SickKids®

Department of Paediatric Laboratory Medicine

555 University Avenue
Room 3416, Roy C. Hill Wing
Toronto, ON, M5G 1X8, Canada

Tel: 416-813-7200 x1
Fax: 416-813-7732

(CLIA # 99D1014032)

Genome Diagnostics
Hereditary Cancer Testing Panel

www.sickkids.ca/en/care-services/for-health-care-providers/lab-testing-services/genome-diagnostics

Patient Name:

Preferred Name (if different):

Date of Birth (DD/MM/YYYY):

Legal Sex: ◯ *Male* ◯ *Female* ◯ *Non-binary/U/X*

Sex Assigned at Birth (if different): ◯ *Male* ◯ *Female* ◯ *Unassigned*

Gender Identity): ◯ *Male* ◯ *Female* ◯ *Non-binary/U/X*

MRN:

Parent's Name:

Address:

For Ontario Only

Provincial Health Card #: *Version:*

*If the patient does not have **valid** coverage under the Ontario Health Insurance Plan (OHIP), please complete page 6 of the requisition*

Testing is provided for medical purposes only and results are not intended for forensic use. The laboratory is not a forensically accredited laboratory.

Ordering Physician:

Name: _____

Institution: _____

Address: _____

Phone: _____

Fax: _____

Email address: _____

Signature *(required)* _____

Copy Report To:

Name: _____

Institution: _____

Address: _____

Phone: _____

Fax: _____

Sample Information:

Date obtained (DD-MM-YYYY): _____-_____-_____

Your referring laboratory reference # / Pathology ID: _____

☐ Blood in EDTA (purple top tube): min. 4 mL (0.5 - 3 mL for newborns)

☐ DNA: min. 5 µg in low TE buffer (Source: _____)

☐ Fibroblasts

For laboratory use only:

Date (DD-MM-YYY) | Time Received

_____-_____-_____ | _____ h

Order #: _____

Specimen type, amt & # of tubes: _____

Comments:

Pedigree No. / Patient No. _____ / _____

Primary Tumour:

_____

Reason for Testing:

☐ Diagnosis

☐ Familial mutation / variant analysis

☐ Tumour finding

Expedited Testing:

☐ Yes ☐ No

Variant information:

Where a familial mutation or variant is known, complete below and attach a copy of the proband's report:

Gene & NM#: _____

Mutation / variant(s): _____

SickKids Laboratory/Order number: _____

SickKids Pedigree/Family number: _____

Name of proband: _____

Name(s) & DOB of other submitted family members: _____

Ordering Checklist:

☐ Specimen tube labeled with at least two identifiers

☐ Completed test requisition form *(pages 1-5)*

☐ Completed billing form *(page 6, if applicable)*

Clinical Diagnostics and Family History:

Draw or attach a pedigree and provide any relevant information below, including clinical and family history details.

Ethnicity: _____

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SYNDROME / DISEASE SITE TESTING PANELS
(sequencing and deletion/duplication; *deletion/duplication analysis only)

√	SYNDROME / DISEASE SITE PANEL (# of genes)	ASSOCIATED GENE(S)
☐	Comprehensive Panel (73)	*AIP, APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, CTNNA1, DICER1, EGFR, EPCAM*, FH, FLCN, GALNT12, GREM1*, HOXB13* (p.Gly85Glu), *KIT, LZTR1, MAX, MBD4, MEN1, MET, MITF* (p.Glu318Lys), *MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PDGFRA, PMS2, POLD1, POLE, POT1, PRKAR1A, PTCH1, PTEN, RAD51C, RAD51D, RB1, RET, RNF43, RPS20, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL*
☐	Hereditary Cancer Core Panel (40)	*APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, CTNNA1, EPCAM*, GALNT12, GREM1*, HOXB13* (p.Gly85Glu), *MBD4, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, RNF43, RPS20, SDHB, SDHD, SMAD4, STK11, TP53*
☐	Hereditary Breast / Ovarian / Prostate (19)	*ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM*, HOXB13* (p.Gly85Glu), *MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53*
☐	Hereditary GI (Lynch, Colorectal, Endometrial, Gastric, Pancreas, Polyposis) (34)	*APC, ATM, AXIN2, BMPR1A, BRCA1, BRCA2, CDH1, CDK4, CDKN2A, CHEK2, CTNNA1, EPCAM*, GALNT12, GREM1*, MBD4, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RNF43, RPS20, SDHB, SDHD, SMAD4, STK11, TP53*
☐	Gastric Cancer (18)	*APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CTNNA1, EPCAM*, MLH1, MSH2, MSH6, PALB2, PMS2, SDHB, SDHD, SMAD4, STK11, TP53*
☐	Polyposis (23)	*APC, AXIN2, BMPR1A, EPCAM*, GALNT12, GREM1*, MBD4, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, RPS20, SMAD4, STK11, TP53*
☐	Pancreatic Cancer (14)	*ATM, APC, BRCA1, BRCA2, CDK4, CDKN2A, EPCAM*, MLH1, MSH2, MSH6, PALB2, PMS2, STK11, TP53*
☐	GIST (8)	*KIT, NF1, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD*
☐	Sarcoma (14)	*APC, ATM, BRCA1, BRCA2, CHEK2, DICER1, EPCAM*, MLH1, MSH2, MSH6, NF1, PMS2, RB1, TP53*
☐	Hereditary Pheochromocytoma and Paraganglioma (PPGL) (12)	*FH, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL*
☐	CNS (20)	*APC, EPCAM*, LZTR1, MLH1, MSH2, MSH6, NF1, NF2, PMS2, POLE, POT1, PTCH1, PTEN, SMARCB1, SMARCE1, SUFU, TP53, TSC1, TSC2, VHL*
☐	Hereditary Melanoma (8)	*BAP1, BRCA2, CDK4, CDKN2A, MBD4, MITF* (p.Glu318Lys), *POT1, PTEN*

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SYNDROME / DISEASE SITE TESTING PANELS cont…
(sequencing and deletion/duplication; *deletion/duplication analysis only)

√	SYNDROME / DISEASE SITE PANEL (# of genes)	ASSOCIATED GENE(S)
☐	Hereditary Renal (15)	*BAP1, FH, FLCN, MET, MITF* (p.Glu318Lys), *PTEN, SDHA, SDHAF2, SDHB, SDHC, SDHD, TP53, TSC1, TSC2, VHL*
☐	Lung Cancer (4)	*EGFR, TP53, ATM, BRCA2*

PEDIATRIC CANCER SYNDROMES PANELS

√	SYNDROME / DISEASE SITE PANEL (# of genes)	ASSOCIATED GENE(S)
☐	Medulloblastoma (7)	*APC, BRCA2, ELP1, PALB2, PTCH1, SUFU, TP53*
☐	Nephroblastoma (Wilms tumour) (13)	*AMER1, ASXL1, BUB1B, CTR9, DICER1, DIS3L2, FBXW7, GPC3, NSD1, REST, TP53, TRIP13, WT1*
☐	Neuroblastoma (16)	*ALK, BARD1, BRAF, EZH2, HRAS, KRAS, LZTR1, MAP2K1, NF1, NRAS, PHOX2B, PTPN11, RAF1, SMARCA4, SOS1, TP53*

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SINGLE GENE SYNDROMES OR SMALL PANELS		
√	**SYNDROME (# of genes)**	**ASSOCIATED GENE(S)**
☐	AXIN2-related Attenuated Familial Adenomatous Polyposis (1)	*AXIN2*
☐	BAP1 Tumour Predisposition Syndrome (1)	*BAP1*
☐	Birt-Hogg-Dube Syndrome (1)	*FLCN*
☐	Carney Complex (1)	*PRKAR1A*
☐	DICER-associated Syndrome (1)	*DICER1*
☐	Dysplastic Nevus Syndrome (2)	*CDK4, CDKN2A*
☐	Familial Adenomatous Polyposis (CHRPE, CMV Thyroid, Desmoid)	*APC* ☐ add *MUTYH*
☐	Familial Isolated Pituitary Adenoma (1)	*AIP*
☐	Hereditary Parathyroid Neoplasia (2)	*CDC73, MEN1*
☐	Hereditary Leiomyomatosis and Renal Cell Cancer (1)	*FH*
☐	Hereditary Multiple Osteochondromas (2)	*EXT1, EXT2*
☐	Li-Fraumeni Syndrome (1)	*TP53*
☐	MBD4 Tumour Predisposition Syndrome (1)	*MBD4*
☐	Multiple Endocrine Neoplasia, Type 1 (2)	*MEN1, CDKN1B*
☐	Multiple Endocrine Neoplasia, Type 2 (1)	*RET*
☐	Neurofibromatosis, Type 1 (1)	*NF1*
☐	Nevoid Basal Cell Carcinoma Syndrome / Gorlin Syndrome (2)	*PTCH1, SUFU*
☐	Nijmegen Breakage Syndrome (1)	*NBN*
☐	Peutz-Jeghers Syndrome (1)	*STK11*
☐	PTEN Hamartoma Tumour Syndrome (1)	*PTEN*
☐	Rare Polyposis Genes (2)	*GALNT12, RPS20*
☐	Retinoblastoma (1)	*RB1*

