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

IWK Health

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

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: specify donor sex _____

☐ Patient has had a previous karyotype
NOTE: If performed at an external laboratory, please attach report or specify karyotype in *Additional Information* section

Request for Expedited Result:

☐ Patient or partner is currently pregnant: indicate EDC _____

☐ 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: Phone #:

Medical Facility: Fax #:

Reason for Testing

Fertility Indications:

☐ Amenorrhea- specify: primary or secondary _____

☐ Azoospermia/Oligospermia

☐ Recurrent miscarriages (≥3): specify number _____

☐ 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 _____

Additional Information

Phenotype/
Family History:

Culture and Storage: NOTE - Available for solid tissue/skin biopsy specimens only

☐ Freeze cultured fibroblast cells (long-term storage)

☐ Extract DNA from cultured cells for Molecular testing
Appropriate requisition must be included

☐ Extract DNA from cultured cells - Test request to follow

☐ Extract DNA from cultured cells for DNA Banking

Refer Out:

☐ Refer out to external laboratory - paperwork must accompany sample

Test name/ID_____

Facility_____

Sample Type

☐ Peripheral blood - Green NaHep 4mL (newborns <1 month: 2mL) IWK meditech CG

☐ Cord blood - see reverse for collection instructions. A maternal EDTA blood sample is also required

☐ Skin biopsy (3mm punch biopsy)

☐ Tissue (fresh surgical/postmortem): specify source _____

Collection Date/Time:

Collection Facility: Collector Initials:

Please see reverse page for collection information and shipping instructions

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CONSTITUTIONAL CYTOGENETIC KARYOTYPE TESTING

Sample Requirements for Constitutional Cytogenetic Testing

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

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: 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. *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.

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, 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. 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 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

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 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