ALBERTA PRECISION LABORATORIES
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Laboratory Bulletin

DATE:	26 May 2025
TO:	All Zones
FROM:	Molecular Genetics Laboratory North & South, Genetics and Genomics, APL
RE:	**Exome Analysis – Launch of APL Testing**

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Key Message

- As of June 16, 2025, routine exome analysis will be available to order through the Molecular Genetics Laboratory North & South within Alberta Precision Laboratories (APL). Exome analysis by next generation sequencing on a genome backbone allows for identification of disease-causing variants associated with hereditary conditions reflective of the patient's phenotype. Secondary analysis designed to identify conditions requiring medical intervention/management will also be available.

Background

- Exome analysis is indicated for patients with a complex phenotype that is most likely explained by a genetic etiology in which the differential diagnosis includes multiple well-defined conditions. Testing has historically been sent to external laboratories through the Genetic Resource Centre (GRC). After completion of the Translational Implementation of Genomics for Rare Disease (TIGeR) project, exome analysis is now available through APL. For more information about eligibility criteria, see the Whole Exome Sequencing Guidelines.

How this will impact you

- Routine whole exome sequencing (WES) should be ordered through APL instead of coordinating a send-out through the GRC. See below for exceptions. Genetic counselling and consenting must also be performed as per clinic processes.
- Scenarios when WES should be ordered through an external laboratory:
  - RUSH testing (i.e. results are needed for patient management within 12 weeks).
  - Prenatal testing
  - Expand to WES (i.e. order WES through the lab that performed the original panel)
  - WES re-analysis (i.e. when WES was performed through an external lab)
- Please note that **saliva is not an accepted specimen type for in-province WES**. In exceptional circumstances when an acceptable specimen type is not available, please contact the lab.

Action Required

- Refer to the test directory for the test description, acceptable specimen types, and restrictions.
- Refer to Whole Exome Sequencing Guidelines for eligibility criteria and reanalysis timelines.
- Testing can be requested in EPIC-ConnectCare or via paper requisition. Note parental genetic information aids in analysis, therefore, collection and sequencing of patient and parental samples is recommended (when available).

  - In ConnectCare testing for both the proband and family members is ordered using the "Exome Analysis" order then selecting which testing is required - proband or family member. Note: separate orders need to be placed for the proband and any associated family members.

- Phenotypic information is required in the form of a clinical description and Human Phenotype Ontology (HPO) terms. Information can be provided at the time of test ordering, in Phenotips within 30 days of ordering, or on the paper requisition.
- Testing will not be initiated until specimens are available on the proband and any family members that will be included in the analysis, and HPO terms have been provided for all individuals.
- Familial testing for identified pathogenic and likely pathogenic variants detected via in-house WES can be requested using the "Familial Variant – WES Follow-up" order. Reanalysis must be requested using the Whole Exome Sequencing (WES) paper requisition.

Effective **June 16, 2025**

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. Lauren MacNeil, Program Site Lead - North, Provincial Genetics and Genomics Program, APL
- Dr. Jillian Parboosingh, Program Site Lead - South, Provincial Genetics and Genomics Program, APL
- Dr. Carolyn O'Hara, Chief Medical Laboratory Officer (Interim), APL