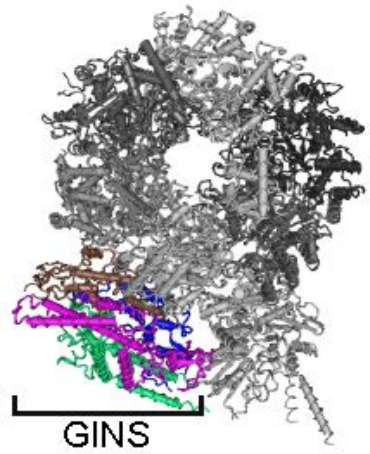
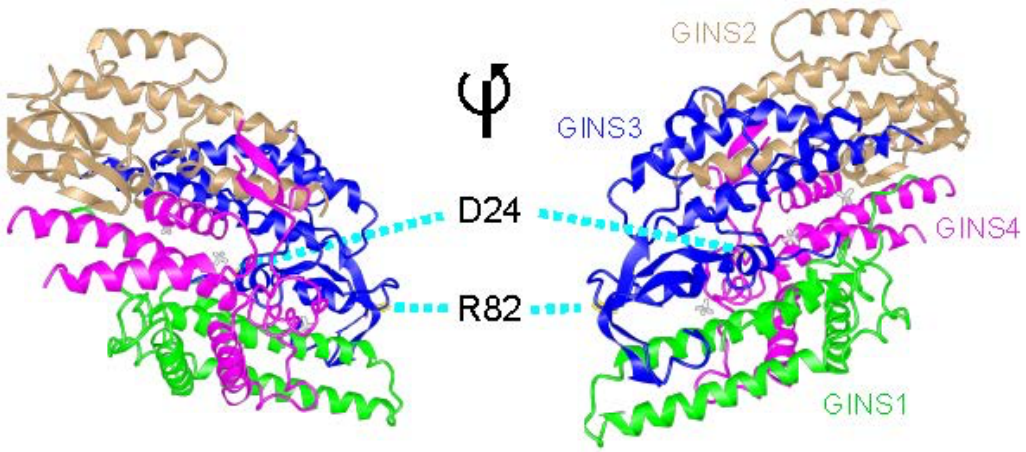
RESEARCH INSTITUTE SEMINAR SERIES

RESEARCH CONNECTIONS

A Research Institute seminar series showcasing successful partnerships between researchers in different research programs or in basic and clinical research. Each seminar highlights the connections that have led to impactful discoveries and stimulate future productivity.





Featured Speakers

PETER KANNU, MB ChB (Otago), PhD, DCH, FRACP

Staff Physician, Clinical and Metabolic Genetics,
Associate Scientist, Developmental & Stem Cell Biology,
SickKids Research Institute;
Associate Professor, Pediatrics,
University of Toronto

ERIC CAMPOS, PhD

Scientist,
Genetics & Genome Biology,
SickKids Research Institute;
Assistant Professor,
Department of Molecular Genetics,
University of Toronto

New hypomorphic mutations affecting the CMG DNA helicase cause Primordial Dwarfism

The evolutionarily conserved CDC45/MCM2-7/GINS (CMG) protein complex is an essential helicase that unwinds DNA at replication forks. We identified four individuals from two unrelated families presenting short stature characterized by unreported mutations in the *GINS3* gene, which encodes a subunit of the CMG helicase. Our analysis suggests that *GINS3* mutations destabilize the GINS subcomplex of the DNA helicase. This in turn affects DNA replication dynamics in mammalian and yeast models.

Panel Discussion with additional Panel Members

Aleixo Muise, MD, PhD, FRCPC Staff Gastroentorologist, Gastroentorology, Hepatology and Nutrition; Co-director, Inflammatory Bowel Disesase Centre;
Senior Scientist, Cell Biology

Rosanna Weksberg, MD, PhD, FRCPC Staff Physician, Clinical & Metabolic Genetics;
Senior Associate Scientist, Genetics & Genome Biology

SickKids RESEARCH INSTITUTE

TUESDAY, OCTOBER 22, 2019, 12 NOON TO 1 PM

PGCRL GALLERY, 2ND FLOOR

PIZZA TO FOLLOW