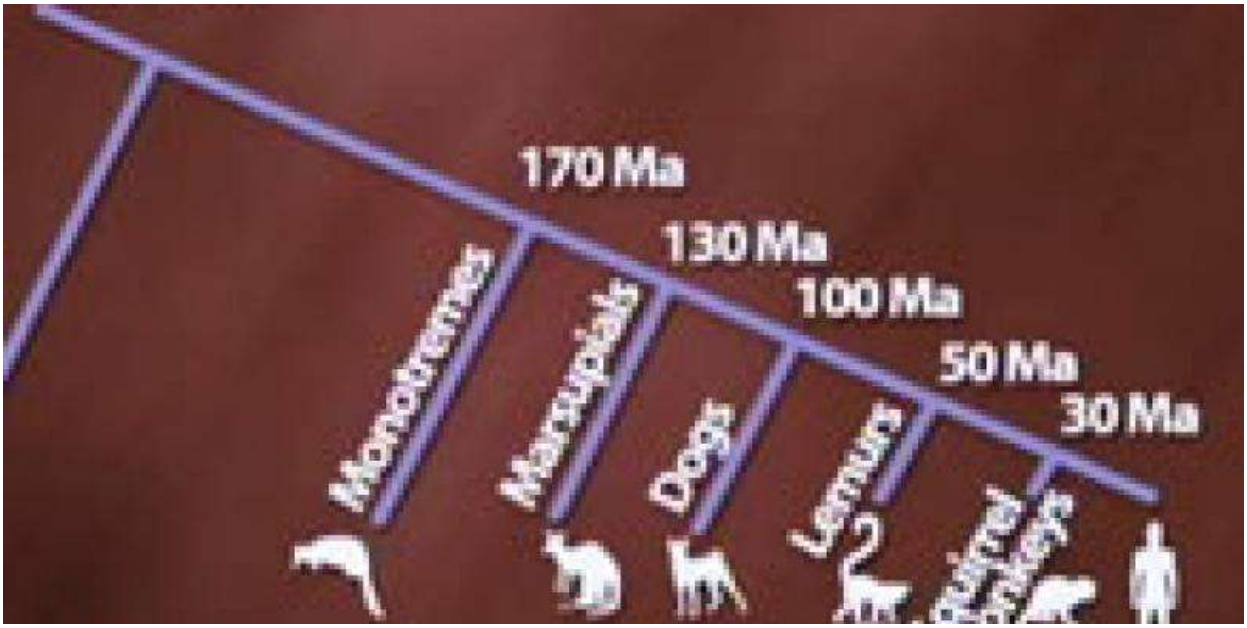
Evolution of the Y Chromosome



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The human X and Y chromosomes are a unique pair. The other chromosome pairs, called the autosomes <http://www.hhmi.org/biointeractive/gender/y_evolution.html#autosome>, appear to be identical twins; they are superficially indistinguishable. In contrast, the X and Y chromosomes appear to be vastly different from one another. Why are the sex chromosomes so different? How did they get that way?

The Y chromosome is only one-third the size of the X. Although the Y has a partner in X, only the tips of these chromosomes are able to recombine. Thus, most of the Y chromosome is inherited from father to son in a pattern resembling asexual, not sexual, reproduction. No recombination <http://www.hhmi.org/biointeractive/gender/y_evolution.html#recombination> means no reassortment, so deleterious mutations <http://www.hhmi.org/biointeractive/gender/y_evolution.html#mutation> have no opportunity to be independently selected against. The Y chromosome therefore tends to accumulate changes and deletions <http://www.hhmi.org/biointeractive/gender/y_evolution.html#deletions> faster than the X. Degradation doesn't occur in X chromosomes because during female meiosis <http://www.hhmi.org/biointeractive/gender/y_evolution.html#meiosis>, the X has the other X as a full partner in recombination.

Clues of how the Y chromosome evolved can be found by comparing the genes and the sequences of X and Y chromosomes as well as homologous genes of different species. One method scientists use to estimate evolutionary time is observing how homologous genes have become different over time in different species. All DNA sequences accumulate random mutations over time, so species that are distant relatives should have more different sequences than close relatives because they have been evolving separately for a longer time. Once recombination stopped between portions of X and Y, genes located on those parts started to evolve separately as homologs. Apparently, this happened in stages, so some X-Y gene pairs are more related than others, meaning they stopped recombining more recently. Also, chunks of genes stopped recombining, and by mapping their positions on the chromosome, one can guess that an event, like an inversion

How did the human Y chromosome become so small relative to its X counterpart? This animation depicts the 300-million-year odyssey of the sex chromosomes that began when the proto X and Y were an identical pair. Over time, structural changes in the Y chromosome resulted in its current form, which is specialized to trigger male development. The evolutionary timescale is represented by positioning the chromosomal remodeling events along an abbreviated vertebrate cladogram, a chart of evolutionary relationships.

