

DATE:	20 January 2025
TO:	All Zones
FROM:	Cytogenetics North and South, Genetics and Genomics, APL
RE:	Targeted Chromosomal Microarray for Copy Number Variant Analysis

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Key Message

- Familial segregation analysis of copy number variants (CNV) identified by chromosome microarray (CMA) will transition from a qPCR-based analysis to CMA limited to the chromosomal region(s) of interest.

Background

- Copy number variation analysis is used for parental follow-up of a CMA result. Based on the characteristics of the CNV, follow-up testing may be by FISH and/or by molecular analysis. Molecular testing was previously limited to qPCR and MLPA techniques.
- Targeted CMA testing, which will be limited to the chromosomal region(s) of interest, is now the preferred methodology for CNV follow-up familial testing. Alternate methods including FISH will be recommended in the proband's report when appropriate. This methodology change will streamline workflow and reduce follow-up costs.
- Parents will be tested sequentially. If testing of the first parent identifies the same CNV identified in the child, testing for the second parent is not required and will be cancelled if a specimen is received. If testing of the first parent is negative, testing will proceed on the second parent. An amended proband report will be issued when all appropriate parental testing is complete.

How this will impact you

- CNV analysis will continue to be ordered using the copy number variant analysis order (LAB4149) in Connect Care or utilizing a Molecular Genetics Laboratory General Requisition. While targeted CMA analysis will be the preferred testing methodology, qPCR and/or MLPA may be utilized under certain and rare circumstances. Refer to the methodology section of the patient's Copy Number Variation Analysis report to determine which methodology was used.

Action Required

- No change in practice required.

Effective Immediately

Questions/Concerns

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Approved by

- Dr. Dennis Bulman, Medical/Scientific Director, Provincial Genetics and Genomics Program, APL
- Dr. Carolyn O'Hara, Chief Medical Laboratory Officer (Interim), APL