



Who does this document apply to?

Information in this document applies to testing performed in the Molecular Genetics Laboratory North.

When should germline testing for a somatic variant be considered?

- The somatic variant is classified as likely pathogenic or pathogenic (Tier I/II).
AND
- The somatic variant allele fraction (VAF) is > 35%.

What sample should be collected for germline testing?

Tumour type	Ideal sample type	Acceptable sample type
Hematological	Fibroblast culture*	Buccal
Solid	Blood	N/A

*Please see “Considerations for Skin Biopsies: Information for Ordering Providers” document for instructions to arrange for a skin biopsy collection and fibroblast culture preparation.

How are buccal samples collected?

Laboratory approved buccal collection kits must be used to collect samples. You may contact the Edmonton Molecular Genetics Laboratory (780-407-1434) to arrange to have a kit delivered to your unit at the University Hospital or mailed to your location outside UAH.

What else should I consider?

If a relative(s) is being considered as a bone marrow donor, a blood sample from the potential donor(s) should be sent at the time of testing the proband. If a germline variant is confirmed, individuals related by blood may have the variant which may impact their suitability to be a donor. This will allow concurrent testing of samples and decrease the overall turnaround time.

How to order germline genetic testing to follow up on a somatic variant?

If the provider is on Connect Care:

Place an Inherited Cancer and Endocrine Gene Panels, Specific Variant (LAB3335) order.

If the provider is not on Connect Care:

Use the Molecular Genetics Laboratory Cancer and Endocrine NGS Requisition (<https://www.albertahealthservices.ca/frm-20897.pdf>).

Please indicate if a transplant is planned. The laboratory may contact the ordering provider to inquire if potential donors are related by blood to recommend concurrent testing.

What happens next?

- The laboratory will review the variant classification and VAF to ensure testing is indicated.
- If testing is indicated the lab will confirm that the sample type provided is acceptable for testing.
- If testing is not indicated or additional information is required, the ordering provider will be contacted.

When can I expect results?

Routine: 6 to 8 weeks, Stat: 1 to 2 weeks

How are results reported?

Results are sent to the ordering provider and available in Netcare and Connect Care.

Contact Information

Laboratory Genetic Counsellors, Genetics & Genomics North Sector (Edmonton) at 780-407-1015.