

## PARTINGTON LECTURE SERIES

### Department of Pediatrics & Department of Psychiatry Combined Grand Rounds

Presented by:



### **Dr. Kym Boycott**

MD, PhD, FRCPC, FCCMG

Clinical Geneticist, Children's Hospital of Eastern Ontario

Senior Scientist, Children's Hospital of Eastern Ontario Research Institute

Professor, Department of Pediatrics, University of Ottawa

## **“Canada’s path forward for rare diseases: Discovery to translation”**

**Date: Tuesday, November 22, 2016**

**Time: 8:00 am – 9:00 am**

**Place: Richardson Labs Amphitheater**

### **Learning objectives:**

1. Summarize the progress and insights from NGS-based rare disease-causing gene discovery;
2. Describe the tools and platforms developed to enable gene discovery and mechanistic investigation of rare disease, and
3. Illustrate the diagnostic and therapeutic opportunities identified.

### **Resident Teaching Session (0930-1130h)**

**“The Role of genome-wide sequencing in clinical care”**

### **Learning Objectives:**

1. Summarize the diagnostic utility of genome-wide sequencing for patients with rare diseases;

2. Describe the current clinical indications of genome-wide sequencing in pediatrics; and,
3. Illustrative cases for discussion.

### **Biography**

Kym Boycott is a Medical Geneticist at the Children’s Hospital of Eastern Ontario (CHEO), Senior Scientist at the CHEO Research Institute, and Professor of Pediatrics at the University of Ottawa.

Dr. Boycott’s research program in rare diseases bridges clinical medicine to basic research and is focused on understanding the molecular pathogenesis of these disorders, enabling the design of new therapies to ultimately benefit patients and their families.

She is the principal investigator of Canada’s national genome-wide sequencing platform for rare disease, Care4Rare Canada, and of the Rare Diseases: Models & Mechanisms Network, established to catalyze connections between clinical investigators discovering new genes in patients with rare diseases and basic scientists who can analyze equivalent genes and pathways in model organisms.

Dr. Boycott moves the international rare disease agenda forward through her role as the Chair of the Diagnostics Committee of the International Rare Diseases Research Consortium.

