Clinical Genomics Laboratory
5850/5980 University Ave, PO Box 9700
Halifax, NS B3K 6R8
Phone: (902) 470-6504 Fax: (902) 470-7466
Email: clinicalgenomics@iwk.nshealth.ca

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 TESTING

Order as: Molecular IWK

Patient Information

Name *(LAST, FIRST MIDDLE)* :

DOB *(dd/mmm/yyyy)* :

Health Card #: Province of Residence:

MRN #:

Accession #:

Phenotips ID (MMGS only):

Sex Assigned at Birth: Legal Gender:

Sample Information

- ☐ Peripheral blood — Lavender **EDTA** 3mL (newborns <1 month: 1mL)
- ☐ Cord blood – see reverse for collection. A **maternal EDTA blood sample** is also required
- ☐ Postmortem blood – Lavender EDTA 3mL ☐ Tissue - Specify: _____
- ☐ Skin biopsy (culture, then extract DNA) ☐ DNA #: _____
- ☐ Skin biopsy (direct DNA extraction) ☐ Cultured cells: _____

Collection Date/Time:

Collection Facility: **Collector Initials:**

Indicate if the Patient has:

- ☐ Had an allogeneic bone marrow transplant
- ☐ A current hematological neoplasm
- ☐ Received blood products containing leukocytes/non-irradiated RBCs in ≤14 days

Indication and Reason for Testing

Request for Expedited Result:

- ☐ Results will impact a pregnancy management decision: indicate EDC _____
- ☐ Medical intervention: specify, include date _____
- ☐ Other: specify _____

Testing Category: Required *(except for DNA storage/MCC requests)*

- ☐ Diagnostic testing *(symptoms of disease/disorder)* — **describe below**
- ☐ Predictive testing *(documented family history)* — by Medical Genetics only
- ☐ Carrier testing
- ☐ Other: specify _____

Clinical Information and Family History

Relevant clinical findings, if symptomatic:

Pedigree/Family History:

Consanguinity: specify _____

Ethnic Background: ☐ Acadian ☐ Ashkenazi Jewish ☐ Indigenous ☐ French Canadian ☐ Hispanic ☐ African ☐ European Caucasian ☐ Asian ☐ Middle Eastern ☐ Other

Please see reverse page for collection information and shipping instructions

Ordering Health Care Provider Information

Name:

Office/Institution:

Phone #: Fax # (**Required**):

Email:

Confirmation of Informed Consent: I (or my designate) have explained the risks, benefits, and limits of the tests requested, and have answered the patient's questions. In my opinion, the patient understands and has given informed consent for this testing.

Signature **(Required)**: _____ _____
Health Care Provider Date signed *(dd/mmm/yy)*

Additional Copies to:

Health care provider:

Facility:

Phone #: Fax #:

Testing Requested

Neuromuscular Disorders:

- ☐ ALS *(C9orf72 only)*
- ☐ Becker/Duchenne Muscular Dystrophy
- ☐ PMP22 dosage- CMT1A
- ☐ PMP22 dosage- HNPP
- ☐ Huntington Disease *(confirmation only)*
- ☐ Myotonic Dystrophy Type 1
- ☐ Spinal Muscular Atrophy (SMN dosage)

Targeted Aneuploidy (QFPCR):

- ☐ Trisomy 21
- ☐ Trisomy 18
- ☐ Trisomy 13
- ☐ Klinefelter Syndrome
- ☐ Atypical genitalia

Microdeletion/Duplication & Imprinting:

- ☐ 22q11 Deletion/Duplication
- ☐ Angelman Syndrome
- ☐ Beckwith Wiedemann Syndrome
- ☐ Prader Willi Syndrome
- ☐ Russell Silver Syndrome
- ☐ Y chromosome microdeletion

Cystic Fibrosis:

- ☐ Infertility (CBAVD)
- ☐ CFTR sequencing

FMR1-Related Tests:

- ☐ Fragile X Syndrome
- ☐ Fragile X Tremor-Associated Syndrome
- ☐ Premature Ovarian Failure

Sequencing* — complete clinical phenotype and family history/pedigree must be provided

- ☐ Panel: _____
- ☐ Gene(s): _____

Refer to **iwkhealth.ca/CGL/TestMenu for available genes, panels, and testing restrictions*

Known Variant Testing: Include proband report when possible

- ☐ Copy number variant (CNV) ☐ Sequence variant

(HGVS/ISCN): _____

Required: Proband Name/DOB: _____
HCN/IWK DNA#: _____ Relationship to patient: _____

Refer Out: *Available testing at **iwkhealth.ca/CGL/TestMenu***

- ☐ Refer to: _____

Portal ID: _____ Test name: _____

Other:

- ☐ MCC — maternal contamination studies
- ☐ Other: _____

DNA Storage:

- ☐ **Extract & Hold (5 year retention):** Testing likely to follow within 5 years
- ☐ **Irreplaceable Storage (25 year retention):** Testing likely to follow beyond 5 years ***AND*** sample cannot be recollected

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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 3mm^3 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/karotype 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*