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SINGLE GENE SYNDROMES OR SMALL PANELS cont…		
√	SYNDROME (# of genes)	ASSOCIATED GENE(S)
☐	Rhabdoid Predisposition Syndrome (2)	*SMARCA4, SMARCB1*
☐	Schwannomatosis (3)	*NF2, LZTR1, SMARCB1*
☐	Sessile Serrated Polyposis Cancer Syndrome (1)	*RNF43*
☐	Small Cell Carcinoma of the Ovary, Hypercalcemic Type (SCCOHT) (1)	*SMARCA4*
☐	Tuberous Sclerosis (2)	*TSC1, TSC2*
☐	Von Hippel-Lindau Syndrome (1)	*VHL*

☐ ASHKENAZI JEWISH PANEL		
	HGVS Nomenclature	ALTERNATE NAME
1	NM_000038.6(APC):c.3920T>A (p.Ile1307Lys)	*APC* I1307K
2	NM_007294.4(BRCA1):c.68_69del (p.Glu23fs)	*BRCA1* (185delAG or 187delAG)
3	NM_007294.3(BRCA1):c.5266dupC (p.Gln1756Profs)	*BRCA1* (5382insC or 5385insC)
4	NM_000059.4(BRCA2):c.5946del (p.Ser1982fs)	*BRCA2* (617delT)
5	NM_007194.4(CHEK2):c.1283C>T (p.Ser428Phe)	*CHEK2* 1283C>T
6	N/A	*GREM1* 40 kb dup
7	NM_000251.2(MSH2):c.1906G>C (p.Ala636Pro)	*MSH2* A636P
8	NM_000179.2(MSH6):c.3984_3987dupGTCA (p.Leu1330Valfs)	*MSH6* c.3984_3987dupGTCA
9	NM_000179.2(MSH6):c.3959_3962delCAAG (p.Ala1320Glufs)	*MSH6* c.3959_3962delCAAG

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BILLING FORM

Completion of Billing Form NOT required for patients with an Ontario Health Card Number.

The referring hospital/laboratory/clinic, insurer (other than OHIP), or a patient/guardian will be billed for the services rendered, as specified in the sections below by the referring physician. Invoices are sent upon completion of each test/service, which are itemized and include the date of service, provider name, patient name, and charge. **Failure to provide appropriate billing information will result in delays to testing.**

How to complete the Billing Form:

- For hospital/laboratory/clinic to be billed: Complete Section 1
- For self-pay: Complete Section 2
- For insurers other than OHIP (including but are not limited to the Interim Federal Health Program, University Health Insurance Program): An approval letter **must** be attached with the requisition

Section 1: Complete to have the Healthcare Provider billed:

Referring Laboratory's Reference #:_____

Billing address of hospital, referring laboratory, clinic, referring physician, or medical group (if different from requisition):

Name: _____
Address: _____
City: _____ Prov/State: _____
Postal/Zip Code: _____ Country: _____
Contact Name: _____ Contact Telephone #: _____

Section 2: Complete to have Patient/Guardian billed directly:

- *Advise the parent/guardian to expect a bill from SickKids*
- *Information provided below should reflect a valid credit card*
- *Personal cheques are a preferred method of payment but will be accepted on a case-by-case basis*

Send bill to (check one): ☐ Patient ☐ Guardian

Method of Payment (check one): ☐ American Express ☐ MasterCard ☐ Visa

Name as it appears on credit card:_____

Credit card #: _____ CVS #: _____

Expiry date on credit card: _____

Signature of credit card holder (Required): _____

Mailing Address of Patient/Guardian (if different from requisition):

Name: _____
Address: _____
_____ Apt. #: _____
City: _____ Prov/State: _____
Postal/Zip Code: _____ Country: _____

Additional Contact Information

Patient's phone # with area code:

_____

-or-

Guardian's phone # with area code:

_____