# CHRD 2024: Abstract Submission Form

**Presenter Name**
Tanvi Kaushal

**Presenter Status**
Undergraduate Students

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

**Research Category**
Clinical

**Title**
Evaluating a mainstreaming approach to whole exome sequencing for inherited metabolic disorders

**Background**
Inherited metabolic disorders (IMDs) are genetic conditions that affect the breakdown or storage of proteins, carbohydrates, and fats, and/or the creation of energy. IMDs have broad, nonspecific clinical presentations and can manifest at any age. The traditional approach for the diagnostic workup of patients with suspected IMDs involves a lengthy, stepwise process of clinical, biochemical, and genetic testing. The Canadian Prairie Metabolic Network (CPMN) offered whole exome sequencing (WES) early in the diagnostic workup of patients with suspected IMDs: the “OMICS First” approach. While CPMN demonstrated that “OMICS First” is more cost-effective than the traditional approach, patients referred by geneticists still faced extensive wait times for their initial consultation in genetics. To further reduce wait times, we propose allowing non-genetics specialists to request WES for select indications. This approach adopts a service delivery model called “mainstreaming”, wherein non-geneticists can initiate genetic testing without first referring patients to a genetics service. The Mainstreaming Genomics in Manitoba study uses a modified approach in which a genetic counsellor supports non-geneticists by obtaining informed consent, facilitating genetic testing, and helping deliver results.

**Objective**
We hypothesize that mainstreaming will reduce wait times in the diagnostic process and as such, will be accepted by patients and their health care providers. One study objective is to evaluate implementation outcomes for this modified mainstreaming approach.

**Methods**
The outcomes we deemed appropriate to assess in this early stage of implementation are acceptability, appropriateness, and feasibility. We will measure the implementation outcomes by surveying genetics professionals (to assess appropriateness and feasibility) and non-genetics specialists (to assess acceptability and feasibility).

**Results**
To date, 16 patients, referred by neurologists, rheumatologists, and immunologists, are enrolled in the study.

**Conclusion**
Measurement of these outcomes will be vital to evaluate the appropriateness of long-term implementation and success of the modified mainstreaming model for the diagnostic work-up of IMDs.

**Do you have a table/figure to upload?**
No

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