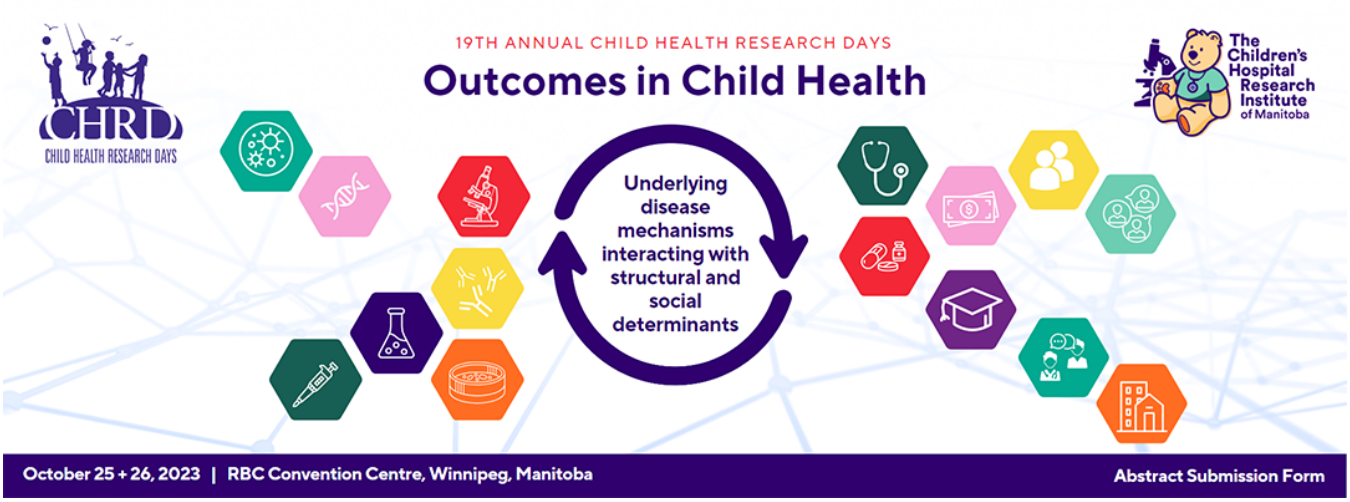

CHRD 2023: Abstract Submission Form

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Presenter Status
Non-Trainee

Research Category
Clinical

Role in the project
Design
Analyze Data
Write Abstract
Review literature

Title
A modified mainstreaming approach to increase access to genome-wide sequencing for patients with suspected inherited metabolic disorders

Background
The traditional approach to the diagnostic workup of patients with suspected inherited metabolic disorders (IMDs) involves protracted stepwise clinical, biochemical, and genetic testing. In Manitoba, this diagnostic odyssey is further hampered by wait times of up to two years for an initial metabolic evaluation. The Canadian Prairie Metabolic Network (CPMN) has offered genome-wide sequencing (GWS) by whole exome and/or mitochondrial DNA sequencing to patients with suspected IMDs who were referred by geneticists and non-geneticists early in the diagnostic workup.

Objective
Based on these findings, we will pilot an expanded mainstreaming approach to GWS.

Methods
Mainstreaming is a model in which non-geneticists integrate genetics into their practices without initial referral of patients to Genetics. This approach typically involves non-geneticists reviewing criteria for genetic testing, gathering relevant history, obtaining informed consent, facilitating genetic testing, and delivering results. In our pilot, we propose a modified approach in which a dedicated nurse specialist and genetic counsellor will support non-geneticists by taking on many of these responsibilities.

Results

To date, CPMN has received 41 referrals from non-geneticists for patients with suspected IMDs. GWS results have been received for 56.1% (23/41) with a current diagnostic yield of 17.4%. Emerging literature demonstrates that a mainstreaming model can reduce overall genetics-related healthcare costs per patient and provide timely access to genetic testing while conserving the resources of genetics services. This allows geneticists to collaborate with non-geneticists in the mainstreaming model by shifting their focus to downstream events such as complex molecular interpretation, confirmatory testing, treatment, patient education, and follow-up family testing.

Conclusion
Results to date support the goal of implementing a mainstreaming approach to GWS in Manitoba, shifting the focus of geneticists to important downstream events, simultaneously raising awareness of IMDs among non-geneticists and ensuring patients have access to the most appropriate, cost-effective, and timely genetic testing.

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