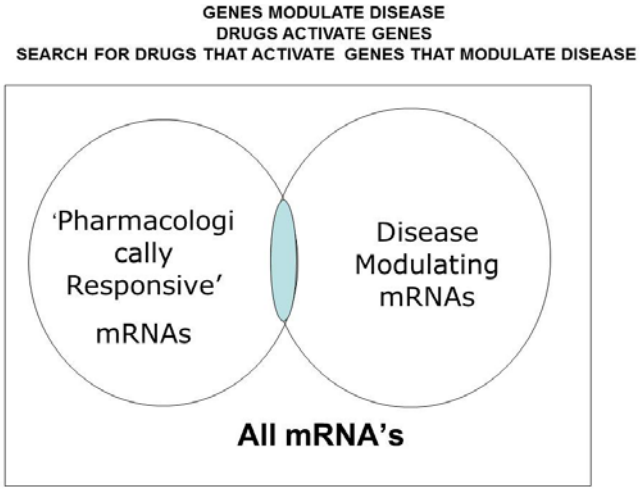
GUEST SPEAKER SEMINAR

Molecular Genetics
UNIVERSITY OF TORONTO

Novel therapeutic approaches for rare genetic disease; after the deluge



The disruptive technology of NGS has resulted in a profusion of novel rare disease (RD) gene discovery underscoring the comparative dearth of effective treatments; over half of the estimated 7000 RDs are solved but there are only 400-500 therapies. At Care for Rare we are attempting to identify novel potentially generalizable approaches for RD therapy. Genetic disorders can be viewed as conditions of pathologic dosage with increased activity or even more frequently decreased dose underlying the pathogenesis; we are screening the modern pharmacopeia for agents which rectify this dosage as well as conducting high throughput screens for reversion of defined RD cellular phenotypes; illustrative cases involving spinal muscular atrophy, myotonic dystrophy and Loeys–Dietz syndrome will be presented.

Dr. Alex MacKenzie

Principal Investigator, Apoptosis Research Centre,
CHEO Research Institute
Physician, Division of Endocrinology, CHEO
Professor, Department of Pediatrics, Faculty of Medicine,
University of Ottawa

Host: Dr. James Dowling

Date: Monday September 15th, 2014
Time: 4PM
Place: Fitzgerald Building, 150 College Street, Room 103