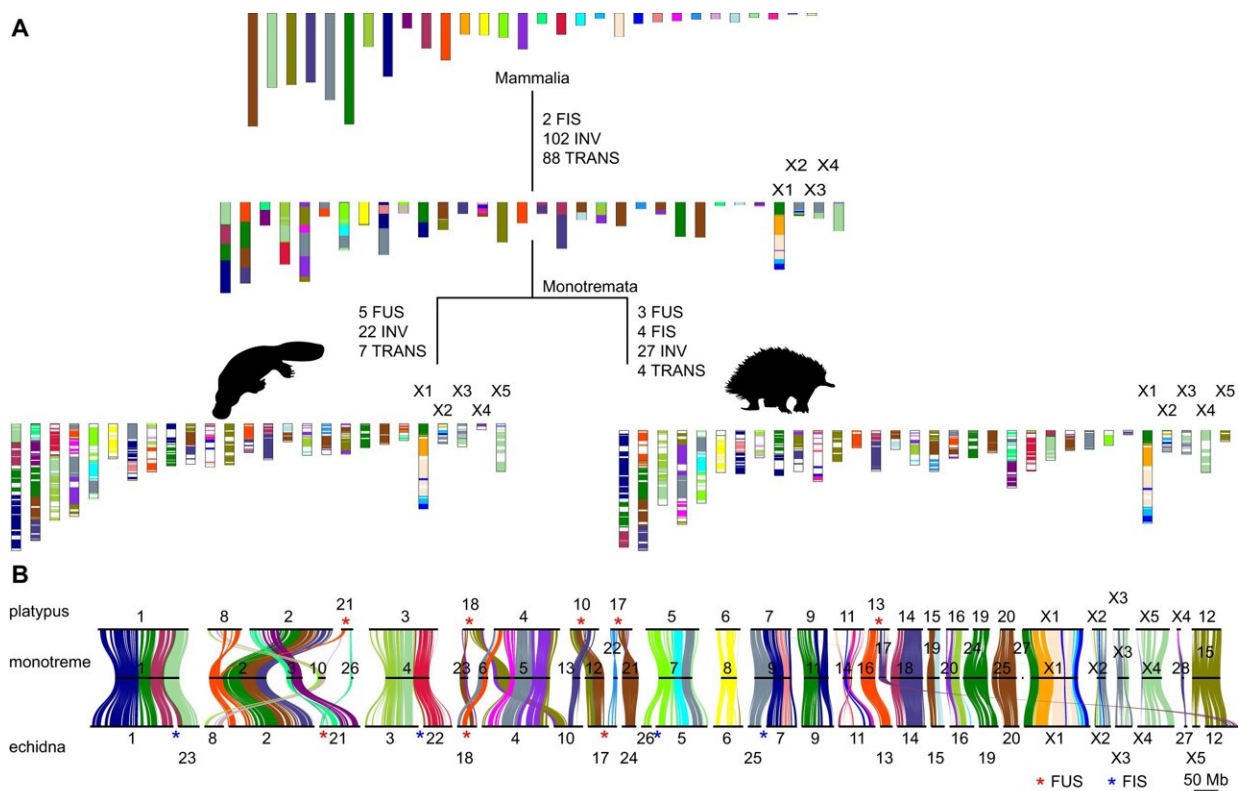


Echidna genome study unravels the events leading to multiple sex chromosomes

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Karyotype evolution of monotremes. Credit: *GigaScience* (2024). DOI: 10.1093/gigascience/giae112

The short-beaked echidna (*Tachyglossus aculeatus*) is one of Australia's most iconic animals. Belonging to a unique group of mammals called "monotremes" (with the platypus as the other prominent member).

Echidnas may at first glance be mistaken for a weird-looking hedgehog, but they are in fact egg-laying mammals.

An international team of researchers, led by Guojie Zhang and Qi Zhou at Zhejiang University, Yang Zhou from BGI-research, and Frank Grutzner from the Adelaide University have now provided a nearly gapless genome sequence of the short-beaked [echidna](#).

