EXCELLENCE IN GENETICS & IMMUNOLOGY
SEMINAR SERIES

Aleixo Muise, MD, PhD, FRCPC

Co-Director SickKids Inflammatory Bowel Disease Centre
Clinician-Scientist, Research Institute Hospital for Sick Children
Associate Professor, Departments of Pediatrics, IMS and Biochemistry
University of Toronto

Title: "Genetic and Functional Studies in Very Early Onset IBD (VEOIBD): Precision Medicine in IBD"

Monday, November 2, 2015

Martin Amphitheatre | Room 504, 12:00 PM

McIntyre Medical Sciences Building

My clinical work and laboratory research is focused on understanding the genetic susceptibility and function of identified genes in pathogenesis of Very Early Onset Inflammatory Bowel disease (VEOIBD; diagnosed prior to 6 years of age and infantile disease). This has led to a number of publications from my laboratory describing novel genetic and functional studies in IBD (E-cadherin, RAC1, and PTPRS) and VEOIBD. Most importantly, our genetic analysis has lead to curative treatments in a number of Canadian and international VEOIBD patients with IL10R, XIAP, and LRBA mutations. Furthermore our lab described a novel form of VEOIBD with severe apoptotic entero-colitis and identified the causative mutations – we termed TTC7A-deficiency. We have shown that mutations in the TTC7A gene result in the severe phenotype through disruption of PI4K signaling and that the PI4K-TTC7A-EFR3B pathway is critical in development of this disease. We identified a causative PLVAP mutation resulting in a novel form of sieving Protein Losing Enteropathy (PLE) characterized by hypoproteinemia, hypoalbuminemia, and hypertriglyceridemia. We have also identified rare functional variants in the NADPH oxidase genes NOX2, NCF1/2/4, RAC1/2, and the enterocyte NADPH oxidases DUOX2 and NOX1, as well as iNOS and IL10R that lead to risk of developing VEOIBD and a hope to identify novel treatment strategies based on these genetic findings.

This seminar is mandatory for Biochemistry Graduate students

LOCATION: **McIntyre Medical Sciences Building, Room 504, 12:00 PM**

HOSTED BY: DRS SAMANTHA GRUENHEID & SILVIA VIDAL