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GENETICS

CLINICAL INFORMATION FORM

Complete and return with Test Requisition Form for Whole Exome Sequencing. Attach relevant medical records and/or lab results.

Patient Information

Last Name: _____ **First Name:** _____ **DOB:** _____ / _____ / _____

Hematological/Immunological

- Allergic rhinitis
- Anemia
- Autoimmunity
Specify: _____
- Coagulation disorder
- Hypoalbuminemia
- Immunodeficiency
Specify: _____
- Increased circulating antibody level
- Myelofibrosis
- Neutropenia
- Pancytopenia
- Recurrent infections
- Thrombocytopenia
- Other: _____

Dermatological

- Abnormal blistering of the skin
- Alopecia
- Angioedema
- Anhidrosis
- Blistering
- Cafe-Au-Lait macules
- Connective tissue abnormality
- Coarse hair
- Cutis laxa
- Eczema
- Hair abnormality
- Hemangiomas
- Hyperextensible skin
- Hyperpigmentation of the skin
- Hypopigmentation of the skin
- Pigmentation abnormality
- Hypohydrosis
- Ichthyosis
- Nail abnormality

- Nail bed hemorrhage
- Skin rash
- Skin tumors
- Sparse hair
- Swollen lip
- Telangiectasia
- Vascular skin abnormality
- Velvety skin
- Other: _____

Cardiovascular

- Abnormality of heart/vessels
- Amyloidosis
- Aneurysm
- Angina pectoris
- Angioedema
- Aortic root dilation
- Arrhythmia
- Arterial calcification
- Arterial dissection
- Arterial tortuosity
- Arteriovenous malformation
- Atrial septal defect
- Bicuspid aortic valve
- Bradycardia
- Cardiomyopathy
- Coarctation of aorta
- Dilated cardiomyopathy
- Epistaxis
- Heterotaxy
- Hypertension
- Hypotension
- Hypertrophic cardiomyopathy
- Hypoplastic left heart
- Lymphedema
- Mitral valve prolapse
- Noncompaction cardiomyopathy

- Patent ductus arteriosus
- Patent foramen ovale
- Pedal edema
- Prolonged QT interval
- Pulmonary hypertension
- Stroke
- Tetralogy of Fallot
- Ventricular septal defect
- Ventricular tachycardia
- Other: _____

Endocrinological

- Delayed puberty
- Diabetes insipidus
- Diabetes mellitus
- Hyperparathyroidism
- Hypoparathyroidism
- Hyperphosphatemia
- Hypophosphatemia
- Hyperthyroidism
- Hypothyroidism
- Maturity-onset diabetes of the young
- Paraganglioma
- Pheochromocytoma
- Rickets
- Other: _____

Gastroenterological

- Abdominal pain
- Chronic intestinal pseudo-obstruction
- Chronic diarrhea
- Constipation
- Dysphagia
- Duodenal stenosis/atresia
- Elevated transaminases
- Exocrine pancreatic insufficiency

- Feeding difficulties
- Gastroesophageal reflux
- Gastroschisis
- Hepatic failure
- Hepatomegaly
- Hirschsprung disease
- Hyperlipidemia
- Inflammatory bowel disease
- Intrahepatic biliary atresia
- Laryngomalacia
- Nausea
- Pancreatitis
- Pyloric stenosis
- Recurrent vomiting
- Splenomegaly
- Tracheoesophageal fistula
- Other: _____

Malformations - Neurological

- Abnormal myelination
- Abnormality of basal ganglia
- Abnormality of brainstem
- Abnormality/agenesis of corpus callosum
- Abnormality of periventricular white matter
- Aplasia/hypoplasia of cerebellar vermis
- Aplasia/hypoplasia of cerebellum
- Arnold Chiari malformation
- Brain atrophy
- Cerebellar atrophy
- Cortical dysplasia
- Hemimegalencephaly
- Heterotopia (Periventricular nodular heterotopia)
- Holoprosencephaly
- Hydrocephalus
- Leukodystrophy
- Lissencephaly
- Macrocephaly
- Microcephaly
- Pachygyria
- Periventricular leukomalacia
- Polymicrogyria
- Ventriculomegaly
- Other: _____

