



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

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

Ordering Laboratory or Institution:

Name: _____
Address: _____
Phone: _____
Fax: _____
Email Address: _____
Ordering Physician: _____
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. _____ / _____

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

Primary Tumour:

Reason for Testing:

- ☐ Diagnosis
☐ Familial mutation / variant analysis
☐ Tumour finding

Expedited Testing:

- ☐ Yes ☐ No

Note: All indications and subpanels are funded for Hereditary Cancer Panel.

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

PEDIATRIC CANCER SYNDROMES PANELS

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



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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:
