C2E2SPOTLIGHT

CENTRE for Clinical Epidemiology and Evaluation

March 11, 2024

Silent Genomes: Economics of Precision Diagnosis in Indigenous Children

Genomic testing is used to diagnose genetic causes of childhood rare diseases. Genomic testing benefits are not equitably distributed to Indigenous peoples because they are underrepresented in the variant libraries needed for results interpretation. Research-based whole-genome sequencing can provide additional diagnoses to Indigenous families. Translating whole-genome sequencing benefits into real-world clinical settings should aim for safe, valued, and accessible implementation, led by Indigenous voices. This talk will provide a brief overview of our Indigenous-partnered Silent Genomes project that looked at how to evolve economic approaches to understand the role of genomic testing to diagnose rare conditions in Indigenous children.

Our learning objectives are to:

1. Discuss Indigenous-partnered research related to economics, innovation, and access;
2. Overview preliminary outcomes of examining the potential role of genomic testing for diagnosing rare disease in children.

Presenter	**Dean Regier, PhD** Senior Scientist, BC Cancer Research Institute Associate Director, Academy of Translational Medicine, University of British Columbia

C2E2 Spotlight is held monthly on Mondays from 12:00 pm to 1:00 pm. To attend in-person or virtually please contact pamela.lee@ubc.ca.