SickKids
THE HOSPITAL FOR SICK CHILDREN
Paediatric Laboratory Medicine

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Genome Diagnostics

www.sickkids.ca/dplm

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:

RNA SEQUENCING

Ordering Physician:

Name: _____

Institution/Facility/Ward/Clinic: _____

Address: _____

_____

Phone _____ Fax _____

Email address: _____

Signature *(required)* _____

Copy Report To:

Name: _____

Institution: _____

Address: _____

Phone _____ Fax _____

Sample Information:

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

Referring laboratory reference #: _____

☐ Cultured Fibroblast (1-2 T25 flask)
☐ Cultured Fibroblast (1x 10*6 cells pelleted frozen on dry ice
☐ Tissue frozen (-80 C min 5-10 mg required) (Source: _____)
☐ Other (please contact lab prior to collecting) _____

Bone Marrow Transplant/Transfusion

Has the patient undergone bone marrow transplant? ☐ Yes ☐ No
Date of bone marrow transplant (DD-MM-YYYY):
Testing for patients who have received an allogenic bone marrow trans-plant must be completed on a pre-transplant sample or a non-hematologic sample

Has the patient received a blood transfusion? ☐ Yes ☐ No
Date of last transfusion (DD-MM-YYYY):
Blood obtained for genetic testing should ideally be collected at least 2-4 weeks after the date of the last transfusion

The specimen should be shipped on a Monday, Tuesday or Wednesday. If there is a delay in the shipping of specimen (i.e. >48 hours), the sample should be placed in the refrigerator and shipped to Genome Diagnostics Laboratory on ice. Please call to inform us when the samples are being sent

RNA Sequencing submission requirements:

Consent:

The test has been discussed with the patient.

Clinical information:

The following information has been provided for the patient on pages 2-4:

- Phenotypic information (PhenoTips or Clinical data sheet)
- Family history (pedigree)
- Previous testing history
- Relevant clinic note(s) and/or letter(s)

☐ Full RNA Sequencing

☐ RNA sequencing for targeted analysis

Complete below and attach a copy of proband's report:

Gene & NM#: _____

Mutation/variant(s): _____

SickKids Laboratory/Order Number: _____

SickKids Pedigree/Family Number: _____

Laboratory Use:

Date (DD/MM/YYYY) | **Time Received**:

_____ - _____ - _____ | _____ h

Order #: _____

Specimen type, amt & # of tubes: _____

Comments:

Proband name: _____ MRN: _____ DOB: _____

CLINICAL DATA SHEET

Previous genetic testing:

- ☐ Single gene/Gene panel (1): _____
  Result: _____
- ☐ Single gene/Gene panel (2): _____
  Result: _____
- ☐ Microarray: _____
- ☐ Other: _____
  Result: _____

Pre/Perinatal History

- ☐ Cystic hygroma
- ☐ Increased nuchal translucency
- ☐ Intrauterine growth retardation
- ☐ Nonimmune hydrops fetalis
- ☐ Oligohydramnios
- ☐ Polyhydramnios
- ☐ Prematurity GA: _____
- ☐ Other: _____

Growth:

- ☐ Growth delay
- ☐ Overgrowth
- ☐ Failure to thrive
- ☐ Hemihypertrophy
- ☐ Short stature
- ☐ Tall stature

Structural Brain Abnormalities

- ☐ Abnormal myelination
- ☐ Abnormality of basal ganglia
- ☐ Abnormality of brainstem
- ☐ Abnormality of periventricular white matter
- ☐ Abnormality of the corpus callosum
- ☐ Aplasia/hypoplasia of cerebellar vermis
- ☐ Aplasia/hypoplasia of cerebellum
- ☐ Cerebellar atrophy
- ☐ Chiari malformation
- ☐ Cortical dysplasia
- ☐ Encephalocele
- ☐ Heterotopia
- ☐ Hemimegalencephaly
- ☐ Holoprosencephaly
- ☐ Hydrocephalus
- ☐ Leukodystrophy
- ☐ Lissencephaly
- ☐ Pachygyria
- ☐ Polymicrogyria
- ☐ Ventriculomegaly
- ☐ Other: _____

