



Clinical Genomics Laboratory

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For additional up-to-date testing information and our most current requisitions,
please visit our website: <http://www.iwk.nshealth.ca/clinical-genomics/>

GENERAL MOLECULAR GENETICS REQUISITION

Order as: *Molecular IWK*

Sample Requirements for Molecular Genetic Testing

Peripheral blood: Collect 3mL in lavender EDTA (newborns <1 month: 1mL). Do not centrifuge or freeze. Ship at room temperature within 72 hours. (Note: if patient has received blood product containing leukocytes or non-irradiated red blood cells in the last 14 days, contact the laboratory before collection as the sample may not be suitable for all testing. Transfusions of irradiated packed red blood cells, plasma, or platelets are accepted as these are not expected to affect this genetic testing.)

Postmortem blood: Collect 3mL in lavender EDTA. Send at room temperature within 72 hours. Alternatively, freeze at -80°C upon collection and send on dry ice.

Skin Biopsy: Collect 3mm³ skin punch into specimen container containing sterile media or saline, taken using aseptic technique. Sample must arrive to the IWK within 24 hours of collection, during regular business hours. To collect: clean skin surface 3 times with 70% isopropyl alcohol by either pouring over the skin surface, or using sterile cotton pads saturated by dipping in alcohol from a sterile container. Allow skin to air dry between applications. Note: Never use betadine as it can inhibit or prevent cell growth. For numbing the area, use 2% lidocaine or 2% lidocaine w/epinephrine 1:100,000 using a small gauge (22G) needle just under the skin to create a bleb. Do not freeze- send at 4°C.

Tissue (surgical/postmortem): Do not place in formalin. If dry ice is available, freeze at -80°C upon collection (with no added saline) and send on dry ice. If dry ice is not available, do not freeze- collect the sample in sterile saline and send at 4°C within 24 hours. NOTE: if cytogenetic cell culture/karyotype is required, do not freeze- order using *CYTOGENETICS CONSTITUTIONAL KARYOTYPE* requisition. For Fetal Tissue (products of conception/fetal demise) please use the *FETAL GENETIC TESTING* requisition.

Cord Blood: NOTE - Please follow all instructions to avoid specimen rejection:

1. Label both specimen and requisition with **neonatal** demographics, including: infant name (or "Baby of *MATERNAL LAST NAME, MATERNAL FIRST NAME*"), infant's date of birth, and infant's HCN (NOTE: if infant's HCN is unavailable, use maternal HCN but clearly indicate "MOM" immediately beside HCN.)
2. Collect 3mL cord blood sample from cord using a syringe, maintaining clean technique to avoid maternal contamination of the specimen. Immediately transfer to labeled lavender EDTA tube.
3. Required: **handwrite** or affix a sticker on the specimen tube indicating "CORD BLOOD".
4. Ship to the laboratory at room temperature within 24 hours. Do not centrifuge or freeze specimen.
5. Note: a maternal peripheral blood specimen to rule out maternal cell contamination is also required- along with a separate *GENERAL MOLECULAR GENETICS* requisition.

Collection information for patients: Peripheral blood can be collected at any blood collection facility convenient for you; fasting is not required.

Informed Consent for Molecular Genetic Testing

Information to be discussed with patient by Healthcare Provider (note: consent for pathology specimens is included under the autopsy consent). Additional information regarding indications for testing and test limitations can be found at iwkhealth.ca/CGL/TestMenu. Turnaround times are available at iwkhealth.ca/CGL/TAT.

General Information about Genetic Testing

1. This test cannot detect every genetic abnormality. Therefore, a normal test result does not rule out all possible genetic conditions.
2. Correlation with clinical information may be required for accurate interpretation. Correct interpretation of results depend on accurate clinical findings, family relationships and other laboratory data provided.
3. This test might reveal:
 - a. Variants of uncertain significance (VUS). These variants may or may not be related to the patient's phenotype or disease.
 - b. Unexpected information about family relationships (e.g. non-paternity, consanguinity).
4. Complete interpretation of test results may require additional follow-up testing on other family members.
5. Test results are confidential, but may be used without identifying information for interpretation of testing for family members.
6. When available, genetic testing will be performed at the IWK Clinical Genomics Laboratory (CGL). When testing cannot be performed at CGL, testing may be referred out to an external laboratory.
7. Results from testing may be submitted to clinical databases anonymously as needed (with all identifying information removed). These clinical databases are used by the laboratory in order to assist in accurate interpretation of results.
8. Genetic counselling through Maritime Medical Genetics Service (MMGS) is available upon request: fax a referral to their department at 902-470-8709 or phone 902-470-8754 to request the appropriate forms.

DNA STORAGE: For more information, refer to our policy at refer to our policy at <https://iwkhealth.ca/CGL/SampleStorage>

Temporary Retention of Residual Samples: 5 years

- Following completion of testing, or when *Extract & Hold* is indicated, DNA will be stored for a minimum of 5 years. (Original specimens are not retained; excluding Medical Examiner tissues).
- Any additional testing of the sample will require a written request from a health care provider including a signed statement that the patient has been consented appropriately for the testing.
- When testing is complete, the laboratory may anonymize and use some of the residual sample or genetic data to improve and develop new testing. Unexpected genetic findings unrelated to the testing indication will not be reported.

Irreplaceable Storage: 25 years

- Long-term DNA storage is available upon request only when testing likely to follow beyond 5 years **AND** sample cannot be recollected.

Sample Disposal/Test Cancellation

- Sample disposal and test cancellation is available. Separate form is required: see *TEST CANCELLATION/SAMPLE DISPOSAL REQUEST FORM*