(Ma = million years ago)

Part 1. Sex chromosomes originated as autosomes

The sex chromosomes began as an ordinary pair of autosomes <http://www.hhmi.org/biointeractive/gender/y_evolution.html#autosome>. During meiosis <http://www.hhmi.org/biointeractive/gender/y_evolution.html#meiosis>. chromosomes replicate their DNA, pair, and exchange genes (recombination <http://www.hhmi.org/biointeractive/gender/y_evolution.html#recombination>; red lines). A mutation <http://www.hhmi.org/biointeractive/gender/y_evolution.html#mutation> in the *SOX3* gene produced the *SRY* <http://www.hhmi.org/biointeractive/gender/y_evolution.html#sry> gene, a critical determinant of maleness, on the proto Y. While the functions of *SRY* and *SOX3* became very different over time, another gene, *RPS4* <http://www.hhmi.org/biointeractive/gender/y_evolution.html#rps4>, retained a similar function on both the X and Y chromosomes.

Part 2. Inversions restrict recombination between the X and the Y chromosome

Inversions <http://www.hhmi.org/biointeractive/gender/y_evolution.html#inversions>, which are internal recombination events, caused a rearrangement of genes on the Y chromosome. These rearrangements meant that large portions of the X and Y chromosome no longer recombined, which made the Y chromosome susceptible to deletions <http://www.hhmi.org/biointeractive/gender/y_evolution.html#deletions>, and it

After our lineage diverged from the ancestors of the monotremes, such as the duck-billed platypus, another inversion further scrambled the genes on the proto Y. In males, only the tips of the Y chromosome were left able to recombine with homologous genes on the X chromosome. In contrast, in females, recombination <http://www.hhmi.org/biointeractive/gender/y_evolution.html#recombination> continued to occur across the full length of the two identical X chromosomes.

Part 4. Autosomal expansion of X and Y chromosomes

About 130 million years ago (Ma), an autosome donated a block of genes that extended the length of both the X and the Y chromosome. The X and Y were able to recombine in these expanded regions of the chromosomes. Subsequently, inversions rearranged the order of genes on the Y chromosome. Additional rearrangements occured almost exclusively on the Y. Without recombination to preserve its integrity, the Y continued to lose genes and, over time, shrank.

Part 5. An autosome contributed a copy of the *DAZ* spermatogenesis gene to the Y chromosome

Sometime after squirrel monkeys diverged from the primates that evolved into humans, an autosome contributed a copy of the *DAZ* spermatogenesis gene to the Y chromosome. The *DAZ* <http://www.hhmi.org/biointeractive/gender/y_evolution.html#daz> gene was copied and copied again and now the modern Y chromosome contains four identical *DAZ* gene sequences. The modern Y chromosome is about one-third the size of its X-chromosome partner.

Learn More: Autosomes

An autosome is any chromosome that is not a sex chromosome. In ancient reptilian creatures, there was no chromosomal basis for sex determination. Scientists speculate that sex was determined by environmental factors such as temperature.

Many genes essential for the production of sperm are located exclusively on the Y chromosome. One of these genes, *DAZ* (*d*eleted in *az*oospermia), was copied from an autosome and was copied twice on the Y chromosome, resulting in four copies of the *DAZ* gene. The Y chromosome is unique because not only are male spermatogenesis genes sequestered on the Y, but they exist as mutiple copies. In fact, the abundance of multiple copies and mirror images of sequences have led researchers to call the Y chromosome a "hall of mirrors." Although this sequence repetition created great challenges in the sequencing of the Y chromosome, the complex structure also serves an important purpose. Multiple copies of essential spermatogenesis genes ensure that in spite of deletion events, which may result in the loss of a single copy of an essential gene, spermatogenesis can still proceed via proteins produced by remaining copies.

Learn More: Deletions

Deletions are uncommon, but relative to inversions, they are not rare events. (Recombination, however, is a common event). Deletions occur particularly in regions of the Y chromosome that do not undergo recombination. The chromosome is mutated, causing a section of DNA to be excised, and the two flanking ends of DNA join to form a continuous strand.

Learn More: Expansion

About 130 Ma, an autosome donated a block of genes that extended the length of both proto X and Y (expansion). The proto X and Y were able to recombine in these expanded regions of the chromosomes. Subsequently, inversions further rearranged the order of genes. Without recombination that preserved the integrity of chromosomes, the proto Y lost genes and, over time, shrank in size.

