Clinical Genomics Laboratory
5850/5980 University Ave, PO Box 9700
Halifax, NS B3K 6R8
Phone: (902) 470-6504
Fax: (902) 470-7466
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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/**

FETAL GENETIC TESTING

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

Patient Information

Fetus of (Name):

DOB:

Health Card #:

Sex Assigned at Birth: Province of Residence:

Hospital #:

Pregnancy Information

Required:

Fetal Anatomic: ☐ Male ☐ Female ☐ Unknown/Uncertain

EDC: *dd/mmm/yyyy*

☐ Parents with ≥ 2 miscarriages ☐ Egg donor pregnancy

☐ Parents known to be related (specify):

Expected fetal ethnic background:

☐ Acadian ☐ Ashkenazi Jewish ☐ French Canadian ☐ Hispanic ☐ African Canadian ☐ First Nations ☐ Middle Eastern ☐ Asian ☐ European Caucasian ☐ Other_____

Ordering Health Care Provider Information

Name:

Office/Institution:

Phone #: Fax #:

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.

Health Care Provider Signature **(Required):** _____

Copies to Additional Health Care Providers

Genetic Counsellor: Phone #:

Email:

Healthcare provider:

Facility:

Phone #: Fax #:

Indication for Testing

Prenatal Samples:

☐ Positive screen (MST +/- soft markers): _____

☐ High risk NIPT results: _____

☐ Abnormal ultrasound (complete *Fetal/Perinatal History* section)

☐ Other: _____

Fetal Loss/ Tissue Samples:

☐ POC/demise (IUFD/Stillbirth): gestational age _____

☐ Congenital anomalies (complete *Fetal/Perinatal History* section below)

Fetal/Perinatal History (check all that apply)

PERINATAL HISTORY
- ☐ Maternal diabetes
- ☐ Teratogenic medication
- ☐ Abnormal fetal Doppler
- ☐ Placental abruption
- ☐ Previous affected pregnancy

CRANIOFACIAL
- ☐ Cleft lip and/or palate
- ☐ Abnormal profile
- ☐ Craniosynostosis
- ☐ Microcephaly
- ☐ Brachycephaly
- ☐ Macrocephaly

GENITOURINARY:
- ☐ Ambiguous genitalia
- ☐ Hypospadias
- ☐ Kidney malformation
- ☐ Ureteral anomaly
- ☐ Megacystis
- ☐ Other:_____

GENERAL FINDINGS
- ☐ Single umbilical artery
- ☐ Fetal growth restriction
- ☐ Fetal macrosomia
- ☐ Macrocephaly
- ☐ Oligohydramnios
- ☐ NT ≥ 3.5mm
- ☐ Other:_____

INTRACRANIAL
- ☐ Ventriculomegaly
- ☐ Absent CSP
- ☐ Cerebellar anomaly
- ☐ Abnormal cisterna magna
- ☐ Agenesis corpus callosum
- ☐ Other:_____

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

GASTROINTESTINAL
- ☐ Gastroschisis
- ☐ Omphalocele
- ☐ Duodenal atresia
- ☐ Tracheoesophageal fistula
- ☐ Diaphragmatic hernia

MUSCULOSKELETAL AND CRANIAL
- ☐ Neural tube defect
- ☐ Thoracic anomaly
- ☐ Talipes/club foot
- ☐ Limb anomaly
- ☐ Polydactyly
- ☐ Acrania/anencephaly
- ☐ Contractures
- ☐ Scoliosis/vertebral
- ☐ Other:_____

Testing Requested

In-House Testing:

☐ Targeted Aneuploidy (QFPCR)

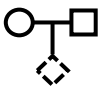
☐ Microarray (eligible indications only- complete *Fetal/Perinatal History* section)
Medical Genetics consult may be required, see reverse page

☐ CFTR Sequencing- *Only Pathogenic/Likely Pathogenic variants reported*

☐ 22q11 deletion

☐ STS gene common deletion

☐ Other (Medical Genetics): _____



Closest relative tested positive:

Name: _____

HCN: _____

IWK DNA #: _____

If not tested at IWK, attach proband report

Refer Out Testing:

☐ NIPT (by MFM or Medical Genetics referral only)

☐ Other (paperwork must accompany)- specify:

Sample Type

Fetal Sample:
- ☐ Amniotic fluid
- ☐ CVS
- ☐ Tissue- please specify source: _____
- ☐ Banked DNA#:_____

Maternal Sample:
(Required, except for Pathology specimens)
- ☐ Buccal swab included
- ☐ DNA sample previously collected
- ☐ Maternal blood, sent separately *(order using "General Requisition")*
- ☐ NIPT streck tube

Collection Date/Time:

Collection Facility: Collectors Initials:

Please see reverse page for collection information and shipping instructions

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Sample Requirements for Prenatal Genetic Testing

Amniotic Fluid: 15-30mL amniotic fluid in sterile 15mL culture tube. Do not centrifuge or freeze. Ship at room temperature within 24 hours.

