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Genome-wide Sequencing Ontario

www.gsontario.ca

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(CLIA# 99D1014032)

*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. 10 mL (3 mL for newborns)

☐ DNA: min.2 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:**

**Pedigree No. /Patient No.** _____/_____

## Requested test:

☐ **Genome-Wide Sequencing**

☐ **Segregation analysis**

## Family member sample submitted:

**Proband name/MRN:** _____

**Relationship to proband:** _____

**Family # (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.**

## 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
**(to 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
**(to 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**