosomes are inherited by sons directly from their fathers, and there is no chance of recombination during meiosis. Without the opportunity to swap genetic material with a homologous chromosome, mutations that knock out Y-linked genes tend to spread through a population. In time, the Y chromosome becomes shrunken, gene poor, and bears little, if any, resemblance to its ancestral X partner.

Mutations and loss of function in the Y-linked genes are rarely fatal because the organism still has a working copy in the X chromosome. Functional loss does, however, reduce the dosage of those gene products.

“The effect on fitness [from losing a gene on the Y] is quite small normally,” Kaiser said, “estimated at about 1–2 percent for genes that are lethal when both copies are lost. But we saw that even such a small reduction in fitness had an effect on selection.”

Kaiser wanted to know if the gene loss was utterly random or if D. miranda was holding on to genes that are more costly to lose because of selective pressures. Previous studies have shown that highly expressed genes, a measure of relative importance, are more likely to be retained in other organisms (such as Paramecium), whereas low-expression genes are more likely to be lost (e.g., in yeast) (Gout et al. 2010).

Kaiser and her team annotated the genomic sequence of D. miranda’s neo-Y chromosome, listing the genes as either functional or not. These they compared with the neo-Y’s homologue in a sister species, Drosophila pseudoobscura. The genetic material newly sex-linked in D. miranda is autosomal in D. pseudoobscura.

Examining the mRNA levels of those autosomal genes in D. pseudoobscura, used as a proxy for the neo-Y’s ancestral state, Kaiser found, to her surprise, a strong correlation between those genes that were highly expressed and those genes that were still functional. Ultimately, the neo-Y chromosome in D. miranda is fated to lose most of its current genes. In time, it will become like the human or Drosophila melanogaster Y chromosome: holding only a few male-specific or highly expressed genes.
“This is an almost unique system to study, fascinating because we can capture the changing Y chromosome in near real-time,” Charlesworth said. “It gives us a natural experiment in what happens when we turn off the process of sexual reproduction.”

Without the chance of recombination, the Y chromosome is inherited as in an asexual system. Observing the neo-Y’s degeneration, then, throws light on why sexual recombination is so valuable. Chromosomes have more freedom to retain ‘good’ or useful mutations, and slough off damaging mutations.

“Without genetic recombination acting in a system, a ‘good’ mutation at one place becomes permanently stuck with a ‘bad’ mutation at another place,” Charlesworth said. “Genetic exchange allows good mutations to separate from bad, and spread unimpeded throughout the population. That is a key advantage.”

**Literature Cited**

Gout JF, Kahn D, Duret L. 2010. The relationship among gene expression, the evolution of gene dosage, and the rate of protein evolution. PLoS Genet. 6:e1000944.