Department of Clinical Laboratory Genetics
Genome Diagnostics – Hereditary Disorders

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Hours of Operation (Mon-Fri) 8:30AM-4:30PM
CAP#: 7175217  CLIA#:99D1106115
IQMH# 4204-site 0141

Patient Information or Hospital Stamp Here
Last Name:
First Name:
Date of Birth (MM/DD/YYYY):
Gender:
Health Card #:
Hospital #:

Instructions:
1. Complete all information as requested
2. Send requisition with specimen to address above
3. Keep specimen at room temperature unless frozen
4. If shipping, send same day or next day delivery
5. Specimen labelling: Name, DOB, MRN#

Referring Physician Signature: __________________________

Information for Reporting:
Full Name of Referring physician:
Hospital/Address:

Phone:
Fax:
Copy Report To: __________________________

Specimen Requirements
- Peripheral blood
  5 mL in EDTA
- Extracted DNA (not accepted for deletion/duplication testing)

Tissue Source ________, Conc. ________, Vol. ________

Collection Information
Collection date/time: __________________________
Collected by: __________________________

Test Indication
- Please provide any available clinical information and/or complete the Clinical Data Information Sheet.

- Diagnosis

- Known Family Variant Analysis - Please provide variant details on Pg. 2 of this requisition
  **If no family member has been tested at UHN a positive genetic test report of a family member is required.

- Other: __________________________

Pedigree
Please draw or attach a pedigree and provide any relevant clinical information.
Ethnicity: __________________________

Please ensure you are using an updated version of this requisition, available at
http://www.uhn.ca/LMP/Health_Professionals/Documents/MolecularDiagnosticsHereditary.pdf
**Molecular Diagnostics Tests**

*Note: Only variants in the genes requested on this requisition will be investigated and reported*

*Indicates genes for which deletion/duplication testing will be completed*

- **Amyloidosis** (APOA1, APOA2, B2M, FGA, GSN, LYZ, TTR)
  - Sequencing

- **Birt-Hogg-Dube** *(FLCN*)
  - Sequencing + Deletion/Duplication

- **Cowden Syndrome** *(PTEN*)
  - Sequencing + Deletion/Duplication

- **Hemochromatosis** *(HFE)*
  - p.Cys282Typr/p.His63Asp

- **Hereditary Leiomyomatosis and Renal Cell Carcinoma** *(FH)*
  - Sequencing + Deletion/Duplication

- **Hereditary Renal Cancer** *(BAP1*, CDC73, Dicer1, FH*, FLCN*, MET, MITF [p.Glu318Lys only], PTEN*, SDHA, SDHAF2*, SDHB*, SDHC*, SDHD*, TMEM127, TP53, TSC1*, TSC2*, VHL*)
  - Sequencing + Deletion/Duplication

- **Hereditary Renal Cancer/ Pheochromocytoma-Paraganglioma Overlap** *(MAX, RET, SDHA, SDHAF2*, SDHB*, SDHC*, SDHD*, TMEM127, VHL*)
  - Sequencing + Deletion/Duplication

- **Lynch Syndrome – Targeted** *(MLH1*, MSH2*, MSH6*, PMS2*)
  - Sequencing + Deletion/Duplication *(includes EPCAM)*
  - Germline Methylation *(MLH1)*

- **Lynch Syndrome – Comprehensive** *(APC, BMPR1A, CHEK2, MLH1*, MSH2*, MSH3, MSH6*, MUTYH, NTHL1, PMS2*, PTEN*, POLD1, POLE, SMAD4, STK11, TP53)*
  - Sequencing + Deletion/Duplication *(includes EPCAM)*

- **Malignant Hyperthermia** *(CACNA1S, RYR1)*
  - Sequencing

- **Melanoma – Familial** *(BAP1*, CDK4*, CDKN2A*, MC1R, MITF [p.Glu318Lys only], POT1)*
  - Sequencing + Deletion/Duplication

- **Thrombosis**
  - Factor V (Leiden)/Prothrombin/Factor II (G20210GA)
  - MTHFR (C677T) *(only if homocysteine is elevated)*

- **Tuberous Sclerosis** *(TSC1*, TSC2*)
  - Sequencing + Deletion/Duplication

- **Von Hippel-Lindau Syndrome** *(VHL*)
  - Sequencing + Deletion/Duplication

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**Known Family Variant Analysis**

Gene/Variant: ________________________________________________

Proband Name/UHN #: ____________________________________

*(If proband or other relatives with a positive result was not tested at UHN, please include copy of report)*

Relationship of this individual to proband: __________________

**Comments/Special Instructions:**

____________________________________________

____________________________________________

____________________________________________

Please ensure you are using an updated version of this requisition, available at


Version 6.0 August 2019
CANCER

- Adrenocortical carcinoma
- Brain
  - Astrocytoma
  - Choroid plexus carcinoma
  - Glioblastoma
  - Hemangioblastoma
  - Medulloblastoma
  - Other
- Breast
  - Ductal carcinoma in situ (DCIS)
  - Invasive ductal carcinoma
  - Invasive lobular carcinoma
  - Lobular carcinoma in situ (LCIS)
  - Phyllodes
  - Other
- Colon
  - Adenocarcinoma
  - Squamous cell carcinoma
  - Other
- Endometrial
- Duodenal
- Hepatobiliary
- Leukemia/lymphoma
  - Acute lymphoblastic leukemia (ALL)
  - Acute myelogenous leukemia (AML)
  - Chronic lymphoblastic leukemia (CLL)
  - Chronic myelogenous leukemia (CML)
  - Hodgkin’s lymphoma
  - Non-Hodgkin’s lymphoma
- Lung
  - Type
- Melanoma
  - Cutaneous
  - Uveal
- Neuroendocrine tumour (site)
- Ovarian
  - Mucinous
  - Papillary
  - Serous
  - Other
- Pancreatic
- Prostate
- Rectal
- Renal
  - Chromophobe
  - Clear cell
  - Collecting duct
  - Oncocytoma
  - Papillary: Type 1________, Type 2______
- Sarcoma
  - Osteosarcoma
  - Soft tissue sarcoma (site)
- Small bowel
- Stomach
- Testicular
- Thyroid
  - Follicular
  - Medullary
  - Papillary
  - Other

OTHER FEATURES

Neurological
- Autism
- Developmental delay/intellectual disability
- Lhermitte-Duclos disease
- Psychiatric disease (type)
- Seizures

Ocular
- Retinal angioma/hemangioma/hamartoma

Auditory
- Endolymphatic sac tumour
- Hearing loss

Endocrine
- Goiter
- Paraganglioma (site)
- Pheochromocytoma
- Thyroid adenoma

Cardiac
- Arrhythmia
- Cardiac rhabdomyoma

Pulmonary
- Lung cysts
- Lymphangiomyomatosis (LAM)
- Pneumothorax

Gastrointestinal
- Colon polyps (approx. number)
- Adenomatous
- Ganglioneuromatous
- Hamartomatous
- Juvenile
- Other

Genitourinary
- Renal angiomyolipomas
- Renal cysts
- Pancreatic cysts
- Uterine fibroids

Skin
- Acral keratoses
- Angiofibromas
- Atypical/dysplastic nevi
- Fibrofolliculomas
- Fibromas
- Lipomas
- Tricheleomomas
- Other

Other
- Fibrocystic breast disease
- Arteriovenous malformations (location)
- Macrocephaly
- Other