



Paediatric Laboratory Medicine

MOLECULAR GENETICS Requisition

MOLECULAR GENETICS LABORATORY (CLIA # 99D1014032)
Roy C. Hill Wing Rm 3421
555 University Ave Toronto ON M5G 1X8 Canada
Tel: (416) 813-7200 x1 Fax: (416) 813-7732

Patient Name
Birthdate (YYYY-MM-DD)
Sex
Parent's Name
Address
Telephone #

For Canada Only:
Provincial Health Card # _____ Version _____
Issuing Province _____

Referring physician: (Please Print)

Name:
Address:
Tel () _____ Fax () _____
e-mail address:
Signature (required) _____

Test request:

Write gene/disease name below or check box on pages 2 and 3.
Testing for a known mutation:
Gene: _____ Mutation: _____
SickKids laboratory number of relative: _____

CC report to:

Name:
Address:
Tel () _____ Fax () _____

Reason for referral:

- Diagnostic testing
Carrier testing
Prenatal Diagnosis
Family study
Family mutation
Bank
Other (specify) _____

Sample information:

Date obtained (YYYY-MM-DD): _____ - _____ - _____
Your Referring Laboratory Reference #: _____
Blood (Room Temp.): 1 Full EDTA tube (min. 4mL) (purple)
0.5-3 mL EDTA- Infant/Newborn
Amniotic fluid: 10 mL minimum
CVS: 10 mg minimum
Cultured cells: 1-2 confluent T25 flask(s) required
DNA: 10 ug minimum (in low TE (pH8.0))
Other (specify) _____

If expedited testing is requested, please indicate reason:

- Pregnancy (gestational age: _____ weeks)
Other (specify): _____

Clinical diagnostics and family history:

(Please provide relevant information below, including the names of any relatives previously tested). Please provide a pedigree, if possible.

Laboratory Use:

Date/Time Received (YYYY-MM-DD): _____ h
Lab #: _____
Specimen type, amt & # of tubes: _____
Comments:
Pedigree No. / Patient No. _____ / _____

Ethnicity: _____

Ordering checklist:

- Completed requisition
Specimen tube labeled (with at least two identifiers)
Complete Clinical datasheet (if applicable)
Tay Sachs requisition (if applicable)

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Sex

Molecular Genetics TESTING

22q11 Deletion Syndrome gene dosage

Angelman Syndrome

- methylation and copy number
- UPD 15 analysis (*please send parental samples*)

AR Hereditary Spastic Paraplegia panel*

- Panel A
 - SPG11 gene sequencing
 - SACS gene sequencing
 - CYP7B1 gene sequencing
- Panel B
 - SPG15 gene sequencing
 - SPG7 gene sequencing
 - PNPLA6 s gene sequencing
- Panel C
 - SPG 20 gene sequencing
 - SPG21 gene sequencing
 - CCT5 gene sequencing
- Familial mutation

Arrhythmogenic Right Ventricular Atrophy*

- Full panel
 - DSC2 gene sequencing
 - DSG2 gene sequencing
 - DSP gene sequencing
 - PKP2 gene sequencing
 - TMEM43 gene sequencing
- Familial mutation

Ashkenazi Jewish Screening panel

Atypical Hemolytic Uremic Syndrome /

Membranoproliferative Glomerulonephritis*

- Panel A
 - CD46 gene sequencing
 - CFB gene sequencing
 - CFH gene sequencing
 - CFHR5 gene sequencing
 - CFI gene sequencing
- Panel B
 - C3 gene sequencing
 - THBD gene sequencing
 - APLN gene sequencing
- Familial mutation

Batten disease

- Recurrent mutation panel
- Full Panel
 - CLN1 gene sequencing
 - CLN2 gene sequencing

- CLN3 gene sequencing
- CLN5 gene sequencing
- CLN6 gene sequencing
- CLN7 gene sequencing
- CLN8 gene sequencing
- CLN10 gene sequencing
- Familial mutation

Beckwith-Wiedemann Syndrome

- methylation and copy number
- CDKN1C gene sequencing
- CDKN1C gene sequencing (*if methylation is normal*)
- UPD 11 analysis (*please send parental samples*)
- Familial mutation

Bone Marrow Transplant

- BMT - monitoring

Branchio-Oto-Renal Syndrome

- EYA1 gene sequencing
- EYA1 gene dosage
- Familial mutation

Charge Syndrome (CHD7)

- CHD7 gene sequencing
- CHD7 gene dosage
- Familial mutation

Cherubism*

- SH3BP2 recurrent mutation in exon 9
- SH3BP2 gene sequencing

Congenital Muscular Dystrophy

- Full panel
 - FCMD gene sequencing
 - FKRP gene sequencing
 - POMGnT1 gene sequencing
 - POMT1 gene sequencing
 - POMT2 gene sequencing
- Familial mutation

