Specimen Requirements – Genome Diagnostics:

- **Peripheral blood**
  - For leukemia/lymphoma - 20 mL in EDTA
  - For circulating tumour (cell free DNA) - 18 mL in STREK tubes
  - For all other testing - 5mL in EDTA

- **Bone marrow aspirate**
  - 1-2 mL in EDTA

- **Extracted DNA or RNA (>1µg)** (please circle nucleic acid)
  - Tissue Source __________________
  - Concentration: _________ Volume: _______

  Extracted nucleic acid will only be accepted from an appropriately qualified laboratory (ex. IQMH or equivalent).

Specimen Requirements – Cytogenetics:

- **Bone marrow aspirate**
  - 1.5-2 mL in sodium heparin

- **Peripheral blood**
  - 7 mL in sodium heparin

- **Tissue biopsy** (5-10mm³ in sterile medium/saline)

Solid Tumour:
- **Tissue or cell block (PREFERRED)**
  - Recut H&E slide (slide will not be returned) and copy of the pathology report is required
  - Note that a 3mm punch biopsy will be taken from block

- **Unstained slides (only if BLOCK is not available)**
  - Cut 8 unstained sections @ 7µm thickness on uncoated slides
  - Air dry (not in oven) unstained sections at room temperature.
  - For all tissues we require 2 H&E stained sections, one cut before cutting slides from block and one cut after.

- **PE tissue (curls)** 5x10µm sections in a sterile Eppendorf tube

- **Fresh tissue**:
  - 5mm³ frozen or in 10 mL sterile medium at room temperature
  - **Fresh/Fixed Cytology fluid**: as much as possible

Please ensure that you are using an updated copy of this requisition available at: http://www.uhn.ca/LMP/Health_Professionals/Documents/CytogeneticsandMolecularDiagnosticstests.pdf

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Clinical Diagnosis/Reason for Referral:

- **Diagnosis:** ____________________________
- **Monitoring:** (for follow-up samples)
- **Other:** ________________________________

Treatment (specify type) _____________________

Date of last treatment _____________________

### Genome Diagnostics Tests - Hematological

#### Leukemia: Single Gene testing

- **^BCR/ABL1 t(9;22)**
- **^ABL1 kinase domain mutation**
- **^RUNX1/RUNX1T1 (AML/ETO) t(8;21)**
- **^CBFB/MYH11 Inv(16) or t(16;16)**
- **PML/RARA t(15;17)**
- **FLT3/NPM1 (new diagnosis)**
- **^FLT3/NPM1 (prevalent case)**
- **^CLL IGHV Somatic Hypermutation (for patients requiring treatment only)**

#### Lymphoma: please attach corresponding pathology report

- **^B-cell Clonality**
- **^T-cell Clonality**
- **^MYD88**

#### Bone marrow/Stem cell transplant monitoring:

- **^15 STRs and amelogenin XY loci**

**Please specify:**

- Donor
- Recipient Pre-SCT
- Recipient Post-SCT

#### Other:

- **^BRAF (p.V600E/K only)** (Hairy cell leukemia, Langerhans cell histiocytosis, Erdheim-Chester)
- **^KIT (Mastocytosis - BM or involved tissue preferred)**
- **^JAK2 (Exon 12 + Exon 14 p.V617F) / CALR (MPD)**

#### Identity Testing (15 STRs and amelogenin XY loci):

- **^Specimen matching (Please provide control specimen, specimen in question and details)**

#### Virus Detection (Nasopharyngeal Carcinoma):

- **^Quantitative EBV from blood plasma**

*Please provide a Karyotype report if analysis was not done at UHN*

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^Indicates a test that will be billed to the referring hospital, laboratory, physician or medical group.
**Patient Information or Hospital Stamp Here**

Last Name:  
First Name:  
Date of Birth (MM/DD/YYYY):  
Gender:  
Health Card #:  
Hospital #:  

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**Note:** Please send requisitions to specimen holding facility to ensure that block/slides accompany requisitions when sent to testing lab

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### Genome Diagnostics Tests – Solid Tumour

#### Adenocarcinoma of the Lung
- EGFR -- Exons 18,19,20,21 (clinically relevant variants only)
- EGFR – p.T790M mutation only (solid tumour/cell block)
- *EGFR* – p. T790M mutation only (Circulating tumour DNA in blood) *peripheral blood in STRECK tube required* (see pg. 1 for specimen requirements)

Please indicate primary EGFR mutation:
- Exon 19 deletion
- Exon 21 L858R
- Other __________

#### Colorectal Adenocarcinoma
- Comprehensive Sequencing (NGS) (BRAF, KRAS, NRAS, PIK3CA)
- MLH1 Methylation (*Please include IHC MMR results if completed*)
- ^MSI - Only performed for cases with equivocal IHC MMR results
  - requires normal reference (normal tissue or blood)

#### Endometrial Carcinoma
- MLH1 Methylation (*Please include IHC MMR results if completed*)
- ^MSI – Only performed for cases with equivocal IHC MMR results
  - requires normal reference (normal tissue or blood)

#### Gastrointestinal Stromal Tumour
- ^Comprehensive Sequencing (NGS) (KIT, PDGFRA)

#### Glioma
- ^MGMT methylation
- ^IDH1/IDH2 Sequencing

#### Melanoma (Somatic)
- BRAF (p.V600E/K only)
- Comprehensive Sequencing (NGS)
- BAP1  CDK6  GNA11  KIT
- BRAF  CDKN2A  GNAQ  NRAS
- CDK4  EIF1AX  HRAS  SF3B1

#### Papillary Thyroid Carcinoma
- ^BRAF (p.V600E/K only)

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^Indicates a test that will be billed to the referring hospital, laboratory, physician or medical group.
**Note:** Please send requisitions to specimen holding facility to ensure that block/slides accompany requisitions when sent to testing lab

### Genome Diagnostics Tests – Solid Tumour

**Polymorphous Low Grade Adenocarcinoma (PLGA) Salivary gland**
- ^PRKD1 Sequencing

**Serous Carcinoma – High Grade (Ovarian/Tubal/Peritoneal)**
- Comprehensive Sequencing (NGS) (BRCA1, BRCA2)

*Tumour Cellularity within circled area_______%*

EXTERNAL pathology review required prior sending material.

**For Solid Tumour:** Circled HE required prior to sending block. **Tumour cellularity must be a minimum of 20% tumour within circled area.** A ~3mm area will be cored from block. If a block cannot be sent, please send sections (see page 1).

**For Cytology Specimens:** **Tumour cellularity must be a minimum of 10% tumour.** Please see page 1 for specimen requirements.

### Virus Detection

- ^HPV (DNA tissue testing-37 genotypes)

^Indicates a test that will be billed to the referring hospital, laboratory, physician or medical group.
A Pathology / Hematology report must accompany or be sent (fax/email) for all bone marrow samples and solid tumour testing. Samples will be banked and testing delayed until this information is received.

### Clinical Diagnosis/Reason for Referral:

- **Diagnosis:**
- **Monitoring:** (follow-up sample) __________
- **Treatment** (specify type) __________
- **Other:** __________

*proper interpretation of post-transplant karyotyping requires information about transplant type*

- Pre-transplant monitoring
- Post-transplant monitoring
  - Type of transplant: □ auto □ allo □ sex-mismatch
  - Date of transplant (mm/yy): __________

### Conventional Cytogenetics

- ^G-banded karyotyping on bone marrow
- ^G-banded Karyotyping on peripheral blood
  (Only to confirm a constitutional abnormality detected on bone marrow karyotype)

#### FISH for Hematologic Disorders

**Chronic Lymphocytic Leukemia**

- ^CLL FISH Panel (11q del, trisomy 12, 13q del, 17p del)
  - diagnostic
  - follow-up (justify): __________

**Chronic Myelogenous Leukemia**

- ^BCR/ABL1 (only for molecular negative)

### Plasma Cell Neoplasms with CD138 Cell Enrichment

(Magnetic separation requires ≥ 1mL marrow aspirate. If other tests are requested, e.g. karyotype, please submit an additional 1.5-2mL of aspirate in a separate tube.)

- ^Multiple Myeloma Panel
- ^Amyloidosis Panel

### Eosinophilia FISH Panel

- PDGFRA / PDGFRB / FGFR1

### MDS/AML Panel

(ordered automatically for failed karyotypes)

- ^Del(5q)/-5 and Del(7q)/-7
- ^KMT2A Break-apart
- ^MECOM 3q26 rearrangement

### Lymphoid Disorders:

**High Grade B-cell Lymphoma Panel**

- ^Entire Panel
- ^MYC
- ^IGH/BCL2 t(14;18)(q32;q21)
- ^BCL6

**Anaplastic large cell lymphoma**

- ^ALK

**MALT lymphoma**

- ^MALT1

**Mantle cell lymphoma**

- ^CCND1/IGH t(11;14)(q13;q32)

**Follicular lymphoma / Diffuse Large B-Cell Lymphoma**

- ^IGH/BCL2 t(14;18)(q32;q21)
- ^BCL6

### Solid Tumour *:

**Breast cancer**

- HER2 Amplification

**Brain Cancer: Gliomas**

- ^1p/19q Co-deletion FISH

**Lung Adenocarcinoma**

- ALK
- ^ROS1

**Sarcoma and Carcinoma FISH**

- ^EWSR1 - EWS-Family Tumours
- ^FUS – Low Grade Fibromyxoid Sarcoma
- ^SS18 – Synovial Sarcoma
- ^MAML2 – Mucoepidermoid Carcinoma
- ^ETV6 – Secretory Carcinoma
- ^CDKN2A (p16) – Malignant Mesothelioma

*tests are performed on FFPE tissue only, see specimen requirements. For cytology specimens please contact the lab to inquire if test is validated.

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For all cytogenetics inquiries please contact canccytogenetics@uhn.ca  Version 6.0  Feb 5  2020