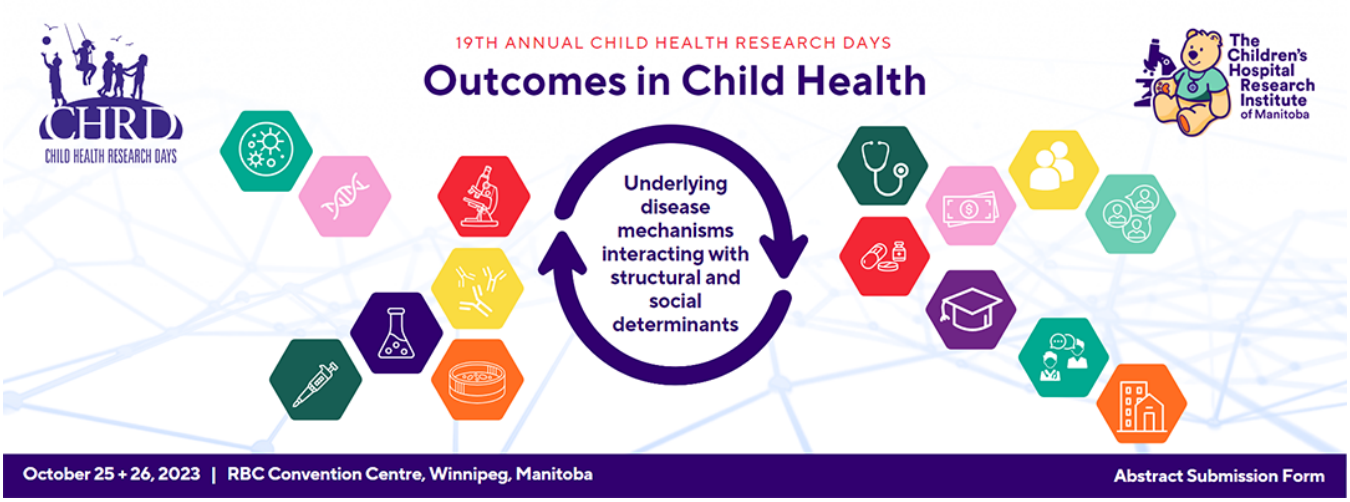


# CHRD 2023: Abstract Submission Form

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**Presenter Status**
Non-Trainee

**Research Category**
Community Health / Policy

**Role in the project**
Design
Analyze Data
Write Abstract

**Title**
Evaluating an “OMICS First” approach to the diagnosis of suspected inherited metabolic disorders: Preliminary analysis of the cost-efficacy of offering genome-wide sequencing early in the diagnostic work-up of patients referred to the clinical metabolic service

**Background**
The Canadian Prairie Metabolic Network (CPMN) provides patients suspected to have an inherited metabolic disorder (IMD) with timely access to genome wide sequencing (GWS) early in their diagnostic work-up – the “OMICS First” approach. The aims of “OMICS First” are to shorten wait-times, increase diagnostic yield, and lower clinical diagnostic costs.

**Objective**
Herein we report the preliminary economic analysis of the “OMICS First” approach using a Markov economic model.

**Methods**
Whole Exome Sequencing (WES) and/or mtDNA sequencing were performed for consenting patients. Cumulative diagnostic costs and clinical wait-times for traditional testing were estimated by reviewing patient charts from 2018-2023 and approximating costs from published sources and local costs. The cost-efficacy of the “OMICs First” approach was examined using a Markov model for participants referred to the clinical Metabolic service after 2018 (n = 71), after which clinical access to GWS became more widely available to patients and families in Manitoba. A decision tree was used to describe the effectiveness of “OMICs First” approach in terms of diagnostic costs and diagnostic yield over the 5-year period from 2018-

2023 and was compared to standard of care (SOC) costs.

**Results**
The “OMICs First” approach was found to be more cost effective than SOC, saving approximately $43,600 for each positive WES or mtDNA diagnosis per 5-year period or $8,715 for each positive diagnosis annually. The accumulated costs incurred for an “average” patient receiving “OMICs First” regardless of diagnostic outcome were $9,703 per 5-year period, whereas the accumulated costs for an “average” patient receiving SOC were $23,368. This translates to a cost-saving of $13,664 per 5-year period. Diagnostic effectiveness was 31% better for the “OMICs First” approach.

**Conclusion**
“OMICs First” WES and mtDNA sequencing improve diagnostic yield and are cost-effective for a cohort that closely resembles the population of patients commonly referred to the clinical Metabolic service.

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