

Biology Seminar



Western
UNIVERSITY · CANADA

12:30 - 1:30 pm
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WSC 240



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A Canadian Framework for Unsolved Genetic Diseases: Matchmaking, Care4Rare-SOLVE and RDMM

Canada's national rare disease (RD) strategy is to understand the molecular pathogenesis of unsolved RDs and improve access to personalized care. Despite recent advances in genome-wide testing, we are still unable to diagnose more than half of RD patients. Diagnosis is "care" for RD patients; without it, there is no prognosis, counseling or specific treatment. Care4Rare is a pan-Canadian consortium focused on improving the diagnosis and treatment of RD's. Its new installment, Care4Rare-SOLVE, aims to facilitate access to clinical genome-wide sequencing across Canada, and increase the solve rate of RD's by employing innovative 'omic approaches. Model organisms remain the most powerful tools to confirm pathogenicity and characterize function of novel genes, as well as identify potential therapies; zebrafish being among the most lucrative. Rare Disease Models and Mechanisms (RDMM) Network was established in Canada to connect clinicians discovering novel RD genes with scientists able to study equivalent pathways in model organisms. Many zebrafish models have been developed through RDMM, mostly using the powerful CRISPR technology. Examples of such models that were able to validate pathogenicity of novel disease genes and identify personalized therapies will be discussed and the local plan will be outlined.

