## SickKids Research INSTITUTE

## 2018 Faculty Candidate Seminar Rare Disease Genetics and Genomics

## Exploring the frontiers of genetic medicine in rare diseases

Dwi Kemaladewi, PhD Research Associate Genetics & Genome Biology Program SickKids Research Institute

Wednesday January 24, 2018 2PM Robert B Salter Auditorium Peter Gilgan Centre for Research and Learning 686 Bay St.



Dr. Kemaladewi has a strong interest in understanding the molecular mechanisms underlying neuromuscular disorders (NMD) and translating this knowledge into a variety of therapeutic strategies. Her entry point to the NMD field was on the development of antisense oligonucleotides and CRISPR/Cas9 as therapeutic tools for Duchenne muscular dystrophy. Leveraging her expertise in splicing modulation, genome editing, and knowledge of NMD pathophysiology, she subsequently embarks on studying LAMA2-deficient congenital muscular dystrophy (LAMA2-CMD). Her current research involves identification of novel disease modifier and utilization of CRISPR/Cas9 technology to correct splicing defect and modulate expression of compensatory genes in LAMA2-CMD mouse and patient-derived cellular models.