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/**

FETAL GENETIC TESTING

Order as: Molecular IWK

Patient Information

Fetus of *(LAST NAME, FIRST MIDDLE)* :

DOB *(dd/mm/yyyy)* :

Health Card #: Province of Residence:

MRN #:

Accession #:

Phenotips ID (MMGS only):

Sex Assigned at Birth: Legal Gender:

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)*

Sample Information

Fetal Sample:

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

Maternal Sample: Required, except Pathology samples

- ☐ DNA (previously collected)
- ☐ Maternal blood, sent separately: order *MCC* on *General Molecular Genetics Requisition*
- ☐ NIPS Roche cfDNA tubes (2x10mL)

Collection Date/Time:

Collection Facility: **Collectors Initials:**

Additional Copies to:

Healthcare provider:

Facility:

Phone #: Fax:

Pregnancy Information

Required: Fetal Anatomic: ☐ Male ☐ Female ☐ Unknown/Uncertain

EDC:

- ☐ Parents with ≥ 3 miscarriages
- ☐ Egg donor pregnancy
- ☐ Parents known to be related (specify): _____

Expected fetal ethnic background: *choose all that apply*

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

Indication for Testing

Prenatal Samples:

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

Fetal/Perinatal History (check all that apply)

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

GENERAL FINDINGS
- ☐ Single umbilical artery
- ☐ FGR (noted <20weeks)
- ☐ Fetal macrosomia
- ☐ Polyhydramnios
- ☐ Oligohydramnios
- ☐ NT≥3.5mm/cystic hygroma
- ☐ Other:_____

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

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

GENITOURINARY
- ☐ Atypical genitalia
- ☐ Hypospadias
- ☐ Cystic/dysplastic kidney
- ☐ Renal malformation
- ☐ Renal agenesis
- ☐ Ureteral anomaly
- ☐ Lower urinary tract obstruct.
- ☐ Other:_____

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

GASTROINTESTINAL
- ☐ Gastroschisis
- ☐ Omphalocele
- ☐ Duodenal atresia
- ☐ Tracheoesophageal fistula
- ☐ Diaphragmatic hernia
- ☐ Other:_____

INTRACRANIAL
- ☐ Ventriculomegaly (≥13mm)
- ☐ Absent CSP
- ☐ Cerebellar anomaly
- ☐ Abnormal cisterna magna
- ☐ Agenesis corpus callosum
- ☐ 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
- ☐ Other (Medical Genetics): _____

Closest relative tested positive:

Name: _____

HCN: _____

IWK DNA #: _____

If not tested at IWK, attach proband report

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

- ☐ NIPS (by MFM or Medical Genetics referral only)
- ☐ Other (paperwork must accompany):

Refer to: _____

Portal ID: _____ Test name: _____

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. Sample must arrive to the laboratory by noon before a weekend or holiday.

NIPS (by Medical Genetics or MFM referral only): Peripheral blood- two 10ml cell free Roche tubes, tubes must be >7ml full. Do not centrifuge or freeze.

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 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): 3mL EDTA tube, separate *GENERAL MOLECULAR GENETICS* requisition also required.

Informed Consent for Fetal 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. 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. 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. 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.
9. 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).
10. Genetic counseling 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.

Fetal Microarray

- Fetal microarray is available only for eligible indications. For more information, visit **iwkhealth.ca/CGL/FetalMicroarray**
- 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.

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*