

Date: June 27, 2024

GENOMICS

Genetic Testing- Pediatric Diagnostic Panel (PEDx)

Date effective: Immediately

Background Information:

The Pediatric Diagnostic panel (PEDx) is a curated panel of 118 variants in 101 genes associated with a hereditary disorder presenting in the pediatric population. This testing is available to confirm a medical diagnosis in a symptomatic infant, child, or youth. This test is intended to provide first line testing prior to (or concomitant with) additional genomic investigations. Most variants in this panel are unique to specific populations in and near Manitoba such as the Hutterite, Mennonite, Ashkenazi Jewish, Inuit, and distinct First Nations communities.

This test is not intended for carrier screening or predictive testing. Furthermore, a diagnosis for a genetic disorder unrelated to the clinical presentation may be identified and reported by this analysis.

Change in Test Procedure:

- Expansion of in-house testing for variants observed within the Manitoba pediatric population.

References/Resources:

- Supplementary Table: Pediatrics Diagnostic Panel Variant List
- A LIM update is in progress
[Laboratory Information Manual- https://apps.sbgh.mb.ca/labmanual/test/findTestPrepare](https://apps.sbgh.mb.ca/labmanual/test/findTestPrepare)
- A SH HSC Molecular Diagnostic Lab Test Requisition R250-10-09 update is in progress

Patient Impact and System Improvements

- Available as first-line testing for acute pediatric cases.
- Rapid turnaround time: 2 weeks (is available for timely medical management and care)
- Non-urgent requests: 12 weeks
- Incidental diagnosis for only pediatric conditions will be reported

More Information:

- Genetic Testing and Screening in Children – Canadian Paediatric Society Position Statement Paediatric Child Health 2022 27(4):243–247

Contact Information: Dr. Ronald Agatep, Molecular Geneticist, Genomics, Diagnostic Services
ragatep@sharedhealthmb.ca