CHRD 2024: Abstract Submission Form

Presenter Name
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Presenter Status
Undergraduate Students

Role in the project
Design
Analyze Data

Research Category
Community Health / Policy

Title
Disparities in Glutaric Aciduria Type 1 Outcomes: Collaborative Research with Anisininew Nations

Background
Glutaric aciduria type 1 (GA1) is an autosomal recessive disorder of lysine, hydroxylysine and tryptophan metabolism. The frequency is 1/400 in the Anisininew Nations due to a founder variant in GCDH. Untreated, GA1 results in severe neurodevelopmental disability. Since inclusion in newborn programs worldwide, presymptomatic treatment is successful in >90% of infants with GA1. However, the outcomes for children from the Anisininew Nations with GA1 identified on newborn screening is less favourable.

Objective
Our long-term goal is to identify the underlying reasons for this disparity, which will ultimately allow development of new treatments. Our immediate objective is to incorporate community perspectives and needs into the research framework, through collaboration with Four Arrows Regional Health Authority (FARHA), community leaders, patients and families.

Methods
We will analyze archived newborn blood spots from 19 GA1 patients from the Anisininew Nations born since 2000 and who are followed in the Shared Health Program of Genetics and Metabolism. Utilizing metabolomics, we will examine these blood spots to identify biomarkers and patterns that may elucidate why ~50% of children with GA1 from the Anisininew Nations still experience severe neurological complications despite receiving evidence-based treatment.

Results
To date, we secured ethics approval. We presented to the FARHA board and senior management for endorsement and are following a more extensive protocol proposed by FARHA. We created a webpage to keep the community informed. The next step is to seek approval from the Chiefs and Councils in the four communities.

Conclusion
Patient engagement, providing support and building trust through meaningful partnerships are vital to successful research. As such, prioritizing the perspectives and needs of patients is essential to developing more effective treatments for the infants diagnosed with GA1. Our foremost priority is to ensure that our research addresses the specific challenges faced by Anisininew families to improve the outcomes for children with GA1.

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