



THE HOSPITAL FOR
SICK CHILDREN

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)

Patient Name: _____

Date of Birth (DD/MM/YYYY): _____

Gender: Male Female

MRN: _____

Parent's Name: _____

Address: _____

Telephone #: _____

For Canada Only (Billing section must be completed for all non-OHIP)

Provincial Health Card #: _____

Version: _____

Issuing Province: _____

Genome Diagnostics

www.sickkids.ca/genome-diagnostics

Referring Physician:

Name: _____

Facility/Ward/Clinic (required): _____

Address: _____

Phone _____ Fax _____

Email address: _____

Signature (required) _____

Copy Report To (if selected, all below information is required):

Name: _____

Address: _____

Phone _____ Fax _____

Sample Information:

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

Referring laboratory reference #: _____

- Blood in EDTA (purple top tube): min. 4 mL (0.5-3 mL for newborns)
- DNA: min. 10 ug in low TE buffer (Source: _____)
- Direct CVS: min. 10 mg direct villi
- Cultured villi: 1-2 confluent T25 flasks
- Cultured amniocytes: 1-2 confluent T25 flasks
- Tissue (Source: _____)
- Other (Specify: _____)

Closed consent:

- (If checked, all remaining DNA will be discarded upon notification by the ordering physician that all DNA testing has been completed)

Laboratory Use:

Date (DD/MM/YYYY) | Time Received: _____ - _____ - _____ | _____ h

Lab #: _____

Specimen type, amt & # of tubes: _____

Comments: _____

Pedigree No. / Patient No. _____ / _____

Test request (write below and/ or check box(es) on pages 2 and 3):

Reason for Testing:

- Diagnosis Carrier testing
- Familial mutation/variant analysis Prenatal testing
- Bank DNA only
- Other (Specify): _____

If expedited testing is requested, indicate reason

- Pregnancy (Gestational age (weeks): _____)
- Other (Specify: _____)

Familial Mutation/Variant Analysis:

For prenatal testing and cases 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: _____

Relationship to proband/fetus: _____

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)
Clinical information must be provided on pages 4 -5 for all Next-Generation Sequencing tests. Testing will not proceed until these are provided.
- 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, as this is important for accurate interpretation of results

Ethnicity: _____

Genome Diagnostics

LIST OF TESTS AVAILABLE BY DISEASE

For prenatal testing and cases where a familial mutation/variant is known, please include information on page 1.

22q11 Deletion Syndrome

- 22q11 deletion/duplication analysis

Angelman Syndrome

- Methylation and deletion/duplication analysis
 UPD15 analysis (*submit parental samples*)

Arrhythmic Right Ventricular Cardiomyopathy

- Sequence analysis panel:
DSC2, DSG2, DSP, PKP2, TMEM43

Ashkenazi Jewish Carrier Screening

Use: Carrier Screening: Tay-Sachs Enzyme Testing & Ashkenazi Jewish Molecular Panel Requisition

Atypical Hemolytic Uremic Syndrome / Membranoproliferative Glomerulonephritis

- Sequence analysis panel

Autoinflammatory Disease *

Clinical information must be provided on pages 4 and 5

- Recurrent Fever Syndrome (RFS) NGS panel
 Hemophagocytic Lymphohistiocytosis (HLH) NGS panel
 Deletion/duplication analysis

Becker Muscular Dystrophy

- DMD* Sequence analysis
 DMD deletion/duplication analysis

Beckwith-Wiedemann Syndrome

- IC1 and IC2 methylation and 11p15 deletion/duplication analysis
 UPD11 analysis
 CDKN1C Sequence analysis

Bone Marrow Transplantation

- Post-transplant monitoring

Branchio-Oto-Renal Syndrome

- EYA1* Sequence analysis
 EYA1 deletion/duplication analysis

Caffey Disease

- COL1A1* recurrent mutation analysis

Cancer Related Tests

Li-Fraumeni Syndrome

- TP53* Sequence analysis
 TP53 deletion/duplication analysis

Rhabdoid Tumour Predisposition Syndrome

- SMARCB1* Sequence analysis
 SMARCB1 deletion/duplication analysis

Charge Syndrome

- CHD7* sequence analysis
 CHD7 deletion/duplication analysis

Cherubism

- SH3BP2* recurrent mutation analysis
 SH3BP2 sequence analysis

Congenital Muscular Dystrophies

- Sequence analysis panel:
FKTN (FCMD), FKRP, POMGnT1, POMT1, POMT2

Connective Tissue Disease *

Clinical information must be provided on pages 4 and 5

If more than one panel is requested, rationale must be provided on page 5.

