



Contents lists available at ScienceDirect

General and Comparative Endocrinology

journal homepage: www.elsevier.com/locate/ygcen

Implications of monotreme and marsupial chromosome evolution on sex determination and differentiation

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ARTICLE INFO

Article history:

Received 18 May 2015

Revised 15 September 2015

Accepted 26 September 2015

Available online xxxx

Keywords:

Karyotype

Genome evolution

Sex chromosomes

Sex determination

Intersex

ABSTRACT

Studies of chromosomes from monotremes and marsupials endemic to Australasia have provided important insight into the evolution of their genomes as well as uncovering fundamental differences in their sex determination/differentiation pathways. Great advances have been made this century into solving the mystery of the complicated sex chromosome system in monotremes. Monotremes possess multiple different X and Y chromosomes and a candidate sex determining gene has been identified. Even greater advancements have been made for marsupials, with reconstruction of the ancestral karyotype enabling the evolutionary history of marsupial chromosomes to be determined. Furthermore, the study of sex chromosomes in intersex marsupials has afforded insight into differences in the sexual differentiation pathway between marsupials and eutherians, together with experiments showing the insensitivity of the mammary glands, pouch and scrotum to exogenous hormones, led to the hypothesis that there is a gene (or genes) on the X chromosome responsible for the development of either pouch or scrotum. This review highlights the major advancements made towards understanding chromosome evolution and how this has impacted on our understanding of sex determination and differentiation in these interesting mammals.

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1. Introduction

Monotremes (prototheria) and marsupials (metatheria) are perhaps the animals most commonly associated with Australasia. Extant monotremes, represented by one species of platypus and four species of echidna, are endemic to Australasia. Marsupials are distributed across the Americas and Australasia but approximately 75% of species, representing four orders, are endemic to Australasia. These two major mammalian lineages last shared a common ancestor around 180 million years ago (Phillips et al., 2009), when the monotreme lineage diverged to become the most basal mammals, prior to marsupials diverging from their eutherians counterparts around 160 million years ago (Luo et al., 2011). Monotremes and marsupials are distinguished by their mode of reproduction, with monotremes being the only egg-laying mammals and marsupials giving birth to highly altricial young that depend on a long lactation to complete their development, typically within a pouch although the types of pouches range from skin flaps on the side of the abdomen to fully enclosing the mammary area (Edwards and Deakin, 2013).

One of the striking differences between these two mammalian lineages, besides their mode of reproduction, is in how their genomes are packaged. Monotreme genomes are divided into six large and easily distinguished pairs of chromosomes and many smaller chromosomes. In contrast, marsupial genomes are divided into a small number of very large chromosomes. However, the most striking difference between monotremes and marsupials is their sex chromosomes: monotremes have complex, multiple sex chromosomes that form a chain during meiosis in males, whereas marsupials typically have a simple XX female/XY male system. This review describes the current understanding of the evolution of monotreme and marsupial chromosomes, focusing on the sex chromosomes, and the latest developments leading to the identification of the genetic control of sex determination and differentiation in these animals.

2. Monotreme chromosomes

The first descriptions of monotreme chromosomes in the mid 20th century proved confusing, with differences reported in the number of chromosomes making up a full diploid complement (reviewed in Murtagh and Sharman, 2009). Eventually, a diploid

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number of 52 chromosomes was reported for the platypus (*Ornithorhynchus anatinus*) (Bick and Sharman, 1975) and 63 and 64 in male and female echidnas respectively (Bick and Jackson, 1967).

Comparisons between the platypus and short-beaked echidna (*Tachyglossus aculeatus*) karyotypes have been made using chromosome painting, where individual chromosome probes for platypus chromosomes have been isolated, fluorescently labeled and hybridized to echidna chromosomes and the reverse experiment has been performed as well (Rens et al., 2007). Eight chromosomes are entirely homologous between the two species (Fig. 1). All other chromosomes have arisen largely from Robertsonian fusion or fission events in either species (Rens et al., 2007). Unfortunately, it is not possible at this stage to determine in which direction these fission/fusion events occurred from a common ancestor or to reconstruct the monotreme ancestral chromosome arrangement, as this would require comparison to outgroup species. Such comparisons would be possible if the platypus genome was anchored to chromosomes but, to date, only about 20% of the genome assembly has been given a chromosomal assignment (Warren et al., 2008).

