



CHEO Genetics Diagnostic Laboratory
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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
 Segregation analysis

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.

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
- Cutsis 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 arteriosus
 - 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
- Tracheoesophageal 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
 - Clinodactyly
 - Ectrodactyly
 - Polydactyly
 - Syndactyly
- 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
- Hypoglycemia
- Increased serum pyruvate
- Ketosis
- Lactic acidosis
- Rhabdomyolysis
- Plasma AA: _____
- Urine OA: _____
- Other: _____
- Hyperammonemia
- Hyperglycemia

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