MEDICAL GENETICS ACADEMIC HALF DAY  
October 11, 2019 – MUHC Glen site, Room B02.9390
[Session from 9:00 – 10:00 will be videoconferenced to the JGH Library Conference room 1.]

9:00 – 10:00  Genetics Research Presentation
[Hosted and Introduced by Geneviève Bernard, MD, MSc, FRCPc, Pediatric Neurologist]  
Stefanie Perrier, PhD Candidate  
Integrated Program in Neuroscience  
McGill University, Department of Neurology and Neurosurgery  
POLR3-Related Leukodystrophy: Defining Novel Phenotypes from Very Mild to Extremely Severe

Learning Objectives:
This presentation will explore POLR3-related leukodystrophy in various levels of severity and describe novel genotype-phenotype correlations.  
At the end of the session, the attendee will be able to:  
- Describe the typical POLR3-related leukodystrophy clinical presentation, disease course, and MRI pattern  
- Identify novel disease presentations from very mild to extremely severe  
- Understand the pathogenicity of the severe disease phenotype on the tissue, cellular, and molecular level

10:00 – 10:30  Clinical Case Review  
Ali López Sarmiento, MD  
Anatomical Pathology, PGY-3  
Respiratory distress in a 1-day-old term newborn with an unexpected chest X-ray

10:30 – 12:00  Resident Teaching  
Active Listening Skills Workshop  
Tel-Aide Training team  
http://www.telaide.org/en/

The Medical Genetics Academic Half Day is a self-approved group learning activity (Section 1) as defined by the Maintenance of Certification program of the Royal College of Physicians and Surgeons of Canada.

Do not hesitate to contact me should you have any questions.

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