Developmental/Behavioral

- ☐ Aggressive behavior
- ☐ ADHD
- ☐ Anxiety
- ☐ Autistic Behavior
- ☐ Autism spectrum disorder
- ☐ Cognitive impairment
- ☐ Delayed speech & language development
- ☐ Developmental regression
- ☐ Fine motor delay
- ☐ Gross motor delay
- ☐ Speech delay
- ☐ Gait disturbance
- ☐ Global developmental delay
- ☐ Hyperactivity
- ☐ Incoordination
- ☐ Intellectual disability
  - ☐ Mild ☐ Profound
  - ☐ Moderate ☐ Severe
- ☐ Learning disability
- ☐ Memory impairment
- ☐ Obsessive-compulsive disorder
- ☐ Sleep disturbance
- ☐ Stereotypy

Neurological

- ☐ Ataxia
- ☐ Chorea
- ☐ Cortical Visual Impairment
- ☐ Dementia
- ☐ Dysarthria
- ☐ Dyskinesia
- ☐ Dysphasia
- ☐ Dystonia
- ☐ Encephalopathy
- ☐ Headaches
- ☐ Hemiplegia
- ☐ Infantile Spasms
- ☐ Migraines
- ☐ Myoclonus
- ☐ Myopathic facies
- ☐ Myopathy
- ☐ Muscle weakness
- ☐ Muscle dystrophy
- ☐ Neuropathy
  - ☐ Motor ☐ Sensory ☐ Sensorimotor
- ☐ Parkinsonism
- ☐ Seizures
- ☐ Spasticity
- ☐ Tremors

Craniofacial dysmorphic features

- ☐ Craniosynostosis
  Specify: _____
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Head shape Specify: _____
- ☐ Facies Specify: _____
- ☐ Forehead Specify: _____
- ☐ Ears Specify: _____
- ☐ Eyes Specify: _____
- ☐ Nose Specify: _____
- ☐ Cleft lip and/or palate
- ☐ Coarse facial features
- ☐ Short neck
- ☐ Synophrys
- ☐ Other: _____

Ophthalmological

- ☐ Anophthalmia
- ☐ Cataracts
- ☐ Coloboma
- ☐ Corneal opacity
- ☐ Ectopia lentis
- ☐ External ophthalmoplegia
- ☐ Microphthalmia
- ☐ Myopia
- ☐ Nystagmus
- ☐ Optic atrophy
- ☐ Ptosis
- ☐ Retinal detachment
- ☐ Retinitis pigmentosa
- ☐ Strabismus
- ☐ Other: _____

Hearing Impairment

- ☐ Abnormal Newborn Screen: _____
- ☐ Conductive hearing impairment
- ☐ Sensorineural hearing impairment

Haematological or Immunologic

- ☐ Anemia
- ☐ Coagulation disorder
- ☐ Immunodeficiency
- ☐ Neutropenia
- ☐ Pancytopenia
- ☐ Recurrent infections
- ☐ Thrombocytopenia
- ☐ Other: _____

Integumental

Skin

- ☐ Abnormal blistering of the skin
- ☐ Anhidrosis
- ☐ Café-Au-Lait macules
- ☐ Cutis laxa
- ☐ Hemangiomas
- ☐ Hyperpigmentation of the skin
- ☐ Hypopigmentation of the skin
- ☐ Ichthyosis
- ☐ Skin rash
- ☐ Telangiectasia
- ☐ Vascular skin abnormality
- ☐ Other: _____

Hair

- ☐ Abnormal texture, distribution, colour, whorls
  Specify: _____
- ☐ Alopecia
- ☐ Coarse hair
- ☐ Sparse hair
- ☐ Other: _____

