

Department of Clinical Laboratory Genetics

Genome Diagnostics – Hereditary Disorders



Toronto General Hospital

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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#**

Information for Reporting:

Full Name of Referring physician: _____

Hospital/Address: _____

Phone: _____

Fax: _____

Copy Report To: _____

Referring Physician Signature: _____

Specimen Requirements

Peripheral blood

5 mL in EDTA

Extracted DNA (not accepted for specific MLPA-based tests)

Tissue Source _____ Conc. _____ Vol. _____

*extracted DNA will only be accepted from an appropriately accredited and qualified laboratory (i.e. IQMH or equivalent)

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 Familial Variant Analysis - Please provide variant details on Pg. 3 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 _____

^ indicates a test that will be billed to a hospital, referring laboratory, referring physician or medical group (hemochromatosis and thrombosis, only). If billing address is different from above, please provide billing information here.

Name: _____ Phone: _____ Fax: _____

Full Address: _____

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

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Hereditary Cancer Panel Testing - Sequencing + CNV analysis
(*CNV only)

Hereditary Breast/Ovarian/Prostate Cancer (*ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM*, HOXB13, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53*)

Hereditary Central Nervous System Tumours (*APC, EPCAM*, LZTR1, MLH1, MSH2, MSH6, NF1, NF2, PMS2, POLE, POT1, PTCH1, PTEN, SMARCB1, SMARCE1, SUFU, TP53, TSC1, TSC2, VHL*)

Hereditary Endometrial Cancer (*BRCA1, BRCA2, EPCAM*, MLH1, MSH2, MSH6, PMS2, POLD1, POLE, PTEN*)

Hereditary Gastric Cancer (*APC, ATM, BRCA1, BRCA2, CDH1, CTNNA1, EPCAM*, MLH1, MSH2, MSH6, PALB2, PMS2, SDHB, SDHD, SMAD4, STK11, TP53*)

Familial GI Cancer (Lynch syndrome, Gastric, Pancreas, Polyposis) (*APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CDKN2A, CHEK2, CTNNA1, EPCAM*, GALNT12, GREM1*, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RNF43, RPS20, SDHB, SDHD, SMAD4, STK11, TP53*)

Familial Gastrointestinal Stromal Tumour (*KIT, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD*)

Lynch syndrome (*MLH1, MSH2, MSH6, PMS2, EPCAM**)

Germline Methylation (*MLH1*)

MMR IHC results (if completed):

Familial Melanoma (*BAP1, BRCA2, CDK4, CDKN2A, MITF, POT1, PTEN*)

Hereditary Pancreatic Cancer (*ATM, BRCA1, BRCA2, CDKN2A, EPCAM*, MLH1, MSH2, MSH6, PALB2, PMS2, STK11, TP53*)

Hereditary Pheochromocytoma-Paraganglioma syndrome (*FH, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL*)

Polyposis (*APC, BMPR1A, EPCAM*, GALNT12, GREM1*, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, RPS20, SMAD4, STK11, TP53*)

Familial Renal Cancer (*BAP1, FH, FLCN, MET, MITF, PTEN, SDHA, SDHAF2, SDHB, SDHC, SDHD, TP53, TSC1, TSC2, VHL*)

Familial Soft Tissue Cancers (*APC, ATM, BRCA1, BRCA2, CHEK2, EPCAM*, MLH1, MSH2, MSH6, NF1, PMS2, TP53*)

Ashkenazi Jewish Panel (*APC [p.Ile1307Lys], BRCA1 [c.185delAG/187delAG; c.5382insC/5385insC], BRCA2 [c.617delT], CHEK2 [c.1283C>T], GREM1 [40 kb dup], MSH2 [p.Ala636Pro], MSH6 [c.3984_3987dupGTCA; c.3959_3962delCAAG]*)

Familial 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: _____

Patient Information or Hospital Stamp Here

Last Name:

First Name:

Date of Birth (MM/DD/YYYY):

Gender:

Health Card #:

Hospital #:

**Single Gene Disorder/Small Panel
Testing – Sequencing + CNV analysis**

- Amyloidosis** (*APOA1, APOA2, B2M, FGA, GSN, LYZ, TTR*)
- AXIN2-related Attenuated Familial Adenomatous Polyposis** (*AXIN2*)
- BAP1 Tumour Predisposition/Mesothelioma** (*BAP1*)
- Birt-Hogg-Dubé syndrome** (*FLCN*)
- Carney Complex** (*PRKAR1A*)
- DICER-associated syndrome** (*DICER1*)
- Dysplastic Nevus syndrome** (*CKD4, CDKN2A*)
- Familial Adenomatous Polyposis (FAP)**
 - APC only*
 - APC + MUTYH*
- Familial Isolated Pituitary Adenoma** (*AIP*)
- ^Hemochromatosis** (*HFE*; p.Cys282Tyr, p.His63Asp)
- Hereditary Hyperparathyroidism** (*CDC73, MEN1*)
- Hereditary Leiomyomatosis and Renal Cell Carcinoma** (*FH*)
- Hereditary Lung Cancer** (*EGFR*; p.Thr790Met, p.Val834Ile, p.Val769Met)
- Li-Fraumeni syndrome** (*TP53*)
- Malignant Hyperthermia** (*CACNA1S, RYR1*)
- Multiple Endocrine Neoplasia, Type I** (*MEN1, CDKN1B*)

- Multiple Endocrine Neoplasia, Type 2** (*RET*)
- Neurofibromatosis, Type I** (*NF1*)
- Nevoid Basal Cell Carcinoma/Gorlin syndrome** (*PTCH1, SUFU*)
- Nijmegen Breakage syndrome** (*NBN*)
- Peutz Jeghers syndrome** (*STK11*)
- PTEN Hamartoma Tumour syndrome** (*PTEN*)
- Rare Polyposis** (*GALNT12, RPS20*)
- Retinoblastoma** (*RB1*)
- Rhabdoid Predisposition syndrome** (*SMARCA4, SMARCB1*)
- Schwannomatosis** (*NF2, LZTR1, SMARCB1*)
- Sessile Serrated Polyposis Cancer syndrome** (*RNF43*)
- Small Cell Carcinoma of the Ovary, Hypercalcemic type (SCCOHT)** (*SMARCA4*)
- ^Thrombosis**
 - Factor V (Leiden)/Prothrombin/Factor II (G20210GA)
 - MTHFR* (C677T) (only if homocysteine is elevated)
- Tuberous Sclerosis** (*TSC1, TSC2*)
- Von Hippel-Lindau syndrome** (*VHL*)

Comments/Special Instructions:

CLINICAL DATA INFORMATION SHEET

PATIENT NAME/DOB: _____

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
 - Trichilemmomas
 - Other _____
- Other**
- Fibrocystic breast disease
 - Arteriovenous malformations (location) _____
 - Macrocephaly
 - Other _____