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Genome-wide Sequencing Ontario

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Patient name:

Date of Birth (DD-MM-YYYY):

Gender: ☐ *Male* ☐ *Female* *MRN:*

Address:

Telephone #:

Ontario health card #: *Version:*

GENOME-WIDE SEQUENCING: FAMILY MEMBER

Ordering physician:

Name: _____

Institution: _____

Address: _____

Phone: _____

Fax: _____

Email address: _____

Copy report to:

Name: _____

Institution: _____

Address: _____

Phone _____

Fax _____

Sample information:

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

Your referring laboratory reference #: _____

☐ Blood in EDTA (purple top tube): min. 2 x 4 mL (0.5-3 mL for newborns)

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

☐ Tissue* (Source:_____)

*Please contact the laboratory directly to discuss prior to sample submission

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 transplant 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

For laboratory use only:

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

_____-_____-_____ | _____h

Order #: _____

Specimen type, amt & # of tubes: _____

Comments:

_____

_____

Requested test:

☐ **Genome-Wide Sequencing** (concurrent sequencing with the proband)

☐ **Segregation analysis** (targeted testing for variant(s) of uncertain significance)

Family member sample submitted:

Proband name/MRN: _____

Relationship to proband: _____

Pedigree # (if known): _____

Clinical status:

This individual was clinically examined: ☐ Yes ☐ No

This individual is: ☐ Affected ☐ Unaffected ☐ Unsure

Requisition and samples must be accompanied by relevant clinical note(s) if this individual is affected.

As the correct assignment of biological relationships is required for the accurate interpretation of test results, a separate test may be performed to confirm the stated family relationships. Segregation testing of the received samples may thus reveal misattributed parentage.

Variant(s) requested for segregation analysis (if applicable):

Sequence variants

Gene	cDNA coordinates	Protein coordinates

Copy number variants / Structural variant

Gene/Region	Type of variant	Genomic coordinates

GWS submission requirements:

Consent:
The test has been discussed with the patient, the consent form has been completed, and decisions have been documented on page 5 of the proband's requisition.

Clinical information:
The following information has been provided for this individual (**if affected**):

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

Name: _____ MRN: _____ DOB: _____

CLINICAL DATA SHEET- FAMILY MEMBER (COMPLETE IF AFFECTED)

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: _____

Name: _____ MRN: _____ DOB: _____

CLINICAL DATA SHEET - FAMILY MEMBER (COMPLETE IF AFFECTED)

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