Dental

- ☐ Specify: _____

Nails

- ☐ Specify: _____

Proband name: _____ MRN: _____ DOB: _____

CLINICAL DATA SHEET

Cardiac
- ☐ Aortic root dilation
- ☐ Arrhythmia / Conduction defect
  - ☐ Bradycardia
  - ☐ Prolonged QTc interval
  - ☐ Ventricular tachycardia
- ☐ Cardiomyopathy
  - ☐ Dilated
  - ☐ Hypertrophic
  - ☐ Noncompaction
- ☐ Congenital heart defect
  - ☐ Atrial septal defect
  - ☐ Bicuspid aortic valve
  - ☐ Coarctation of aorta
  - ☐ Hypoplastic left heart
  - ☐ Patent ductus arteriosis
  - ☐ Patent foramen ovale
  - ☐ Tetralogy of Fallot
  - ☐ Ventricular septal defect
- ☐ Heterotaxy
- ☐ Mitral valve prolapse
- ☐ Sudden death
- ☐ Syncope
- ☐ Other: _____

Endocrine
- ☐ Early puberty
- ☐ Delayed puberty
- ☐ Diabetes Insipidus
- ☐ Diabetes mellitus
- ☐ Hyperparathyroidism
- ☐ Hypoparathyroidism
- ☐ Hyperthyroidism
- ☐ Hypothyroidism
- ☐ Hypogonadism
- ☐ Hypophosphatemia
- ☐ Rickets
- ☐ Other: _____

Gastrointestinal
- ☐ Chronic intestinal pseudo-obstruction
- ☐ Duodenal stenosis/atresia
- ☐ Diaphragmatic hernia
- ☐ Elevated transaminases
- ☐ Exocrine pancreatic insufficiency
- ☐ Feeding difficulties
- ☐ Gastroesophageal reflux
- ☐ Hepatomegaly
- ☐ Hepatic failure
- ☐ Hirschsprung disease
- ☐ Inflammatory bowel disease
- ☐ Intrahepatic biliary atresia
- ☐ Laryngomalacia
- ☐ Omphalocele
- ☐ Pyloric stenosis
- ☐ Splenomegaly
- ☐ Tracheoesohageal fistula
- ☐ Other: _____

Genitourinary
- ☐ Ambiguous genitalia
- ☐ Cryptorchidism (undescended testes)
- ☐ Cystic renal dysplasia
- ☐ Horseshoe kidney
- ☐ Hydronephrosis
- ☐ Hypospadias
- ☐ Inguinal hernia
- ☐ Infertility
- ☐ Micropenis
- ☐ Nephrolithiasis
- ☐ Polycystic kidney disease
- ☐ Renal agenesis or dysgenesis
- ☐ Renal tubulopathy
- ☐ Other: _____

Musculoskeletal
- ☐ Abnormal connective tissue
- ☐ Abnormal form of the vertebral bodies
- ☐ Abnormality of the digits
  - ☐ Arachnodactyly ☐ Polydactyly
  - ☐ Clinodactyly ☐ Syndactyly
  - ☐ Ectrodactyly
- ☐ Abnormality of the limb(s)
  Specify: _____
- ☐ Abnormality of the ribs
- ☐ Arthralgia
- ☐ Arthrogryposis
- ☐ Contractures
- ☐ Decreased muscle mass
- ☐ Exercise intolerance
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Joint hypermobility
- ☐ Myalgia
- ☐ Osteoarthritis
- ☐ Osteopenia
- ☐ Pectus carinatum
- ☐ Pectus excavatum
- ☐ Recurrent fractures
- ☐ Scoliosis
- ☐ Skeletal dysplasia
- ☐ Other: _____

Respiratory
- ☐ Bronchiectasis
- ☐ Pneumothorax
- ☐ Pulmonary fibrosis
- ☐ Respiratory insufficiency
- ☐ Other: _____

Tumour / Malignancy
Type: _____
Location: _____
Age of onset: _____

Vascular System
- ☐ Angioedema
- ☐ Aneurysm
- ☐ Arterial calcification
- ☐ Arterial dissection
- ☐ Arterial tortuosity
- ☐ Arteriovenous malformation
- ☐ Bruising susceptibility
- ☐ Epistaxis
- ☐ Lymphedema
- ☐ Pulmonary hypertension
- ☐ Stroke

