Department of Molecular and Cellular Biology

Graduate Seminar MCB*6500

Friday, Sept. 14, 2018 in SSC 1511 @ 12:45 p.m.

presented by:

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(Advisor: J. Dawson)

"Characterizing hypertrophic cardiomyopathy linked S271F ACTC variant and dilated cardiomyopathy linked T126I ACTC variant"

Heart disease is the number one cause of death worldwide, specifically costing the Canadian economy 21.2 billion dollars yearly. The most commonly inherited heart disease is cardiomyopathy and is the result of abnormal cardiac muscle. The two main types of cardiomyopathy are dilated cardiomyopathy (DCM), where the walls of the ventricles are thin due to weakened cardiac muscle, and hypertrophic cardiomyopathy (HCM), where the walls of the ventricles are thick and stiff. Abnormal cardiac muscle can result in systolic dysfunction and even sudden cardiac death. Many genetic mutations, including mutations in *ACTC1* encoding α-cardiac actin (ACTC), have been linked to the pathogenesis of DCM and HCM. The ACTC protein plays a critical role in the contractility of the heart muscle. Changes in ACTC affect its intrinsic properties and interaction with other proteins, and potentially muscle contractility. Within the field, the general model is that HCM is the result of increased contractility, while DCM is more commonly the result of decreased contractility. Currently, there are 16 ACTC variants found independently in patients with cardiomyopathy. Of these, the DCM-linked T126I ACTC variant and the HCM-linked S271F ACTC variant have yet to be characterized. I aim to determine if these variants exhibit characteristics consistent with DCM or HCM, respectively. The findings of my research will shine light on the molecular mechanisms contributing to cardiomyopathy and heart failure.