



MEDICAL GENETICS ACADEMIC HALF DAY

March 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 room change for 10:30 Resident Teaching session, below.

9:00 – 10:00 Genetics Rounds

[Sponsored by Isabelle De Bie, MD, PhD and the Prenatal Diagnosis team of the MUHC]

Karen Wou, MDCM, FRCSC

Obstetrician & Gynecologist

Maternal-Fetal Medicine Specialist

Clinical Geneticist

Department of Obstetrics and Gynecology, Division of Maternal Fetal Medicine, MUHC

Cree Board of Health and Social Services of James Bay - Chief of OBGYN service

Prenatal Applications of Whole Exome Sequencing

Learning objectives:

At the end of this session, participants will be able to:

- Describe the landscape of prenatal diagnosis
- Review the current literature around prenatal WES
- Assess the WES testing process with promises and pitfalls
- Discuss the clinical utility of diagnostic WES in the prenatal setting through cases

10:00 – 10:30 Journal Club

Andrea Gomez, PhD

Clinical Specialist in Medical Biology

Core Molecular Diagnostic Laboratory, MUHC

CCMG Fellow in Molecular Genetics, Department of Human Genetics

Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy

Mercuri E et al. *N Engl J Med* 2018; 378:625-35.

[Please see attached article.]

10:30 – 12:00 Resident Teaching

B 02.6645

Stephanie Fox, MSc, MS, CGC, CCGC

Risk Assessment III: Bayes' Theorem and Risk Communication

Please bring a calculator and paper to work on problems. Also, please read:

- Bennett et al. Genetic Counselling and Screening of Consanguineous Couples and their Offspring: Recommendations of the National Society of Genetic Counselors - *J Genet Counsel* 2002; 11(2)
- Optional: Ogino & Wilson. Bayesian Analysis and Risk Assessment in Genetic Counseling and Testing – *Journal of Molecular Diagnostics* 2004; 6(1)

Canada.

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

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