Metabolic
- ☐ Abnormal activity of mitochondrial respiratory chain
- ☐ Abnormal Newborn Screen: _____
- ☐ Elevated CPK
- ☐ Elevated hepatic transaminase
- ☐ Hypoammonemia ☐ Hyperammonemia
- ☐ Hypoglycemia ☐ Hyperglycemia
- ☐ Increased serum pyruvate
- ☐ Ketosis
- ☐ Lactic acidosis
- ☐ Rhabdomyolysis
- ☐ Plasma AA: _____
- ☐ Urine OA: _____
- ☐ Other: _____

Other investigations
(Please provide copy or report if possible)

Echo: _____
EEG: _____
EMG: _____
MRI: _____
Muscle biopsy: _____
Ultrasound: _____
X-ray: _____

Additional clinical findings:
_____
_____
_____
_____
_____
_____

FAMILY HISTORY

Please draw or attach pedigree
- ☐ Consanguinity

Requisition and samples must be accompanied by additional clinical notes

Proband name: _____ MRN: _____ DOB: _____

PATIENT SUMMARY (all sections must be completed)

Phenotypic category
- ☐ Syndromic developmental delay (DD) or intellectual disability (ID)
- ☐ Moderate-severe isolated DD or ID
- ☐ Single system disorder without DD or ID
- ☐ Multisystem disorder without DD or ID
- ☐ Multiple congenital anomalies without DD or ID

Age of onset
- ☐ Prenatal
- ☐ At birth (<12mo)
- ☐ Childhood (1-10yrs)
- ☐ Adolescence (11-17yrs)
- ☐ Adulthood (>18)

Ethnicity (all applicable)
- ☐ Black, African-American, African
- ☐ East Asian ☐ South Asian
- ☐ White ☐ Indigenous
- ☐ French-Canadian
- ☐ Middle Eastern, North African
- ☐ Latino, Hispanic, Spanish
- ☐ Unknown ☐ Other: _____

Previous test history (all applicable)
- ☐ No previous genetic testing
- ☐ Chromosome microarray
- ☐ Single gene test
- ☐ Gene panel (<100 genes)
- ☐ Gene panel (≥100 genes)
- ☐ Targeted testing (e.g. Prader-Willi)
- ☐ Unknown

ATTESTATION

Attestation (must meet all items):

YES NO

☐ ☐ **I confirm that all the following conditions have been met:**
- Detailed phenotypic characterization (physical examination, investigations) has been documented
- Pretest genetic counselling and consent has been completed
- Chromosomal microarray or other previous genomic testing has been completed and does NOT explain the patient's phenotype
- Other causative circumstances (e.g. environmental exposures, injury, and infection) do NOT explain the patient's clinical presentation, based on the most complete clinical history

YES NO

☐ ☐ **I confirm that the patient does NOT have:**
- Isolated mild intellectual disability or learning disabilities
- Isolated non-syndromic autism
- Isolated neurobehavioural disabilities (e.g. attention deficit disorder)
- A phenotype highly specific to a known genetic condition for which an optimized genetic panel exists, or for which all known gene-disease associations could be assessed. If so, then the targeted gene panel should be given priority assuming it is more sensitive (e.g. Noonan spectrum disorders)

YES NO

☐ ☐ **I confirm that I:**
- Practice in the area of genetics (as a geneticist/genetics consultant or in a clinic where a genetic counsellor has been integral to the care of the patient)
- Have expertise in performing a clinical genetics evaluation including family history, genetic-focused medical history and physical examination, and have a critical understanding of the prior genetic evaluations undertaken in the patient
- Have expertise in determining whether clinical RNA sequencing is the test of choice for the specific clinical indication, prioritizing other available tests as appropriate
- Have expertise in providing adequate pre-test counselling, including informed consent for primary and incidental findings
- Have the ability to interpret the results of the clinical RNA sequencing and provide adequate post-test counselling

PROVIDER ATTESTATION

By signing here I attest that the above the information is an accurate and comprehensive summary of this patient's clinical history.

Ordering physician signature: _____ Date: _____

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Tel: 416-813-7200 x1
Fax: 416-813-7732
(CLIA # 99D1014032)

Patient Name:

Date of Birth (DD/MM/YYYY):

Gender: ☐ *Male* ☐ *Female* *MRN:*

Genome Diagnostics

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: Interm 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:

_____