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/**

POSTNATAL MICROARRAY 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)
- ☐ Postmortem blood — Lavender EDTA 3mL
- ☐ DNA#: _____ ☐ Cultured cells: _____
- ☐ Tissue — specify source: _____
- ☐ Cord blood - see reverse for collection instructions. A maternal EDTA blood sample is also required.

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: specify EDC vjhvj
- ☐ Medical intervention: specify, include date vhjvj
- ☐ Other: specify _____

Testing Category: Required

- ☐ Developmental Delay/Intellectual Disability
- ☐ Autism Spectrum Disorder
- ☐ Multiple Congenital Anomalies
- ☐ Other: _____

Additional Molecular Test Requests:

- ☐ Fragile X* ☐ Angelman Syndrome
- ☐ Prader-Willi Syndrome ☐ Rett Syndrome (*MECP2*)
- ☐ Other: _____

*refer to iwkhealth.ca/CGL/TestMenu for Fragile X testing restrictions

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

Pedigree/Additional Information:

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 #:

Clinical Phenotype: check all that apply

DEVELOPMENTAL
- ☐ Fine motor delay
- ☐ Gross motor delay
- ☐ Speech delay
- ☐ Other:_____

GROWTH
- ☐ Failure to thrive
- ☐ Overgrowth
- ☐ Short stature
- ☐ Other:_____

ENDOCRINE/METABOLIC
- ☐ Hypocalcemia
- ☐ Hypercalcemia
- ☐ Hypogonadism
- ☐ Hypothyrodism
- ☐ Other:_____

GASTROINTESTINAL
- ☐ Gastroschisis
- ☐ Hirschprung disease
- ☐ Omphalocele
- ☐ Pyloric stenosis
- ☐ Tracheoesophageal fistula
- ☐ Other:_____

MUSCULOSKELETAL
- ☐ Contractures
- ☐ Club foot
- ☐ Diaphragmatic hernia
- ☐ Limb anomaly
- ☐ Polydactyly
- ☐ Scoliosis
- ☐ Syndactyly
- ☐ Vertebral anomaly
- ☐ Other:_____

NEUROLOGICAL
- ☐ Ataxia
- ☐ Dystonia
- ☐ Chorea
- ☐ Hypotonia
- ☐ Neural tube defect
- ☐ Seizures
- ☐ Spasticity
- ☐ Structural brain anomaly
- ☐ Cerebral palsy
- ☐ Other:_____

CRANIOFACIAL
- ☐ Cleft lip
- ☐ Cleft palate
- ☐ Coloboma of eye
- ☐ Craniosynostosis
- ☐ Dysmorphic facial features
- ☐ Ear malformations
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Other:_____

PERINATAL HISTORY
- ☐ Prematurity
- ☐ Intra-uterine growth retardation
- ☐ Oligohydramnios
- ☐ Polydramnios
- ☐ Other:_____

CUTANEOUS
- ☐ Hyperpigmentation
- ☐ Hypopigmentation
- ☐ Other:_____

COGNITIVE
- ☐ Learning disability
- ☐ Intellectual disability
- ☐ Other:_____

BEHAVIOURAL
- ☐ Autistic features
- ☐ Autism spectrum disorder
- ☐ Obsessive-compulsive disorder
- ☐ Other:_____

GENITOURINARY:
- ☐ Atypical genitalia
- ☐ Hydronephrosis
- ☐ Hypospadias
- ☐ Cryptorchidism
- ☐ Kidney malformation
- ☐ Ureteral obstruction
- ☐ Other:_____

CARDIAC
- ☐ Atrial septal defect
- ☐ Atrioventricular canal defect
- ☐ Coarctation of the aorta
- ☐ Hypoplastic left heart
- ☐ Tetralogy of Fallot
- ☐ Ventricular septal defect
- ☐ Other:_____

FAMILY HISTORY
- ☐ Parents with ≥ 3 miscarriages
- ☐ Other relative with similar clinical history (describe):

_____

_____

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POSTNATAL MICROARRAY TESTING

Order as: Molecular IWK

Sample Requirements for Microarray 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.

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 (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.

Postnatal Microarray

- Microarray testing looks broadly at an individual's genetic information for areas of copy number variation (deletions/duplications) and absence of heterozygosity.
- Microarray is available only for patients who meet eligible indications. For more information, visit **iwkhealth.ca/CGL/PostnatalMicroarray**. If clinical criteria are not met, the request may be cancelled by the laboratory. Eligible indications include:
  - Developmental Delay/Intellectual Disability
  - Autism Spectrum Disorder
  - Multiple Congenital Anomalies

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*