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Clinical

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

Background
Early diagnosis of inherited metabolic disorders (IMDs) is crucial for timely initiation of clinical management, which can reduce morbidity and mortality. However, diagnosis is often hampered by long wait times for genetics consultations and a lengthy, iterative diagnostic work-up including biochemical and genetic testing. The Mainstreaming Genomics in Manitoba (MGM) project implements a modified mainstreaming model whereby non-genetics specialists can request exome sequencing (ES), with support from a triage team to determine eligibility and a genetic counsellor to facilitate consent, sample collection, and result disclosure. We hypothesize that this modified mainstreaming model will be well-received by patients and caregivers.

Objective
Evaluate acceptability of the modified mainstreaming model for ES from the patient and caregiver perspective.

Methods
We assessed acceptability among MGM participants using a survey adapted from the local Program of Genetics and Metabolism. Invitations were sent to patients/caregivers one week after they received ES results, with a reminder two weeks later.

Results
Sixty patients were referred to MGM, of whom 40 consented to participate and were approved for ES. To date, 13 participants received results with a 38% diagnostic yield. Eight participants completed the survey (62% response rate), five of whom received a diagnosis. All eight respondents rated their interaction with the health care provider(s) disclosing their results as excellent. Most participants reported very good to excellent understanding of the study purpose, next steps, and implications of their results. Interactions with the research team, care received, and information provided were uniformly rated as excellent. All respondents were glad to have been referred to the study.

Conclusion
The modified mainstreaming model appears to be well-received by and acceptable to patients and caregivers, as shown by the high levels of satisfaction. Future work will include gathering more participant responses, and unique surveys for genetics and non-genetics specialists regarding acceptability, appropriateness and feasibility of the model.

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