A first look at monotreme meiotic recombination

Master's thesis

Sonia Horan
Supervisors:
Dr. Frank Grützner
Dr. Enkhjargal Tsend-Ayush
Dr. Tasman Daish

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Abstract

Recombination is a critical event in meiosis required for proper chromosome segregation; however, despite its importance, crossovers have not yet been analysed in egg-laying mammals, representing an extant species from the most basal of the contemporary mammalian lineage. The platypus is a unique species in that it has a complex system of ten sex chromosomes that form a chain in meiosis, and the segregation of this chain raises many questions regarding the state of meiotic recombination among chain members. This study aimed to set a foundation for recombination studies in the platypus. 15 genes essential for crossover formation were analysed; while most of these genes showed generally high levels of conservation, surprisingly it was found that Mei4 and Rec114 are not expressed during platypus meiosis. This raises several questions about the platypus double-strand break induction mechanism in which these genes have an essential function in other mammals. Furthermore, characterisation of the crossover hotspot-determining protein, Prdm9, shows that the platypus protein differs significantly from the Prdm9 of eutherians, suggesting that the gene underwent rapid evolution after the divergence of monotremes nearly 200 Million years ago. This study also aimed to examine the distribution of crossovers in meiotic cells using Mlh1 as a marker for crossover events. Despite conservation of the protein and expression and some specific staining overall, crossover counts could not be analysed in platypus male meiotic cells. Surprisingly another protein which can be found in earlier meiotic stages, Dmc1, showed a very distinct pattern in platypus pachytene cells. It was observed that an average of 24 Dmc1 foci were present in platypus pachytene, long after Dmc1 is expected to have diminished from chromosome cores. These foci seemed to localise to the ends of chromosomes and were more prominently associated with the sex chromosomes, particularly the pseudoautosomal regions. Together with other evidence this suggests that the Dmc1 antibody may be cross-reacting with Rad51, which plays a structural role in pachytene, holding chromosomes together at the telomeres. Overall this work provides first evidence for a general conservation of genes involved in the recombination pathway but has also identified some genes that may have diversified in their function.

Thesis declaration

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Sonia Adeline Horan
postgraduate student

* Figure 1 (page 15 Whitby, 2005), Figure 2 (page 21 Shinohara & Shinohara, 2004), Figure 3 A (page 28 Oliver-Bonet, Turek, Sun, Ko, & Martin, 2005), Figure 4 (page 37 Grützner, Deakin, Rens, El-Mogharbel, & Marshall Graves, 2003), Figure 5 (page 38 Daish, Casey, & Grutzner, 2009), and Figure 37 (page 94, Veyrunes et al., 2008).

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