

# A first look at monotreme meiotic recombination

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Master's thesis

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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

This work contains no material which has been accepted for the award of any other degree or diploma in any university or other tertiary institution to Sonia Adeline Horan and, to the best of my knowledge and belief, contains no material previously published or written by another person, except where due reference has been made in the text. I give consent to this copy of my thesis, when deposited in the University Library, being made available for loan and photocopying, subject to the provisions of the Copyright Act 1968.

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\* 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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