- Ehlers Danlos Syndrome NGS panel
 Osteogenesis Imperfecta NGS panel
 Osteopetrosis and Disorders of Increased Bone Density NGS panel
 Bone Involvement NGS panel
 Deletion/duplication analysis

Craniosynostosis

- Apert Syndrome (*FGFR2* recurrent mutations analysis)
 Crouzon Syndrome (*FGFR2, FGFR3* recurrent mutation analysis)
 Pfeiffer Syndrome (*FGFR1, FGFR2, FGFR3* recurrent mutation analysis)
 Saethre-Chotzen Syndrome (*TWIST1* sequence analysis and *FGFR3* recurrent mutation analysis)
 Non-Syndromic Craniosynostosis (*FGFR3* recurrent mutation analysis)
 TWIST1 deletion/duplication analysis

Cystic Fibrosis

For prenatal echogenic bowel, ensure parental samples are linked to each other on both requisitions with at least two identifiers. Ensure "echogenic bowel" is indicated on the Family History section on Page 1.

- CFTR* recurrent mutation analysis
 CFTR Sequence analysis
 CFTR deletion/duplication analysis

Dopamine Beta-Hydroxylase Deficiency

- DBH* Sequence analysis

Duchenne Muscular Dystrophy

- DMD* Sequence analysis
 DMD deletion/duplication analysis
 DMD mRNA analysis (*contact the laboratory before ordering*)

Fabry Disease

- GLA* Sequence analysis
 GLA deletion/duplication analysis
 GLA mRNA analysis (*contact the laboratory before ordering*)

Focal Segmental Glomerulosclerosis

- Sequence analysis panel

Fragile X Syndrome

- FMR1* trinucleotide repeat analysis

Patient Name:

Date of Birth (DD/MM/YYYY):

Gender: Male Female

MRN:

Genome Diagnostics

LIST OF TESTS AVAILABLE BY DISEASE

For prenatal testing and cases where a familial mutation/variant is known, please include information on page 1.

Fragile X E Syndrome **

- AFF2 trinucleotide repeat analysis
(See testing requirements)

Gaucher Disease

- GBA recurrent mutation analysis

Hearing Loss: Non-Syndromic, Autosomal Recessive

- GJB2 Sequence analysis
 GJB6 deletion/duplication analysis

Hearing Loss: Pendred Syndrome

- SLC26A4 Sequence analysis
 SLC26A4 deletion/duplication analysis

Hereditary Hearing Loss *

Clinical information must be provided on pages 4 and 5

When the Common and Non-syndromic Hearing Loss Panel is requested, STRC dosage is tested.

- Common and Non-syndromic Hearing Loss NGS panel
 Usher Syndrome NGS panel
 Stickler Syndrome NGS panel
 Alport Syndrome, Norrie Syndrome, Treacher Collins Syndrome, Waardenburg Syndrome NGS panel
 Deletion/duplication analysis

Hereditary Hemorrhagic Telangiectasia

- ACVRL1 Sequence analysis
 ENG Sequence analysis
 ACVRL1 and ENG deletion/duplication analysis
 SMAD4 Sequence analysis

Hereditary Spastic Paraplegia *

Clinical information must be provided on pages 4 and 5

- Autosomal Dominant HSP NGS panel
 Autosomal Recessive HSP NGS panel
 X-Linked HSP NGS panel
 Deletion/duplication analysis

Hunter Disease

- IDS Sequence analysis
 IDS deletion/duplication analysis
 IDS mRNA analysis (contact the laboratory before ordering)

