



Medical Genetics Seminar Series

Emma Gillesse, BSc

Department of Medical Genetics
University of Calgary

“Experimental validation of novel variants detected by genome sequencing”

Friday, May 10, 2023
9:00 am – 10:00 am
HSC O’1500

LEARNING OBJECTIVES:

- 1) Discuss approach to genome analysis and overall genetic testing for individuals with rare genetic disease
- 2) Evaluate the value of yeast as a model for functional validation of novel variants and variants of unknown significance detected in the gene *ALG1*
- 3) Outline an approach to functional validation of a recurrent non-coding variant detected in a Canadian founder population