

**Date:** July 29, 2019  
**To:** All Zones  
**From:** Alberta Public Laboratories (APL)  
**Re:** Change to Chromosomal Microarray (CMA) Testing Method

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### Key Message:

- Effective September 3, 2019, CMA will be performed using a single nucleotide polymorphism (SNP) array instead of a comparative genomic hybridization (CGH) array. A SNP array provides slightly increased sensitivity and will detect absence of heterozygosity (AOH) which can provide additional diagnostic information.
- Chromosomal microarray (CMA), detects chromosome copy number variants (CNVs) (i.e. missing or extra genetic information) that are smaller than can be detected by karyotyping and can more precisely characterize chromosome imbalances.
- For most patients previously tested using a CGH array, repeat CMA with the SNP array is not necessary, unless there is consanguinity. Other genetic investigations may be more appropriate and referral to Medical Genetics may be considered.

### Why this is important:

- CMA can be ordered by any relevant medical specialist for the approved Indications, including:
  - dysmorphic features
  - autism or intellectual disability
  - multiple congenital anomalies or isolated congenital heart defect/cleft lip and/or palate
  - familial or de novo CNV
  - unexplained stillbirth or neonatal death ( > 20 weeks gestational age)
  - third and/or subsequent miscarriage (*tissue or products of conception*)
  - nuchal translucency  $\geq 3.5$  mm, intrauterine growth retardation or structural ultrasound abnormality (*prenatal*)
- Referring health care practitioners should be aware of the changes to CMA reports which will include both copy-number information and, where relevant, information on regions with absence of heterozygosity.

### Action Required:

- CMA information and the *Chromosomal Microarray Requisition* are found on the Genetics and Genomics webpage: <https://www.albertahealthservices.ca/lab/page8667.aspx>

### Inquiries and feedback may be directed to:

- APL Genetic Counsellors in Calgary 403-955-3097 and Edmonton 780-407-1015

### This bulletin has been reviewed and approved by:

- Dr. Judy Chernos and Dr. Sherry Taylor, Interim Co-Medical/Scientific Directors, Genetics & Genomics
- Dr. Carolyn O'Hara, Interim Chief Medical Laboratory Officer, Alberta Public Laboratories