Identity Testing

- Zygosity studies

- Maternal Cell Contamination Studies
(maternal sample required)

Neurofibromatosis type 1/Legius syndrome *

Clinical information must be provided on pages 4 and 5

- NF1 sequence analysis
 NF1 deletion/duplication analysis
 SPRED1 sequence analysis
 SPRED1 deletion/duplication analysis

Neuronal Ceroid Lipofuscinoses (Batten Disease)

- PPT1 (CLN1), TPP1 (CLN2) and CLN3 recurrent mutation analysis
 Sequence analysis panel:
PPT1(CLN1), TPP1 (CLN2), CLN3, CLN5, CLN6, CLN7, CLN8, CLN10

Noonan Syndrome and RASopathies *

Clinical information must be provided on pages 4 and 5

- Noonan Syndrome and RASopathies panel
 Deletion/duplication analysis for SPRED1 only

Prader-Willi Syndrome

- Methylation and deletion/duplication analysis
 UPD15 analysis (parental samples required)

Russell-Silver Syndrome

- IC1 methylation and 11p15 deletion/duplication analysis
 UPD7 analysis (parental samples required)

Shwachman-Diamond Syndrome

- SBDS Sequence analysis

Simpson-Golabi-Behmel Syndrome

- GPC3 sequence analysis and GPC3 and GPC4 deletion/duplication analysis

Skeletal Dysplasia

- Achondroplasia (FGFR3 recurrent mutation analysis)
 Hypochondroplasia (FGFR3 recurrent mutation analysis)
 Thanatophoric Dysplasia (FGFR3 recurrent mutation analysis)

Spinal and Bulbar Muscular Atrophy

- AR trinucleotide repeat analysis

Spinal Muscular Atrophy

- SMN1 and SMN2 deletion/duplication analysis

Trismus Pseudocamptodactyly Syndrome

- MYH8 Sequence analysis

X-Inactivation Analysis

- Other: _____

* Next-Generation Sequencing (NGS) testing will only be initiated if the clinical information sections, located on pages 4 and 5 of the requisition form, are completed. For more information on our Next-Generation Sequencing (NGS) panels, including the list of genes tested, please visit our website: www.sickkids.ca/genome-diagnostics

** For information on the testing requirement for Fragile X E, please visit the Specimen Requirements section for Fragile X E Syndrome on our website: www.sickkids.ca/genome-diagnostics/FragileXE

Genome Diagnostics

Clinical Information (Required)

DISEASE SPECIFIC FEATURES

Autoinflammatory Disorders (RFS/HLH)

- Abnormal inflammatory response
- Fevers
- Arthritis
- Pulmonary complications
- Gastrointestinal irritation
- Hepatosplenomegaly
- Lymphadenopathy
- Hemophagocytosis
- Oral ulcers
- Rash, specify: _____
- Ocular inflammation specify: _____
- Edema (periorbital, optic disk)
- Vision loss
- Other: _____

Hearing Loss

- Age of onset: _____
- Sensorineural hearing loss
- Conductive hearing loss
- Mixed hearing loss
- Bilateral Unilateral
- Syndromic Non-syndromic
- Ear anomalies Ear tags
- Eye anomalies Renal anomalies
- White forelock Cardiac anomalies
- Hirschsprung disease
- Other: _____

Hereditary Spastic Paraplegia (HSP)

- Abnormal corpus callosum
- Cognitive impairment
- Ataxia Spasticity
- Hyperreflexia Seizures
- Hypertonia Hypotonia
- Dystonia Dysarthria
- Extensor plantar reflex
- Other: _____

The following investigations are required before molecular testing of HSP is undertaken:

- MRI – Brain and spinal cord
- Biochemical testing - Vitamin B12, vitamin E, very long chain fatty acids, lysosomal work-up, plasma amino acids and serum lipoprotein analysis (as appropriate)