Craniosynostosis

- Apert/Pfeiffer Syndrome (*recurrent mutation in FGFR1 gene*)
- Crouzon Syndrome (*select exons of FGFR2 and FGFR3 gene*)
- Saethre-Chotzen Syndrome (*TWIST seq & select exons in FGFR3*)
- Craniosynostosis Non-Syndromic (*select exons of FGFR3 gene*)
- Gene dosage for FGFR2, FGFR3 & TWIST

Cystic Fibrosis

- CFTR mutation panel
- CFTR gene sequencing
- CFTR gene dosage
- Familial mutation

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Sex

Molecular Genetics TESTING

Dopamine beta-hydroxylase deficiency

- DBH gene sequencing

Duchenne Muscular Dystrophy

- DMD gene dosage
- DMD gene sequencing
- DMD gene mRNA analysis (*contact the laboratory*)
- Familial mutation

Becker Muscular Dystrophy

- DMD gene dosage

Fabry Disease

- GLA gene sequencing
- GLA gene dosage
- GLA gene mRNA analysis (*contact the laboratory*)
- Familial mutation

Focal Segmental Glomerulosclerosis*

- Full panel
 - ACTN4 gene sequencing
 - CD2AP gene sequencing
 - NPHS1 gene sequencing
 - NPHS2 gene sequencing
 - TRPC6 gene sequencing
- Familial mutation

Fragile X (FMR1)

Fragile X E (FMR2)

Gaucher Disease (recurrent mutations)

Hearing Impairment

- Full panel
 - Non-syndromic genes
 - GJB2 gene sequencing and GJB6 deletion
 - GJB2 gene sequencing only
 - Aminoglycoside-Induced or Maternally Inherited Hearing Loss (MTRNR1 (12s rRNA), MTT51 (tRNA-SER))
 - Pendred Syndrome/Hearing loss with EVA
 - SLC26A4 gene sequencing
 - SLC26A4 gene dosage
- Familial mutation

Hereditary Hemorrhagic Telangiectasia

- ACVRL1 gene sequencing
- ENG gene sequencing
- SMAD4 gene sequencing
- ACVRL1 & ENG gene dosage
- Familial mutation

Hunter Disease

- IDS gene sequencing
- IDS gene dosage analysis
- IDS gene mRNA analysis (*contact the laboratory*)
- Familial mutation

Identity studies

- Maternal cell contamination studies
- Zygoty studies

Li-Fraumeni Syndrome*

- TP53 gene sequencing
- TP53 gene dosage
- Familial mutation

Prader-Willi Syndrome

- methylation and copy number
- UPD 15 analysis (*please send parental samples*)

Russel Silver Syndrome

- H19 methylation & copy number
- UPD 7 analysis (*please send parental samples*)

Schwachman-Diamond Syndrome

- SBDS gene sequencing

Simpson-Golabi-Behmel Syndrome

- GPC3 gene sequencing
- GPC3 & GPC4 gene dosage
- Familial mutation

Skeletal Dysplasia

- Achondroplasia (*recurrent mutation in FGFR3*)
- Hypochondroplasia (*recurrent mutation in FGFR3*)
- Thanatophoric dysplasia (*recurrent mutation in FGFR3*)

Spinal and Bulbar Muscular Atrophy (SBMA) (AR gene)

Spinal Muscular Atrophy (SMA)

- SMN1 & SMN2 gene dosage

Trismus-Pseudocamptodactyly (TRISM)

- MYH8 gene sequencing

X Inactivation Analysis

Other:

* Clinical datasheet required. Please contact the laboratory to obtain this form.



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LABORATORY TESTING: Billing Form

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

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At your direction, we will bill the hospital, referring laboratory, referring physician, or a patient/guardian, for the services we render

- Invoices are sent upon completion of each test/service.
• Invoices are itemized and include the date of service, patient name, CPT code, test name and charge.
• Contact SickKids' Molecular Genetics Laboratory at (416) 813- 7200 x1 with billing inquiries.

How to complete the Billing Form:

- Referring Physician completes the appropriate section below to specify billing method.
• Send requisition and completed "Billing Form" with specimen.

Section 1: Complete to have the Healthcare Provider billed:

Your 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: _____

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

If you elect to have patient/guardian billed:

- Patient/Guardian billing information below must be complete; otherwise, the healthcare provider will be billed.
• Please advise the patient/guardian to expect a bill from our laboratory.
• Provide us with patient's valid credit card information.
• Unfortunately, we cannot accept personal checks.
• In this case, the patient/guardian is solely responsible for the charges.

Send bill to (check one):

- Patient Guardian
 American Express Master Card Visa

Method of Payment (check one):

Name as it appears on credit card: _____
Credit card #: _____
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: () _____ - _____