MEDICAL GENETICS ACADEMIC HALF DAY  
February 9, 2018  
MUHC Glen Site  
Room B 02.9390  
[Sessions from 9:00 – 10:30 will be teleconferenced to the JGH, Room H-347.]  
Please note time and room change for Resident Teaching session, below.

9:00 – 10:00  Genetics Rounds  
*Dysmorphology Bootcamp, Continued!*  
Discussion led by Mehdi Yeganeh, MD, PhD  
PGY3 in Genetics

Learning objectives:  
At the end of this session, participants will be able to:  
1. Identify and define the terminology used to describe physical features  
2. Use proper terminology to describe patients with dysmorphic features  
3. Identify various genetic syndromes based on dysmorphic features

10:00 – 10:30  Journal Club  
Jennyphane Alexandre, BA  
1st year genetic counselling student  
*A Homozygous Potentially Pathogenic Variant In The PAXBP1 Gene In A Large Family With Global Developmental Delay And Myopathic Hypotonia*  
[Please see attached article.]

10:30 – 12:00  Resident Teaching  
B 02.6645  
Stephanie Fox, MS, MSc, CGC, CCGC  
Genetic Counsellor, MUHC  
*Introduction to Risk Assessment & Pedigree Analysis*  
Reading assignment will follow!

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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