

## Biology 305 Exam II Abstract

*This abstract will be part of the first exam. You do not need to read the actual paper. Your goal before the exam is to make sure you understand the concepts and ideas presented in the abstract. You may talk with friends (in fact I hope that you do) and ask me anything about the information presented below.*

Evans *et al.* (2006) Germline mosaicism for a MECP2 mutation in a man with two Rett daughters. *Clinical Genetics* **70**:336-8.

Rett syndrome is a severe neurodevelopmental disorder that is caused by mutations in the X-linked gene, methyl-CpG binding protein 2 (MECP2). The majority of cases are sporadic, but rarely germline mosaicism can lead to familial cases. Here, we report the first case where germline mosaicism for a MECP2 mutation has been shown in a man. He has two affected daughters who are half sisters, and both have the c.808delC mutation. We show that this mutation is present at a low level in DNA extracted from the patient's semen. This case has implications for genetic counseling, and pre-natal testing should be offered for the partners of men who have a daughter with Rett syndrome.