**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 CYTOGENETIC KARYOTYPE TESTING

*Order as: Cytogenetics 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 Type

- ☐ Peripheral blood — Green **NaHep** 4mL (newborns <1 month: 2mL)
- ☐ Cord blood — see reverse for collection. A maternal EDTA blood sample is also required
- ☐ Skin biopsy (3mm punch biopsy)
- ☐ Tissue, fresh surgical/postmortem: specify source _____

**Collection Date/Time:** **Collector Initials:**

**Collection Facility:**

**Indicate if the Patient has:**

- ☐ Had an allogeneic bone marrow transplant: specify donor sex _____
- ☐ Had a previous karyotype — **NOTE:** If performed at an external laboratory, please attach the report or specify the karyotype in *Additional Information* section

## Indication and Reason for Testing

**Request for Expedited Result:**

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

**Fertility Indications:**

- ☐ Amenorrhea: specify primary or secondary _____
- ☐ Azoospermia/Oligospermia
- ☐ Recurrent miscarriages ( ≥3 ): specify number **(Required)** _____
- ☐ Premature ovarian failure
- ☐ IVF/ICSI Candidate
- ☐ Infertility

**Sex Chromosome Indications:**

- ☐ Atypical genitalia
- ☐ Klinefelter Syndrome
- ☐ Turner Syndrome

**Other Indications:**

- ☐ Family study: specify in *Additional Information* section
- ☐ Microarray follow-up: specify DNA # _____
- ☐ Targeted Aneuploidy follow-up: specify DNA # _____
- ☐ Other: specify _____

**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: bsofbbib

Facility:

Phone #: Fax #:

## Additional Information

**Phenotype/Family History:**

**Refer Out:** External laboratory paperwork must accompany sample

- ☐ Refer to : _____

Portal ID : _____ Test name : _____

**Culture and Storage:** (Available only for cultured cells from solid tissue/skin biopsies)

- ☐ Freeze cultured fibroblast cells (Long-term storage)
- ☐ Extract DNA for testing: Appropriate Molecular requisition must be included
- ☐ Extract DNA & Hold (5 year retention): Testing likely to follow within 5 years
- ☐ Extract DNA for Irreplaceable storage (25 year retention): Testing likely to follow beyond 5 years **AND** sample cannot be recollected

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# CONSTITUTIONAL CYTOGENETIC KARYOTYPE TESTING

*Order as: Cytogenetics IWK*

## Sample Requirements for Constitutional Cytogenetic Testing

**Peripheral Blood:** Collect 4mL peripheral blood in green NaHep (newborns <1 month: 2mL). Do not centrifuge or freeze. Ship to the laboratory at room temperature within 48 hours.

**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:** Collect 1cm$^3$ (when possible) fresh surgical or postmortem tissue using aseptic technique into a specimen jar containing sterile tissue media or saline. Do not freeze or place in formalin. Ship at 4˚C within 48 hours. *Note: cytogenetic karotype testing is no longer routinely performed for Fetal Tissue (products of conception/fetal demise); please use the FETAL GENETIC TESTING requisition for current testing options for these specimens.*

**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 green NaHep 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.

## Information about Cytogenetic 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. Structural rearrangements, mosaicism, or full or partial aneuploidy which may have reproductive implications for this individual or related family members.
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.

**SAMPLE STORAGE:** For more information, refer to our policy at refer to our policy at **https://iwkhealth.ca/CGL/SampleStorage**

**Temporary Retention of Fixed Cell Pellets**

- Following testing, fixed cell pellet from blood specimens will be stored for 6 months. 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.

**Fibroblast Cell Storage**

- Fibroblasts requested for long term cell storage will be frozen and retained for a minimum of 25 years.

**Temporary Retention of Residual DNA Samples**

- 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 DNA 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*