



Molecular analysis may be performed on DNA extracted from cultured skin biopsies (fibroblast culture). Such testing may be indicated for patients who have had a bone marrow transplant, patients with hematological malignancies and when testing an alternative tissue type is necessary.

For Patients Seen in Edmonton

1. Complete the relevant Molecular Genetics Laboratory (MGL) requisition. MGL North (Rm 8-26 MSB) extracts DNA from culture and performs germline testing:
 - Mark the specimen type “Other: skin biopsy”
 - Write “culture done in Mol Path, cells to come to MGL for extraction” on the first page.
 - If targeted variant testing is being ordered to confirm a somatic cancer variant in germline DNA, complete a Cancer and Endocrine NGS Requisition (<https://www.albertahealthservices.ca/frm-20897.pdf>). Under “Section V – Specific Variant Testing”, provide the gene, transcript, HGVS variant description and variant allele frequency. Indicate that germline testing is being requested for a somatic variant.
 - Make a copy of the completed requisition to go with the specimen and give the original to MGL. (This step will ensure MGL accessioning will be expecting the culture from Mol Path when ready).
2. Arrange for collection of biopsy:
 - Call the Senior Technologist in Mol Path (7-2895) prior to biopsy (and ask for aliquoted MEM media, if needed; someone will need to go pick it up at WMC 4B4.30).
 - Enter order in Connect Care for Fibroblast culture (LAB4017) and use Clinic Collect to complete the order once the sample is collected.
 - Once biopsy is collected in media, label sample and deliver the sample to Mol Path (WMC 4B4.30) with a copy of the requisition.
3. Culturing is generally expected to take 3-4 weeks, and in-house testing typically takes 8 weeks.

When testing multiple family members, please send samples from all family members at the same time. This information sheet does not cover the investigation of cytogenetic mosaicism.

For Patients Seen in Calgary

1. Transport media can be obtained frozen from the Alberta Children’s Hospital (ACH) Cytogenetics Lab and must be thawed and warmed to ambient temperature for 30 minutes prior to use. Do NOT put tissue in formalin. To coordinate pick up of transport media from ACH in advance of your patient’s biopsy, contact the Genetics and Genomics South Genetic Counsellors (403-955-3097).
2. Place a Connect Care order for Cytogenetic Analysis:
 - Select type of cytogenetic testing: Constitutional/Inherited condition
 - Select test: Culture for alternative testingIf you do not have access to Connect Care, complete a Cytogenetic Analysis, Constitutional Requisition (<https://www.albertahealthservices.ca/frm-21890.pdf>), and select “Culture for Alternative Testing” under the test requested section.
3. Complete the relevant Molecular Genetics Laboratory or Genetic Resource Centre requisition for the molecular test requested:
 - If targeted variant testing is being ordered to confirm a somatic cancer variant in germline DNA, complete a Cancer and Endocrine NGS Requisition (<https://www.albertahealthservices.ca/frm-20897.pdf>). Under “Section V – Specific Variant Testing”, provide the gene, transcript, HGVS variant description, and indicate that germline testing is being requested for a somatic variant.
 - If out-of-province testing is being coordinated through the Genetic Resource Centre, complete a Genetic Resource Centre Requisition (<https://www.albertahealthservices.ca/frm-21887.pdf>).

Send the completed requisition to the ACH Cytogenetics Lab by email or by fax (E: aplggcytoaccessioningsouth@albertahealthservices.ca, F: 403-955-3000). This requisition does NOT need to accompany the fibroblast sample.



4. Transport the fibroblast specimen at room temperature to the ACH Cytogenetics Lab, B0-150. Sample transport can be facilitated through an APL collection site. For additional information about turnaround times and sample requirements, please refer to the APL Test Directory page “Fibroblast Culture - Cytogenetics South”.

Contact Information

Location

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