Department of Clinical Laboratory Genetics

Genome Diagnostics – Hereditary Disorders

Toronto General Hospital
Eaton Wing 11-444, 200 Elizabeth Street
Toronto, Ontario M5G 2C4
Director: Tracy Stockley, PhD, FCCMG, FACMG
Phone: (416) 340-4800 x5739/7624
Fax: (416) 340-3596
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):

Sex assigned at birth:

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**)
- ❑ **Saliva** (Oragene Kit: OG-500/OG-600 only)
- ❑ **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** - symptoms/features of condition in THIS individual; please provide clinical details
- ❑ **Known Familial 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** – Please provide details/justification here

Clinical/Family History Information

TESTING MAY BE PUT ON HOLD IF INSUFFICIENT CLINICAL DETAILS PROVIDED

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
https://www.uhn.ca/Labs/services_clinicians#Requisitions

UHN Toronto General Toronto Western Princess Margaret Toronto Rehab Michener Institute
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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)*
- ❑ **Familial Melanoma** *(BAP1, BRCA2, CDK4, CDKN2A, MITF, POT1, PTEN)*
- ❑ **Lynch syndrome** *(EPCAM*, MLH1, MSH2, MSH6, PMS2)*
  - ❑ Germline Methylation *(MLH1)*

MMR IHC results (if completed):

_____

- ❑ **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])*
- ❑ **Comprehensive Cancer Panel** *(AIP, APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, CTNNA1, DICER1, EGFR [p.Val769Met, p.Thr790Met], EGLN1, EPCAM*, EXT1, EXT2, FH, FLCN, GALNT12, GREM1*, HOXB13, KIT, LZTR1, MAX, MEN1, MET, MITF, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PDGFRA, PMS2, POLD1, POLE, POT1, PRKAR1A, PTCH1, PTEN, RAD51C, RAD51D, RB1, RECQL, RET, RNF43, RPS20, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL)*

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: _____

Comments/Special Instructions/Add'l Clinical History:

_____

_____

_____

_____

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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)*

Pharmacogenomic Testing:

- ❑ *DPYD**2A, *DPYD**9B, *DPYD**13, HapB3 prior to fluoropyrimidine treatment

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 I____ 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
- ☐ Trichelemommas
- ☐ Other _____

Other

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