Neurofibromatosis type 1 (NF1) / Legius Syndrome

- The patient meets the NIH criteria for a clinical diagnosis of NF1 (≥2 of the clinical features below).**
- Café-au-lait macules ≥6 CALS (#: _____)
- Neurofibromas, ≥ 2 or ≥ 1 Plexiform
- Freckling, axillary or inguinal
- Optic glioma
- ≥2 Lisch nodules (iris hamartomas)
- Osseous lesion (type: _____)
- First degree relative diagnosed with NF1 by above criteria
- Other: _____
- The patient does not meet the NIH diagnostic criteria for NF1. Rationale for testing must be provided on page 5.**

Connective Tissue Disorders (CTD)

Ehlers Danlos Syndrome (EDS)

Indicate the suspected clinical diagnosis in the patient:

- Classic Vascular
- Kyphoscoliotic Other: _____

Note: Genetic testing is not offered for joint hypermobility alone. If testing is requested for joint hypermobility, provide rationale on page 5.

Check applicable CTD features below.

Osteopetrosis and Disorders of Increased Bone Density

Check applicable CTD features below.

CTD Related Clinical Features:

- Joint hypermobility: Beighton score: _____
- Arterial aneurysms, dissection or rupture
- Intestinal rupture
- Molluscoid pseudotumors
- Subcutaneous spheroids
- Loose/stretchable skin
- Smooth/velvety skin
- Widened atrophic scars
- Recurrent spontaneous tendon rupture

Osteogenesis Imperfecta (OI)

If the patient does not present with one of the test indications below, rationale for testing must be provided on page 5.

- Fetal findings on anatomy ultrasound consistent with OI.
- Fractures with minimal or no trauma in the absence of other known disorders of bone metabolism.
- Vertebral fractures
- Dentinogenesis imperfecta
- Low ALP for age/gender (ALPL analysis only will be performed)

Check applicable CTD features below.

Bone Involvement

Check applicable CTD features below.

- Easy bruising
- Myopia
- Lens dislocation
- Blue/gray sclerae
- Thumb or wrist sign
- Club foot
- Scoliosis
- Marfanoid habitus
- Short stature
- Shortened long bones
- Recurrent pneumothoraces
- Joint subluxations/dislocations
- Fractures
- Bone deformity
- Wormian bones
- Increased bone mineral density
- Diaphyseal sclerosis
- Hearing loss
- Osteosclerosis
- Other: _____

Noonan Syndrome and RASopathies

- Increased nuchal translucency
- Developmental delay
- Characteristic facies
- Broad or webbed neck
- Heart defect (specify: _____)
- Hypertrophic cardiomyopathy
- Short stature (%ile: _____)
- Pectus deformity
- Lymphatic dysplasias
- Characteristic hematological abnormality (specify: _____)
- Other RASopathy features: (specify: _____)
- For postnatal patients: The patient must present with ≥2 of the above features for molecular testing to be undertaken.**

FAMILY HISTORY

Please draw or attach a pedigree and provide any relevant information below, including clinical and family history details, as this is important for accurate interpretation of results.

Ethnicity: _____

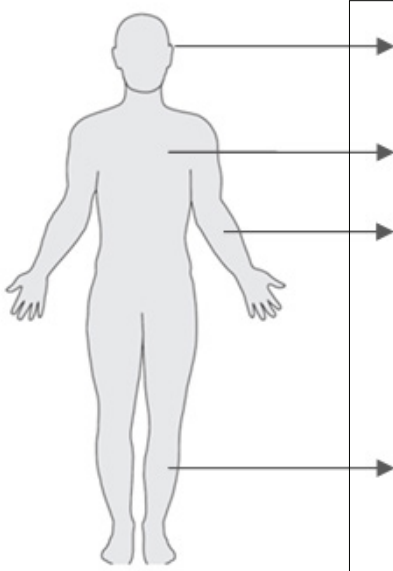
Genome Diagnostics

Clinical Information (Required)

ADDITIONAL RELEVANT CLINICAL INFORMATION

Previous Genetic Testing

- No
 Yes – Test Results



GENERAL CLINICAL FEATURES

Perinatal history

- Premature birth
 IUGR
 Oligohydramnios Polyhydramnios
 Other: _____

Growth

- Failure to thrive
 Growth retardation/short stature
 Overgrowth
 Macrocephaly Microcephaly
 Other: _____

