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How Ancient Sex Drove Mammalian Lineage Evolution

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In many animals and some plants sex chromosomes play the key role in controlling sex determination. They carry genes to regulate the development of reproductive organs and other sexual characteristics. The evolution of sex determining systems is often connected to sex chromosome turnover – initially a drastic chromosomal change in a single individual that, in spite of potential chromosomal incompatibility, somehow propagates to a whole population. The variety of known sex determining systems suggests, that over evolutionary periods nature again and again found solutions to establish such drastic changes. The question remains do sex chromosome rearrangements promote speciation or are they rather a consequence of it?

In her BioEssays review,[1] Jennifer A. M. Graves's (2016) exciting journey through the evolution and turnover of mammalian sex chromosomes examines this question by looking at their possible relevance to lineage separation. Some 200 million years ago (mya) in the reptile-like synapsid ancestor of mammals, sex-determination was probably similar to the bird ZW system that is driven by the expressed dosage of the Z-borne DMRT1 gene (a double dosage of Z determines a male). In monotremes, the first divergence of mammals, the platypus has ten sex chromosomes (X_{1-5} and Y_{1-5}), and the echidna has nine (X_{1-5} and Y_{1-4}, two fused Y chromosomes) with extensive homologies to bird ZW chromosomes. However, in both monotreme lineages the DMRT1 gene is located on an X chromosome, and an AMH gene located on Y_5 or Y_4 probably took over the male-determining function. Early in therian evolution, 190–166 mya, in the common ancestor of marsupials and eutherians an ordinary autosomal pair of chromosomes underwent extreme rearrangements that initiated a novel sex-determining system. Originating from the drastically modified SOX3 gene, the male-dominant SRY gene located on the novel Y chromosome took over sex determination. Sequences for male-specific function such as spermatogenesis accumulated in the vicinity of the SRY gene. The X chromosome still carries the original SOX3 gene and acquired genes related to sex and intelligence. As the mechanism of dosage compensation evolved, one of the two X chromosomes in females was silenced. In early eutherians, a Robertsonian fusion of the new XY chromosomes with autosomes (A in **Figure 1**, modified after Graves 2016) occured that established our current composition of the XY chromosomes. However, in some mammalian lineages, the XY system might be just an evolutionary intermediate, and, for example, in rodents the Y chromosome may be removed from service, step-by-step, a tendency, also recognized for our own Y chromosome. A turnover of sex chromosomes in spiny Japanese rats shows that an obsolete, error-prone XY system can be reprogrammed for sex determination by a new autosomal gene, and consequently lead to a genetic barrier and speciation. Accordingly, the significant sex chromosome divergences in major mammalian groups should have produced reproductive barriers, and thus, Jennifer A. M. Graves suggests that sex chromosome turnover precipitated the divergence of monotremes, marsupials, and eutherian lineages.

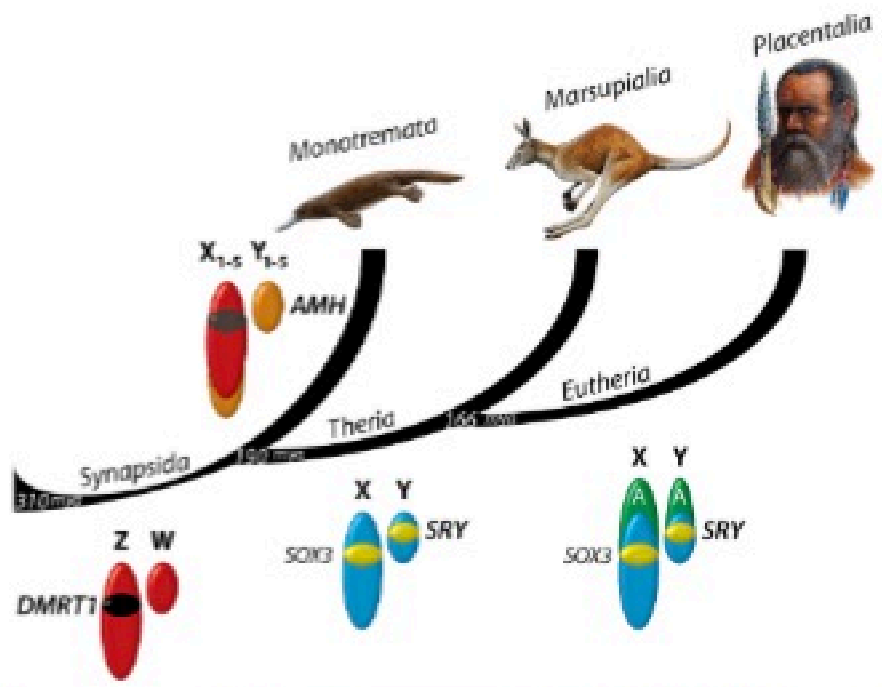

Figure 1. Turnover of sex chromosomes in mammals described in Graves (2016). Animal paintings by Jon Baldur Hlidberg.

Early mammals were not the only vertebrates in which a transition between ZW and XY sex-determination systems occured. While in amphibians and squamate reptiles there are no differences between the transition-rates of ZW and XY, in fishes the transition of ZW to XY was prevalent.[2]

The connection between sex chromosome rearrangements and speciation is being intensively investigated. It was shown that non-recombining regions of any chromosome might present hotspots for incompatibility that may be a key to speciation.[3] In this article, Irwin argues that in ZW and XY sex-determination systems, the Z and X chromosomes can play a dominant role in speciation. However, more empirical data are required to finally conclude which came first: mammalian sex-chromosomal turnover or progressive mammalian divergence.

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[1] J. A. M. Graves, *BioEssays* **2016**, *38*, 734.
[2] M. W. Pennel, J. E. Mank, C. L. Peichel, *Mol. Ecol.* **2018**, https://doi.org/10.1111/mec.14540.
[3] D. E. Irwin, *Mol. Ecol.* **2018**, https://doi.org/10.1111/mec.14537.