The Rare Disease Discovery Hub - Referral Form

PATIENT INFORMATION					
Last Name:		First Name:		Sex:	☐F ☐M
Address:					
DOB:		PHN:		MRN:	

FAMILY CONTACT INFORMATION			
Name:		Relationship:	
Email:		Phone Number:	

PREVIOUS GENETIC TESTING INFORMATION *(raw data will not be requested unless the patient consents to the study)*			
Type of Testing:	☐ Exome ☐ Panel ☐ Other:	Testing Provider:	
Individuals Tested:	☐ Proband Only ☐ Trio ☐ Other:	ID:	

By signing the form below, I, _____ confirm the following:

1. The patient, named above, has been notified about The Rare Disease Discovery Hub study and is interested in learning about the study in greater detail. They have agreed to release their contact information and are expecting a phone call from a member of the study team.
2. Should the patient agree to participate in the Hub study:
   - 2.1. I will endeavor to set appropriate patient participant expectations, both during or after participation in research activities. Direct benefits to participants might be nil or negligible.
   - 2.2. I will be considered a research collaborator of the Hub and am expected to be involved in evaluating new emerging data or evidence as I am considered an expert on this patient's medical history and current condition.
   - 2.3. I understand that I am responsible for, at my discretion, return of any findings.
   - 2.4. If possible, feasible and desirable, I will attempt to confirm the study's research-grade conclusions in a certified diagnostic laboratory. Any actionable incidental findings in particular will need to be confirmed in a clinical laboratory; the Hub will not arrange clinical testing.
   - 2.5. I will ensure arrangement of an alternate collaborating physician should I leave my current position.
   - 2.6. I have read *The Discovery Hub Protocol* and the *General Guidance for Physicians* documents.
3. If I publish findings generated using Hub resources, I will acknowledge the BCCHRI Rare Disease Discovery Hub and its funding through BCCHF, and, if individual Hub members contributed to a degree compatible with authorship, I will include these persons as co-authors.
4. If I receive data generated from Hub participation I will follow the REB-approved protocol for either 1) securely using and storing this data solely on PHSA/UBC servers or 2) creating, through the Hub, a Data Transfer Agreement prior to sending this data elsewhere.

Referring Physician:					
Last Name:		First Name:		Phone:	
Signature of Referring Physician:					

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CLINICAL INFORMATION (required for all genomic analysis/reanalysis requests)

Pre/Perinatal History
- ☐Cystic hygroma
- ☐Increased nuchal translucency
- ☐Intrauterine growth restriction
- ☐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 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: _____

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: _____

Developmental/Behavioural
- ☐Aggressive behaviour
- ☐ADHD
- ☐Anxiety
- ☐Autistic behaviour
- ☐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 ☐Moderate ☐Severe
- ☐Learning disability
- ☐Memory impairment
- ☐Obsessive-compulsive disorder
- ☐Sleep disturbance
- ☐Stereotypy

Neuromuscular
- ☐Ataxia
- ☐Chorea
- ☐Cortical visual impairment
- ☐Dementia
- ☐Dysarthria
- ☐Dyskinesia
- ☐Dysphagia
- ☐Dystonia
- ☐Encephalopathy
- ☐Headaches
- ☐Hemiplegia
- ☐Infantile spasms
- ☐Migraines
- ☐Myoclonus
- ☐Myopathic facies
- ☐Myopathy
- ☐Muscle weakness
- ☐Muscle dystrophy
- ☐Neuropathy
- ☐Motor ☐Sensory ☐Sensorimotor
- ☐Parkinsonism
- ☐Seizures
- ☐Spasticity
- ☐Tremors

Haematological/Immunologic
- ☐Anemia
- ☐Coagulation disorder
- ☐Immunodeficiency
- ☐Neutropenia
- ☐Pancytopenia
- ☐Recurrent infections
- ☐Thrombocytopenia
- ☐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

Integumental

Skin:
- ☐Abnormal skin blistering
- ☐Anhidrosis
- ☐Café-au-lait macules
- ☐Cutis laxa
- ☐Hemangiomas
- ☐Hyperpigmentation
- ☐Hypopigmentation
- ☐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: _____

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

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

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

Genitourinary
- ☐Ambiguous genitalia
- ☐Cryptorchidism
- ☐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 limbs 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: _____

Vascular system
- ☐Angioedema
- ☐Aneurysm
- ☐Arterial calcification
- ☐Arterial dissection
- ☐Arterial tortuosity
- ☐Arteriovenous malformation
- ☐Bruising susceptibility
- ☐Epistaxis
- ☐Lymphedema
- ☐Pulmonary hypertension
- ☐Stroke
- ☐Other: _____

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

Tumour/Malignancy

Type: _____
Location: _____
Age of onset: _____

ADDITIONAL CLINICAL FINDINGS: