March 15, 2023

GENOMICS

Chromosome Microarray (CMA) testing

Date effective: Immediately

Background Information:

Whole genome chromosome microarray (CMA) testing is used to detect small gains or losses of chromosome material, also known as copy number variants (CNV), which are not detected by conventional cytogenetic techniques. This test is often ordered for children presenting with developmental delay, intellectual disability, autism spectrum disorder, and/or multiple congenital anomalies.

The Cytogenetics laboratory has validated the clinical use of the Illumina Infinium GDA-Cyto Assay as its in-house microarray test, allowing for repatriation of this test. For operational reasons, this repatriation will be staggered such that some samples will be tested in-house while others will continue to be sent to a referral laboratory outside of Manitoba for testing. Please continue to order this test using both the Shared Health Constitutional Cytogenetics and the GeneDx requisitions.

Change in Test Procedure:

- Postnatal peripheral blood samples received in the Cytogenetics laboratory for microarray testing will be triaged for microarray testing either in-house or at GeneDx.
- The targeted turnaround time for in-house testing is 4 weeks.
- Prenatal samples for microarray testing will continue to be sent to GeneDx.

References/Resources:

- Lab Information Manual (sbgh.mb.ca) (will be revised once repatriation is fully completed)
- R250-10-71 *Cytogenetics & FISH – Constitutional Requisition*

Patient Impact:

- Better coverage of coding regions of clinically significant genes
- Improved turnaround time

System Improvements:

- Better utilization of healthcare dollars

More Information:

Miller D.T. *et al.* Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. Am J Hum Genet. 2010 May 14;86(5):749-64. PMID: 20466091

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