

DATE:	2 February 2026
TO:	All Zones
FROM:	Molecular Genetics Laboratory North & South, Genetics & Genomics, APL
RE:	Exome Analysis Guideline Update

PLEASE POST OR DISTRIBUTE AS WIDELY AS APPROPRIATE

Key Message

As of January 20, 2026, the [Exome Analysis Guideline](#) has been updated in the following sections.

- Section 1.3 has been revised to provide further clarification of the ordering criteria. When a genetic etiology is the most likely explanation for a patient's phenotype, routine exome analysis can now be ordered if one or more of the following criteria are met. The patient's clinical presentation includes:
 - Moderate, severe or profound intellectual disability in the absence of known risk factors
 - Global developmental delay (age 0 to 5 years)
 - Moderate-severe autism or mild intellectual disability **AND** clinical features suggestive of a genetic syndrome
 - Suspected ataxia onset between ages 0 and 5 years
 - Two or more congenital anomalies suggestive of a genetic condition
 - Suspected genetic condition with at least one of the following: multisystem involvement, progressive clinical course and/or severe developmental or functional impairment
 - A broad differential diagnosis which includes two or more well-defined conditions requiring evaluation by multiple targeted gene panels
 - Multiple congenital anomalies in a fetus OR a single complex fetal anomaly that is considered highly genetically heterogenous
 - Comprehensive genetic testing is required to determine transplant eligibility (age 0 to 5 years)
- Additional testing scenarios have been added to Section 3:
 - for patients who have been previously tested at an external lab
 - when exome reanalysis and resequencing are indicated
 - when quad exome analysis is indicated

Background

- Exome analysis has been available through APL since June 16, 2025. Over the last year, the Exome Analysis Guideline has been revised based on current evidence and is in alignment with other Canadian institutions offering exome analysis.
- Additional testing scenarios have been added to the document to direct the approach for complex testing scenarios.
- For more information about eligibility criteria, see the [Exome Analysis Guideline](#).



How this will impact you

- Requests for routine clinical exome analysis will be accepted if the updated eligibility criteria are met. The test name, turnaround time and acceptable sample types are unchanged.

Action Required

- Refer to the Test Directory for the test description, acceptable specimen types, and restrictions.
- Refer to the [Exome Analysis Guideline](#) for eligibility criteria and testing scenarios to guide testing approach for complex cases.

Effective Immediately

Questions/Concerns

- G&G North MGL Genetic Counsellors at 780-407-1015
- G&G South MGL Genetic Counsellors at 403-955-3097

Approved by

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