



## Specialized Services for Children and Youth invites you to join us for Breakfast at SSCY, our monthly learning series!

Topic: Inborn Errors of Metabolism and "OMICS First" DNA Testing
Presented by: Dr. Cheryl Greenberg MD, CM, FRCPC, FCCMG and Angela Krutish MSc, MSc,
CGC, CCGC

Inborn errors of metabolism (IEMs) are genetic conditions that affect the way the body breaks down food and turns it into energy. To diagnose these conditions, doctors usually start with metabolite testing (chemical tests in blood and urine) and then follow with DNA testing. It often takes several years to diagnose an IEM this way. But, DNA testing has improved over time and we now have access to new DNA tests that can give results in a matter of weeks. Will this change the way we diagnose IEMs in the future?

The Canadian Prairie Metabolic Network (CPMN) is a research study funded by Genome Prairie/Genome Canada. In this study, consenting patients who are thought to have an IEM will have DNA testing before metabolite testing — this is called the "OMICS First" approach. We want to know if this approach can lead to faster diagnosis, faster access to treatment, fewer tests, fewer hospital stays, the same or lower cost, and the same or better quality of care for patients in the Prairies.

Join us to learn more about the evolving world of DNA testing and how the CPMN hopes to integrate a new testing strategy for IEM patients across the Prairies!

## **Learning Objectives:**

- Define inborn errors of metabolism and list common signs and symptoms of these conditions
- Recognize different types of genetic testing (including single-gene tests, multi-gene panels, whole exome sequencing, and whole genome sequencing)
- Contrast the "OMICS First" approach with the traditional approach to diagnosing inborn errors of metabolism
- Consider the potential benefits and drawbacks of using an "OMICS First" DNA testing approach for the diagnostic work-up of a wide range of genetic conditions

When: 8:00-9:00am, Tuesday, April 12th, 2022; Via Zoom Register to receive a link to this event:

https://rccinc-ca.zoom.us/meeting/register/tZltfuuqqD4qGN3wM8YHUr6hYfhQskobDt2O

Mark your calendar for our upcoming sessions: 8:00-9:00 am, Tuesday May 10<sup>th</sup>, 2022 Presenter: Tricia Kell

Previously recorded Breakfast at SSCY sessions and links to this recording will be found on the SSCY Website landing page below:

<u>Breakfast at SSCY Learning Series | Specialized Services for Children and Youth - Services spécialisés pour enfants et jeunes</u>

Everyone is welcomed to attend!