CVS: 25-50mg in sterile culture media. Do not freeze. Ship at room temperature within 24 hours.

NIPT (by Medical Genetics or MFM referral only): Peripheral blood- 10ml cell free streck tube, tube must be full. Do not centrifuge or freeze. (IWK meditech: NIPT)

Fetal Tissue (POC/fetal demise): 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 72 hours. **First trimester samples:** preferred tissue type: identifiable fetal tissue from pathology examination. Otherwise, CVS sample (collected by a specialist prior to evacuation of the uterus- indicate on requisition sample is from IUFD). **Second trimester or later:** Listed in order of preference (where possible): psoas(100mg), liver(100mg), cord(1cm), placenta(100mg). *Note: If an IWK autopsy is planned, an appropriate sample will be automatically collected by the pathologist and sent to the Clinical Genomics Lab as part of the autopsy investigation.*

Maternal Buccal swab (no separate requisition required) for MCC: Collect two cytobrushes:

1. Wait 1 hour after eating or drinking before collecting. Open package and remove brush without touching any other surfaces.
2. Brush up and down the entire length of the inside of the cheek (at least a 1" path) ten times using firm light pressure. Rotate the handle while brushing to ensure all surfaces of the cytobrush come in contact with the cheek.
3. Return swab back into the package without touching the brush portion to any other surfaces. Repeat the process with a second cytobrush on the opposite cheek.
4. Label both cytobrushes with patient's identifiers, and ship with labelled fetal sample.

Maternal Peripheral blood (separate requisition required): 4mL EDTA tube, separate General Testing requisition also required.

Informed Consent for Prenatal Genetic Testing

Information to be discussed with patient by Healthcare Provider. Visit our Testing web page for details and limitations regarding prenatal testing.
*Consent for pathology specimens is included in the fetal autopsy.

TEST METHODS AND LIMITATIONS

See specific full test descriptions and limitations at **https://www.iwk.nshealth.ca/clinical-genomics** for further information regarding indications for testing, test procedures, test limitations and turnaround times.

- For all fetal genetic testing, there is a possibility of test failure due to degraded DNA and/or maternal DNA contamination, especially when testing fetal tissues in the setting of IUFD and early pregnancy loss.
- Targeted Aneuploidy Detection tests only for limited genetic conditions (21, 18, 13, X and Y). Microarray testing looks broadly at an individual's genetic information for areas of copy number variation (deletions/duplications).

GENERAL INFORMATION ABOUT GENETIC TESTING

1. Testing 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. Some tests might reveal:
   a. Variants of uncertain significance (VUS). These variants may or may not be related to the patient's phenotype or disease.
   b. Clinical information unrelated to the reason for testing. Any such results that are medically actionable will be reported to the Healthcare Provider(s).
   c. Unexpected information about family relationships (e.g. non-paternity, consanguinity).
4. Test results might 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. Genetic counseling is available if requested. This will require a referral to Maritime Medical Genetics (MMGS).
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.

FETAL MICROARRAY

- Fetal microarray is available only for eligible indications. For more information, visit **https://www.iwk.nshealth.ca/clinical-genomics**
- For microarray testing on fetal specimens, a referral to Medical Genetics is:
  - **Required** for all on-going pregnancies.
  - *Recommended* for IUFD/fetal tissue testing when anomalies have been identified, as testing beyond microarray may be indicated.
  - **Required** for IUFD/fetal tissue testing if no anomalies have been identified in the fetus, prior to consideration of microarray testing.

SAMPLE RETENTION

Following testing, the DNA sample will be stored indefinitely (excluding DNA from buccals, which will be discarded). Any additional testing of the sample will require a written request from the physician including a signed statement that the patient has been consented appropriately for the additional testing. If there is enough sample remaining when testing is complete, it may be used anonymously (identifying information will be removed) for research and quality assurance purposes.