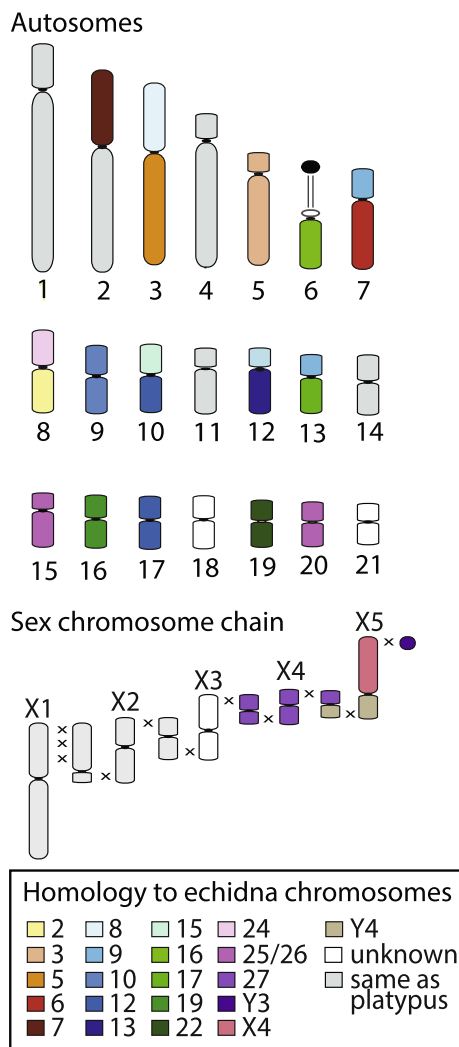


Fig. 1. Homology of platypus autosomes and the male sex chromosome chain to echidna chromosomes. Regions pairing between X and Y chromosomes are indicated by crosses.

2.1. Monotreme sex chromosomes

The most controversial point regarding monotreme chromosomes revolved around the identification of the sex chromosomes, a problem that was not resolved until a molecular cytogenetic technique known as chromosome painting was used to distinguish some of the very small chromosomes, demonstrating that there are multiple sex chromosomes in monotremes: five different X and Y chromosomes in platypus (Grützner et al., 2004; Rens et al., 2004) and the short beaked echidna has five X and four Y chromosomes (Rens et al., 2007). In both species, these chromosomes form a multivalent chain during male meiosis (Murtagh, 1977).

In the platypus, the sex chromosomes in the multivalent chain share a homologous region (pseudautosomal) with the next chromosome in the chain that permits the chromosomes to pair during meiosis (Fig. 1). This means that a region on X_1 pairs with one end of Y_1 , the opposite end of Y_1 pairs with X_2 and so forth (Grützner et al., 2004; Rens et al., 2004). There are similar pseudautosomal regions on echidna sex chromosomes (Rens et al., 2007). From chromosome painting results, it remained unclear whether there were pseudautosomal regions present on echidna Y_3 that would allow pairing with X_3 and X_4 . Mapping of Major Histocompatibility Complex (MHC) genes to platypus and echidna chromosomes revealed a region of homology shared between echidna X_3 and Y_3 (Dohm et al., 2007). However, a region of homology between Y_3 and X_4 is yet to be detected.

The chromosomes involved in this multivalent chain are not entirely homologous between the platypus and echidna. By using platypus chromosome probes in chromosome painting experiments on echidna chromosomes and vice versa, it became clear that there are some major differences. Firstly, the smaller number of Y chromosomes in the echidna is accounted for by a fusion between the equivalent of platypus Y_3 and Y_5 (Dohm et al., 2007; Rens et al., 2007). Secondly, platypus X_4 corresponds to an echidna autosome (chromosome 27) (Fig. 1). Similarly, echidna X_5 is homologous to the short arm of platypus chromosome 12. This difference in chain content between the species is explained by a chain of eight chromosomes having formed through a series of fusions between the original sex chromosome and autosomes prior to platypus/echidna divergence. Subsequent fusions of different autosomes into the chain occurred independently in the different lineages (Rens et al., 2007).

Although it has been difficult to reconstruct the general evolutionary history of monotreme genomes, a more concerted effort has been made to assign sequence to their sex chromosomes, permitting comparisons to outgroup species, such as chicken and other mammals (Veyrunes et al., 2008). Such comparisons have resulted in intriguing findings regarding the evolution of the unusual sex chromosome system. Assignment of platypus genome sequence scaffolds to chromosomes, in combination with additional gene mapping, has shown that four of the 10 sex chromosomes in males share homology with the chicken Z but have no homology to X of therian (marsupial and eutherian) mammals (Veyrunes et al., 2008). This means that the same genes are present on the sex chromosomes in a female heterogametic sex chromosome system in birds and a male heterogametic sex chromosome system in monotremes.

2.2. Candidate sex determination gene

A major goal in understanding the evolutionary history of monotreme sex chromosomes is to uncover the master sex determining switch. In eutherian mammals, the sex determination cascade is switched on by the *SRY* gene (Foster et al., 1992; Koopman et al., 1991). This gene evolved from *SOX3* (Foster and Graves, 1994), an autosomal gene in monotremes, after the divergence of

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