GUEST SPEAKER SEMINAR

Molecular Genetics
UNIVERSITY OF TORONTO

Pathogenesis of FIG4 mutations and phosphoinositide deficiency in neurological disorders

Neurons as post-mitotic cells are susceptible to defects in protein turnover and lysosome function. Positional cloning of a spontaneous mouse mutant with degeneration of the CNS and PNS led us to characterize the endolysosomal signalling lipid PI(3,5)P2 and the effects of deficiency in the nervous system. FIG4 mutations in patients with Charcot-Marie-Tooth disease, Yunis-Varon Syndrome and polymicrogyria with epilepsy define an allele series with a spectrum of clinical severity. Mechanistic studies in conditional mouse mutants and cultured cells suggest potential therapeutic pathways for these debilitating neurological disorders.

Dr. Miriam Meisler

Myron Levine Distinguished University Professor
Professor of Human Genetics, Professor of Neurology
University of Michigan

Host: Dr. Lucy Osborne

Date: Monday October 6th, 2014
Time: 4PM
Place: Fitzgerald Building, 150 College Street, Room 103