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/**

CONSTITUTIONAL MOLECULAR GENETIC TESTING

Hours of Operation: Monday to Friday: 0800-1700. After hours call: 902-470-8289

Patient Information

Name *(LAST, FIRST MIDDLE)* :

DOB *(dd/mmm/yyyy)* :

Health Card #: Province of Residence:

Hospital #:

Site Visit # (IWK patients only):

Sex Assigned at Birth: Legal Gender:

Indicate, if Applicable:

- ☐ Patient has had an allogeneic bone marrow transplant
- ☐ Patient has a current hematological neoplasm
- ☐ Patient has received a blood product containing leukocytes or non-irradiated RBCs in the last 14 days

Request for Expedited Result:

- ☐ Patient or partner is currently pregnant: indicate EDC_____
- ☐ Medical intervention: specify, include date _____
- ☐ Other: specify _____

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*

Copies to Additional Health Care Providers

Genetic Counsellor: Phone #:

Email: Fax #:

Health care provider:

Facility:

Phone #:: Fax #:

Indication and Reason for Testing

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

- ☐ Confirm a clinical diagnosis (patient is clinically affected) - **describe below**
- ☐ Rule out a clinical condition (some symptoms, but no clinical diagnosis) - **describe below**
- ☐ Predictive testing (documented family history) - by Medical Genetics only
- ☐ Carrier testing: Family History - **provide pedigree below**
- ☐ Carrier testing: Ethnic risk - **Indicate relevant ethnicity below**
- ☐ Other:_____

Relevant clinical findings, if symptomatic:

_____

_____

Pedigree/Additional Information:

Consanguinity: specify _____

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

Sample Type

- ☐ Peripheral blood - Lavender EDTA 5mL (newborns <1 month: 2mL) IWK meditech DNAM
- ☐ Post-mortem blood - Lavender EDTA 5mL
- ☐ Cord blood - see reverse for collection instructions. A maternal EDTA blood sample is also required
- ☐ Tissue - specify source: _____ ☐ Banked DNA#: _____
- ☐ Skin biopsy (for direct DNA extraction) ☐ Cultured cells: _____
- ☐ Skin biopsy (culture & extract DNA from cultured cells)

Collection Date/Time:

Collection Facility: Collector Initials:

Please see reverse page for collection information and shipping instructions

Testing Requested

Neuromuscular Disorders:

- ☐ ALS Screen (*C9orf72* and *SOD1*)
- ☐ Becker/Duchenne Muscular Dystrophy
- ☐ CMT1A/HNPP (*PMP22* dosage)
- ☐ DRPLA
- ☐ Friedreich Ataxia
- ☐ Huntington Disease (confirmation only)
- ☐ Myotonic Dystrophy Type 1
- ☐ Myotonic Dystrophy Type 2
- ☐ Oculopharyngeal Muscular Dystrophy
- ☐ Spinal and Bulbular Muscular Atrophy
- ☐ Spinal Muscular Atrophy (SMN dosage)

Male Infertility:

- ☐ *CFTR* sequencing (CBAVD)
- ☐ Y chromosome microdeletion

Targeted Gene Sequencing:

- ☐ Cystic Fibrosis (*CFTR*)
- ☐ Fabry Disease *(GLA)*
- ☐ Hereditary Transthyretin Amyloidosis *(TTR)*
- ☐ Non-Syndromic Deafness (*GJB2/6*)

FMR1-Related Tests:

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

Microdeletion/Duplication and Imprinting disorders:

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

Targeted Aneuploidy (QFPCR)

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

Other:

- ☐ MCC- maternal cell contamination
- ☐ Other: _____
- ☐ Refer out to:_____
  Portal ID: _____
  Test name: _____

☐ **Sequencing -** complete clinical phenotype and family history/ pedigree must be provided
Gene or Panel*: _____
*refer to website for available genes, panels, and testing restrictions

Known Variant Testing: Include proband report when possible

☐ Copy number variant (CNV) ☐ Sequence variant
(HGVS/ISCN Nomenclature):_____

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

DNA Storage:

- ☐ DNA banking - Irreplaceable sample
- ☐ Test request to follow

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GENERAL CONSTITUTIONAL MOLECULAR GENETIC TESTING

Sample Requirements for Molecular Genetic Testing

Peripheral blood: Collect 5mL in lavender EDTA (newborns <1 month: 2mL). *IWK Meditech: DNAM.* 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.)*

Post-mortem blood: Collect 5mL 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 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 the 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.

Tissue (surgical/post-mortem): 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 ("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. *IWK Meditech: order under infant- pneumonic DNAM.*
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 CONSTITUTIONAL MOLECULAR GENETIC TESTING* requisition.

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, test procedures, test limitations and turnaround times can be found on our website at **https://www.iwk.nshealth.ca/clinical-genomics**.

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

Temporary Retention of Residual Samples

- Following completion of testing, or when *Test Request to Follow* is indicated, DNA samples will be stored for a minimum of 5 years.
- 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.

DNA Banking

- Long-term DNA banking is available upon request for ongoing complex diagnostic analyses, future investigations, or future testing of other family members where extended storage of genetic material for potential future use in clinical molecular diagnostic testing. Banked samples will be retained for a minimum of 25 years.
- For more information, please refer to our DNA banking policy on our website at https://www.iwk.nshealth.ca/clinical-genomics