Malformations - Skeletomuscular

- Abnormal connective tissue
- Abnormal face shape (dysmorphic features)
Specify: _____
- Abnormality of the ear
- Abnormal uvula morphology
- Arachnodactyly
- Arthralgia
- Arthrogyposis
- Brachycephaly
- Bruising susceptibility
- Cleft lip/palate
- Clinodactyly
- Club foot/feet
- Coarse facial features
- Craniosynostosis
- Decreased muscle mass
- Ectrodactyly
- Exercise intolerance
- Fatigue
- Hemihypertrophy
- Hypertonia
- Hypotonia
- Joint hypermobility
- Limb anomaly
- Muscle weakness
- Myalgia
- Myopathic facies
- Myopathy
- Osteoarthritis
- Osteopenia
- Overgrowth
- Pectus carinatum
- Pectus excavatum
- Polydactyly
- Recurrent fractures
- Rhabdomyolysis
- Scoliosis
- Short neck
- Short stature
- Tall stature
- Skeletal dysplasia
- Syndactyly
- Synophrys
- Vertebral anomaly
- Other: _____

Metabolic

- Abnormal circulating lipid concentration
- Abnormal creatine phosphokinase
- Abnormal mitochondrial metabolism
- Abnormal newborn screen
Specify: _____
- Decreased HDL cholesterol concentration
- Elevated alanine
- Elevated hepatic transaminase
- Elevated pyruvate
- Failure to thrive
- Hyperammonemia
- Hypoammonemia
- Hyperglycemia
- Hypoglycemia
- Increased serum pyruvate
- Ketosis
- Lactic acidosis
- Plasma AA
Specify: _____
- Urine OA
Specify: _____
- Other: _____

Genitourinological

- Abnormality of the kidney
- Ambiguous genitalia
- Anuria
- Cryptorchidism
- Cystic renal dysplasia
- Hematuria
- Horseshoe kidney
- Hypogonadism
- Hydronephrosis
- Hypospadias
- Infertility
- Inguinal hernia
- Nephrolithiasis
- Nephrotic syndrome
- Polycystic kidney disease
- Proteinuria
- Renal agenesis
- Renal dysgenesis
- Renal tubulopathy
- Umbilical hernia

- Undescended testis
- Other: _____

Neurological and Developmental

- Abnormality of nervous system
 - Absent speech
 - ADHD
 - Aggressive behavior
 - Anxiety
 - Ataxia
 - Autism spectrum disorder
 - Cerebral palsy
 - Chorea
 - Cognitive impairment
 - Cortical visual impairment
 - Delayed speech and /or language development
 - Dementia
 - Developmental delay
 - Developmental regression
 - Dysarthria
 - Dyskinesia
 - Dysphasia
 - Dystonia
 - Encephalopathy
 - Fine motor delay
 - Gait disturbance
 - Gross motor delay
 - Global developmental delay
 - Hearing loss
 - Conductive hearing impairment
 - Sensorineural hearing impairment
 - Hemiplegia
 - Hyperactivity
 - Hypoesthesia
 - Incoordination
 - Intellectual disability
 - Learning disability
 - Infantile spasms
 - Migraines
 - Recurrent headaches
 - Myoclonus
 - Obsessive-compulsive disorder
 - Paresthesia
 - Parkinsonism
 - Peripheral neuropathy
 - Psychiatric symptoms
- Specify: _____

- Seizures
- Sensory neuropathy
- Spasticity
- Syncope
- Tremors
- Vertigo
- Other: _____

Neuromuscular

- Ataxia
- Chorea
- Dystonia
- Hypotonia
- Hypertonia
- Muscle weakness
- Muscular dystrophy
- Neuropathy
- Spasticity
- Other: _____

Ophthalmological

- Abnormal eye movement
- Abnormal vision
- Anophthalmia
- Blindness
- Cataracts
- Coloboma
- Corneal opacity
- CPEO
- Ectopia lentils
- External ophthalmoplegia
- Microphthalmia
- Myopia
- Nystagmus
- Optic atrophy
- Optic neuropathy
- Ptosis
- Retinal detachment
- Retinitis pigmentosa
- Strabismus
- Other: _____

Pre/Perinatal History

- Cystic hygroma
- Diaphragmatic hernia
- Encephalocele
- Growth delay
- Increased nuchal translucency
- Intrauterine growth retardation

- Nonimmune hydrops fetalis
- Oligohydramnios
- Polyhydramnios
- Prematurity GA
- Prolonged neonatal jaundice
- Other: _____

Respiratory

- Asthma
- Bronchiectasis
- Dyspnea
- Hyperventilation
- Hypoventilation
- Pneumothorax
- Pulmonary fibrosis
- Respiratory insufficiency
- Other: _____

Tumors and Malignancies

- Cancer
- Type: _____
- Location: _____
- Age of onset: _____
- Other: _____

Other Testing/Imaging

- Genetic testing
- Specify: _____
- Results: _____
- Lab: _____
- Echo
- EEG
- EMG
- Microarray
- MRI
- Muscle biopsy
- Ultrasound
- X-rays

Additional clinical information:
