The birth of a male sex chromosome in Atlantic herring

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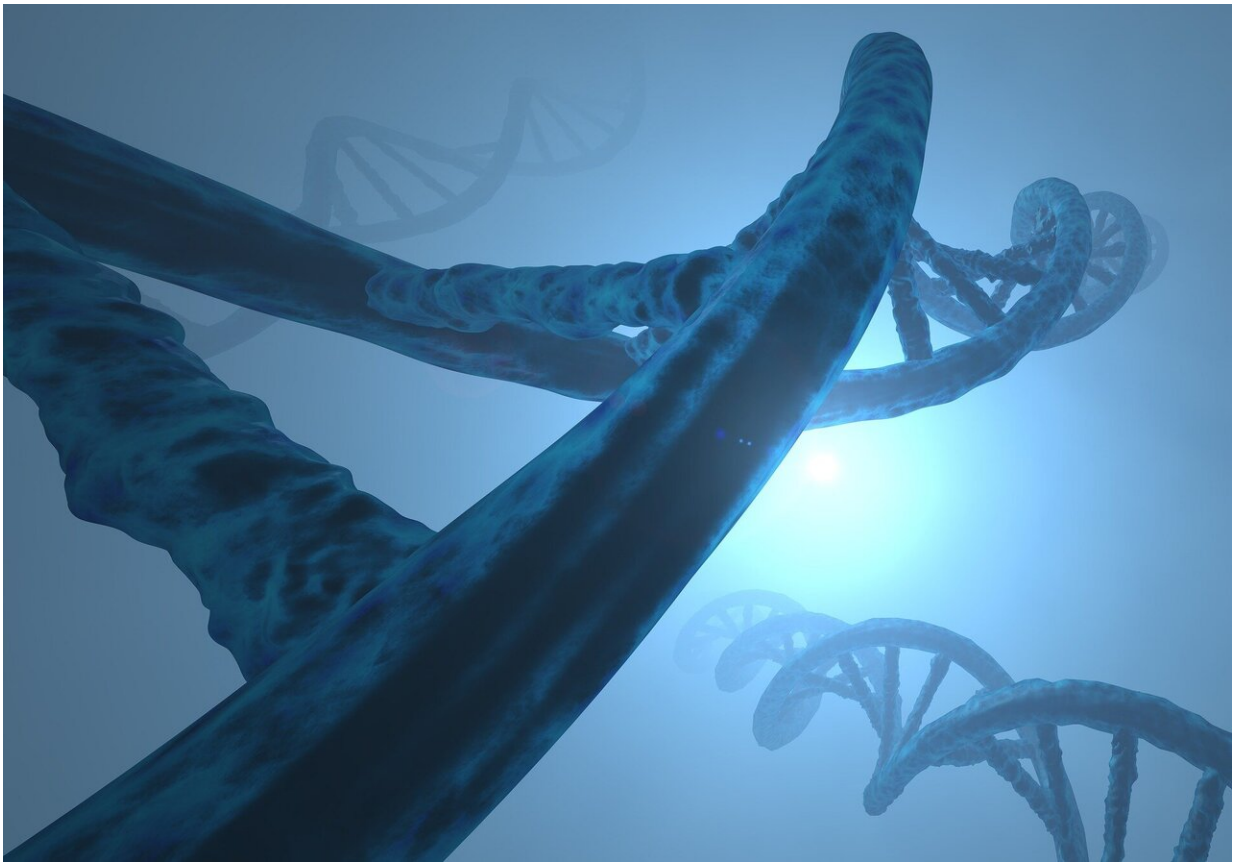

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The evolution of sex chromosomes is of crucial importance in biology as it stabilizes the mechanism underlying sex determination and usually results in an equal sex ratio. An international team of scientists, led by

researchers from Uppsala University, now reports that they have been able to reconstruct the birth of a male sex chromosome in the Atlantic herring. The male-specific region is tiny and contains only three genes: a sex-determining factor and two genes for sperm proteins.

It is hard to study the early evolution of sex chromosomes because it usually happened a long time ago and the sex-determining chromosomes usually rapidly degenerate and accumulate repetitive sequences. For instance, humans have an X/Y system of sex determination and the presence of Y determines male sex. The human Y chromosome, which was established more than 100 million years ago, evolved from a chromosome identical to the X chromosome but has since lost most of the genes present on X and is now only about a third the size of the X chromosome. The Atlantic herring also has an X/Y system but it is young and evolved much more recently. In the herring X and Y are almost identical in gene content, the only difference being that Y has three additional genes: a sex-determining factor (BMPR1BBY) and two sperm protein genes predicted to be essential for male fertility.

"The unique feature of this study is that we have been able to reconstruct the birth of a sex chromosome. The evolution of the herring Y chromosome in fact resembles the process when my son makes a construction with pieces of Lego," says Nima Rafati, scientist at Uppsala University and first author on the paper.

Two of the building blocks were formed when extra copies of two different genes emerged and were translocated to what became a male-specific region that cannot exchange genetic material with the X chromosome. This was followed by the incorporation of a third gene to the male-specific region and its loss from the X chromosome.

"The Y-specific gene BMPR1BBY is most certainly the sex-determining factor in Atlantic herring since it belongs to a family of proteins with a

critical role in inducing the development of testis. The evolution of BMPR1BBY is a wonderful example of molecular evolution in action. It shows how random mutations and natural selection can 'create' a new gene," says Amaury Herpin, scientist at INRAE, France's new National Research Institute for Agriculture, Food and Environment, and one of the shared first authors.

BMPR1BBY contains about 50 mutations compared with the autosomal copy but it maintains its ability to promote testis development and has evolved an ability to act independently of some of the cofactor the autosomal copy requires. It therefore provides a shortcut to the induction of testis development.

"It has previously been proposed that the presence of a sex-determining factor is not sufficient for the evolution of a sex chromosome, it requires a close association between a sex-determining factor and one or more genes beneficial for that sex," explains Manfred Schartl, professor at Würzburg University and one of the co-authors of the study. "This is exactly what the herring Y chromosome provides, a male-determining factor (BMPR1BBY) and two genes for sperm proteins predicted to be essential for male fertility."

"We are now working on a follow-up study by making an assembly of the sprat genome. Sprat is a close relative to the herring and this analysis will allow us to make a more precise estimate of when this Y chromosome evolved, how stable it is and how rapidly it evolves," says Professor Leif Andersson, Uppsala University, who led the study.

More information: Rafati et al. (2020) Reconstruction of the birth of a male sex chromosome present in Atlantic herring, *PNAS*, DOI: 10.1073/pnas.2009925117

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