



## **Seminar Announcement**

"A Drosophila model of 15q Duplication Syndrome"



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Friday August 14, 2015 | 11:00 a.m. Donnelly Centre Red Seminar Room

## **Abstract:**

My laboratory utilizes the powerful genetic model organism Drosophila melanogaster (fruit flies) to investigate the functions of genes involved in human neurogenetic disease. The main focus of the laboratory is autism spectrum disorders, specifically 15q Duplication syndrome. We have established in clinical studies of interstitial duplication 15q cases that maternal duplication of this region results in the ASD phenotype, clearly implicating the human UBE3A gene in the pathogenesis. However, other phenotypes in the syndrome like seizure may not be driven by UBE3A alone. We have now identified 50 potential Dube3a regulated proteins and are actively validating these interactions using whole genome molecular methods in human cell lines, genetic suppressor/enhancer screens, protein expression in fly neurons, and changes in synaptic function and stability in flies. Dr. Reiter will present new data from a whole genome deficiency screen that suggests a role for both GABA B3 and glial cells in the seizure phenotype observed in 15q Duplication syndrome.

**Host: Timothy R. Hughes**