Physical/cognitive development

- Delayed fine motor development
 Delayed gross motor development
 Delayed speech and language
 Autistic behavior
 Intellectual disability
 Developmental regression
 Other: _____

Behavioral

- Autistic features
 Obsessive-compulsive disorder
 Other psychiatric symptoms
 Other: _____

Cancer/Malignancy

- Age of onset: _____
 Tumor type: _____
 Location(s): _____

Craniofacial/Ophthalmologic

- Abnormal face shape
 Blindness Cataracts
 Coloboma Optic atrophy
 Ophthalmoplegia Ptosis
 Retinitis pigmentosa
 Oral cleft
 Other: _____

Brain malformations/ abnormal imaging

- Abnormality of the basal ganglia
 Agenesis of the corpus callosum
 Brain atrophy
 Cortical dysplasia
 Hemimegalencephaly
 Heterotopia
 Holoprosencephaly
 Hydrocephalus
 Lissencephaly
 Periventricular leukomalacia
 Other: _____

Cardiac/congenital heart malformations

- ASD VSD
 Coarctation of aorta
 Hypoplastic left heart
 Tetralogy of Fallot
 Cardiomyopathy
 Arrhythmia/conduction defect
 Other: _____

Gastrointestinal

- Gastroschisis/omphalocele
 Gastrointestinal reflux
 Pyloric stenosis
 Tracheoesophageal fistula
 Hepatic failure
 Chronic intestinal pseudo-obstruction
 Hirschsprung disease
 Recurrent vomiting
 Chronic diarrhea
 Constipation
 Other: _____

Genitourinary abnormalities

- Ambiguous genitalia
 Cryptorchidism
 Hypospadias
 Hydronephrosis
 Kidney malformation
 Renal agenesis
 Proximal renal tubulopathy
 Other: _____

Endocrine

- Diabetes mellitus Type 1
 Diabetes mellitus Type 2
 Hypothyroidism
 Hypoparathyroidism
 Pheochromocytoma/paraganglioma
 Other: _____

Neurological/Muscular

- Ataxia Hypotonia
 Chorea Hypertonia
 Dystonia Spasticity
 Exercise intolerance/ easy fatigue
 Headache/migraine
 Muscle weakness
 Seizures (type: _____)
 Stroke/stroke-like episodes
 Other: _____

Skeletal/Limb abnormalities

- Contractures Club foot
 Polydactyly Syndactyly
 Vertebral anomaly Scoliosis
 Other: _____

Skin/Hair

- Abnormality of the hair pattern,
quantity
 Abnormal nail growth
 Abnormal pigmentation
 Café-au-lait macules
 Neoplasms of the skin
 Neurofibromas
 Blistering
 Ichthyosis
 Other: _____

Patient Name: _____

Date of Birth (DD/MM/YYYY): _____

Gender: Male Female

MRN: _____

Genome Diagnostics

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

BILLING FORM

The hospital, referring laboratory, or a patient/guardian will be billed for the services rendered.

- Invoices are sent upon completion of each test/service.
- Contact SickKids' Genome Diagnostics 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.

Option 1: Complete to have the Healthcare Provider billed:

Your Referring Laboratory's Reference #: _____
 Billing address of hospital, referring laboratory:
 Name: _____
 Address: _____
 City: _____ Prov/State: _____
 Postal/Zip Code: _____ Country: _____
 Contact Name: _____
 Contact Telephone #: _____

Option 2: Interim Federal Health Program (IFHP)

Submit a copy of the Interim Federal Health Certificate (Refugee Protection Claimant Document) with the photo and UCI# visible for coverage to be confirmed.

UCI# _____
 ICD code (lab use only): _____

Option 3: 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.**

Relation to patient (check one): Patient Guardian/Parent

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

Name as it appears on credit card: _____

Credit card #: _____

Expiry date on credit card: _____

CVS#- found on back of card (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: _____