The authors used the new data to better understand the evolutionary origin of the highly complex configuration of multiple sex chromosomes, which is characteristic for monotremes. The work is [published](#) in the journal *GigaScience*.

Among the many unusual features of monotremes, one is their position in the tree of life: they separated from other mammals very early, making them the oldest branch in the mammalian tree. This is reflected in their genomes and, in particular, in the way sex is determined genetically.

In the majority of sex determination systems, sex chromosomes typically come as a pair, such as in the familiar X/Y system in humans and in most other mammals.

In this well-known setup, sex is primarily determined by which pair of sex chromosomes the offspring inherit. The sperm carry either an X or a Y chromosome and the egg can carry only an X chromosome.

At fertilization, the sex of the offspring is mainly driven by which of the sperm chromosomes enters the egg, with females having two X chromosomes and males having one X and one Y.

In contrast, monotremes have a much more complex sex determination system, featuring multiple X and Y chromosomes.

Furthermore, there are also differences between the two monotreme groups that separate the echidnas from the platypus families. In male echidna there are nine sex chromosomes while platypus has an even 10.

The male echidna has five X chromosomes, but only four Y chromosomes, whereas the male platypus has five X and five Y).

During production of sperm and eggs (meiosis), these sex chromosomes pair in a head-to-tail manner and form chains, rather than the side-by-side pairing typically seen in organisms with only two sex chromosomes. The head-to-tail pairing in monotremes allows the chromosomes to be distributed in an orderly manner to the germ cells and in a way that doesn't lead to genetic problems.

The echidna genome sequence provided here helps researchers trace the genetic events that led to this remarkable chromosomal arrangement. The high-quality sequence with very few gaps presented here allows scientists to infer events like chromosomal rearrangements in the ancestry line of monotremes.

Zhang says, "The high-quality reference genome of echidna allows us to infer which chromosomes are shared between echidna and platypus since they diverged from other mammals, and which [chromosomes] were newly evolved after the two monotreme families split."

The evolutionary lineages of echidnas and platypus diverged around 55 million years ago, and there is evidence that chromosome fusion and fission events might have occurred since their divergence.

Using both the new echidna genome sequence and an existing platypus genome sequence, the international team of researchers reconstructed the likely chromosome composition of the monotreme ancestor.

They also used chromosomal assemblies of placentals (human, bovine and sloth), marsupials (opossum and Tasmanian devil), and reptilian outgroups (chicken, turtle and the common wall lizard) in their research.

Analysis of these sex chromosomes revealed that the ancestral X chromosome of monotremes had exchanged segments with ancestral non-sex chromosomes (autosomes), which gave rise to their complex system.

In addition to these structural changes in genome organization, the authors also found signals of gene family expansion, meaning that additional copies of specific genes arose during the evolution of monotremes.

One such event concerns a gene called SYCP3Y. This gene encodes a component of a protein structure called the synaptonemal complex, which connects chromosome pairs during the formation of germ cells.

SYCP3Y is duplicated from autosomes and expanded in both platypus and echidna, suggesting that the gene expansion may be associated with the evolution of the sex chromosome complex or play a role in its organization.

Overall, the new results provide a detailed evolutionary history of the highly unusual multiple-sex chromosome system in monotremes and demonstrate how today's high-resolution genomic methods can uncover complex evolutionary events that shaped the mammalian tree of life millions of years ago.

The details of this process, and the intriguing question of how and why this complicated system originated in the first place, is still subject to ongoing research, but the data here take us a step further into understanding these events and contributing to answering these questions.

More information: Guojie Zhang et al, Chromosome-level echidna genome illuminates evolution of multiple-sex-chromosome system in monotremes, *GigaScience* (2024). [DOI: 10.1093/gigascience/giae112](https://doi.org/10.1093/gigascience/giae112)

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