Learn More: Inversions

On an evolutionary timescale, large inversions, such as those shown in the animation,

large inversions resulted in the relocation of *SRY* to the "top" of the Y chromosome relative to its former partner *SOX3*, which remains near the "bottom" of the X chromosome.

Learn More: Meiosis

To produce sperm or eggs, germ cells undergo the process called meiosis. Chromosomes replicate and pair up, resulting in a 4n quantity of chromosomes. Two nuclear divisions follow, so that the gametes have a haploid (1n) number of chromosomes.

Learn More: Mutation

Mutations are alterations in the DNA sequence that occur randomly and can have little or great consequences, depending on the location of the mutation. When a mutation altered *SOX3*, the testes-determining gene *SRY* resulted. This mutation had great consequences: The evolution of unique sex chromosomes began.

Learn More: Recombination

During meiosis, chromosomes with substantial DNA sequence homology will pair and exchange pieces of DNA, a process called genetic recombination. This process provides not only a source of genetic variability but also a way in which deleterious mutations are eliminated by not being passed on to future generations. (Recombination allows for reassortment and thus a way for selecting out deleterious mutations without needing to eliminate the entire set of alleles on which the mutation arose.) In this way, recombination preserves the integrity of the chromosomes. The X and Y chromosomes are notably different with respect to recombination. During female meiosis, the two X chromosomes undergo recombination throughout their entire length (illustrated by red lines). In contrast, during male meiosis, the Y chromosome recombines with the X chromosome only at its tips. Thus, over time,

RPS4 (ribosomal protein small subunit, protein 4) is a gene essential for ribosome formation. Identical copies of *RPS4* are found on both the X and the Y chromosome, and the same function has been retained over timeósince before the creation of SRY and through millions of years of divergence between the X and Y. However, the location of *RPS4* has changed as a result of a large inversion.

Learn More: *SRY*

The *SRY* gene regulates the formation of the testes from the undifferentiated embryonic gonad. The location of *SRY* and *SOX3* on the X and Y chromosomes of several species has been compared, an analysis that has allowed scientists to roughly date when changes occurred. Gene pairs, such as *SRY* and *SOX3*, allow researchers to map changes and rearrangements on chromosomes. Specifically, 300 million years ago (Ma), scientists speculate that *SOX3* was in the same location on the proto X and Y chromosomes. A mutation in *SOX3* created the gene *SRY* on the Y chromosome. Researchers found that monotremes are the most ancient mammals that have the *SRY* gene, whereas all earlier ancestors do not. These data allowed researchers to estimate when the mutation that created *SRY* occurred.

Learn more about *SRY* at: "Gender Testing of Female Athletes" http://www.hhmi.org/biointeractive/gendertest/gendertest.html <http://www.hhmi.org/biointeractive/gendertest/gendertest.html>

Resources

Web Resources

"The Book of Y: An Unfinished Story About Maleness" <http://www.hhmi.org/bulletin/bookofy/> Describes the research of David Page and other members of his lab. http://www.hhmi.org/bulletin/bookofy/ <http://www.hhmi.org/bulletin/bookofy/>

The Meaning of Sex: Genes and Gender Lecture Four <http://www.hhmi.org/biointeractive/gender/lectures.html> presented by David Page: "Sexual Evolution: From X to Y" http://www.hhmi.org/biointeractive/gender/lectures.html <http://www.hhmi.org/biointeractive/gender/lectures.html>

"Gender Testing of Female Athletes" http://www.hhmi.org/biointeractive/gendertest/gendertest.html <http://www.hhmi.org/biointeractive/gendertest/gendertest.html>

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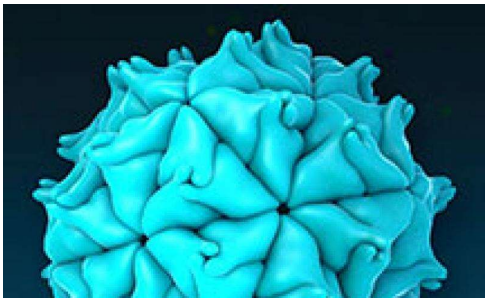
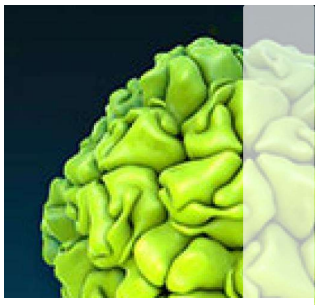

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