

GENETICS LABORATORY WHOLE EXOME SEQUENCING TEST REQUEST FORM

Patient Name: _____ DOB: _____

REASON FOR TESTING, CLINICAL DIAGNOSIS AND ICD-10 CODES

Please include any additional clinical information (medical records, pictures, family history) to aid in result interpretation.

	Clinical Description:	Phenotypic Description:	ICD-10 Codes (required):
<input type="checkbox"/> Diagnostic <input type="checkbox"/> Prenatal <input type="checkbox"/> Carrier Screening <input type="checkbox"/> Family History <input type="checkbox"/> No Family History			

WHOLE EXOME SEQUENCING (WES)

Acceptable sample: EDTA blood, cultured cells, fresh/frozen tissue

Specific Test Instructions:

- Rapid Infant Exome** (does not include analysis for secondary findings)
 - Reflex to Full WES Trio** (can opt-in to analysis for secondary findings)
- Whole Exome Sequencing** (can opt-in to analysis for secondary findings)

ADDITIONAL CLINICAL INDICATIONS

Perinatal history

(Please check all the apply)

- Prematurity
- Intrauterine growth retardation
- Oligohydramnios
- Polyhydramnios
- Cystic hygroma / increased NT
- Shortened fetal long bones
- Choroid plexus cyst
- Ventriculomegaly
- Echogenic bowel
- Fetal pyelectasis/fetal renal pelvic dilatation
- Single umbilical artery (SUA)
- Maternal diabetes mellitus

Growth

- Failure to thrive
- Growth retardation / short stature
- Overgrowth
- Macrocephaly
- Microcephaly

Physical/Cognitive Development

- Fine motor delay
- Gross motor delay
- Speech delay
- Intellectual disability
- Learning disability
- Developmental regression

Skin, Hair, & Nail Abnormalities

- Nails: _____
- Pigmentation: _____
- Connective tissue: _____
- Blistering
- Ichthyosis
- Skin tumors/Malignancies
- Other: _____

Craniofacial/Ophthalmologic/Auditory

- Cataracts
- Cleft lip/palate
- Coloboma of eye
- CPEO (ophthalmoplegia)
- Ptosis
- Blindness
- Optic atrophy
- Retinitis pigmentosa
- Hearing loss
- Ototoxicity (aminoglycoside-induced)
- External ear malformation
- Facial dysmorphism

Describe: _____

Neurological/Muscular

- Ataxia
- Chorea
- Dystonia
- Hypotonia
- Hypertonia
- Seizures (type _____)
- Spasticity
- Exercise intolerance/easy fatigue
- Muscle weakness
- Stroke/stroke-like episodes
- Recurrent headache/migraine

Endocrine

- Diabetes mellitus Type I
- Diabetes mellitus Type II
- Hypothyroidism
- Hypoparathyroidism
- Pheochromocytoma/paraganglioma

Behavioral

- Autism spectrum disorder
- Autistic features
- Obsessive-compulsive disorder
- Stereotypic behaviors
- Other: _____

Brain Malformations/ Abnormal Imaging

- Agenesis of the corpus callosum
- Holoprosencephaly
- Lissencephaly
- Cortical dysplasia
- Heterotopia
- Hydrocephalus
- Brain atrophy
- Periventricular leukomalacia
- Hemimegalencephaly
- Abnormalities of basal ganglia
- Other: _____

Cardiac/Congenital Heart Malformations

- ASD
- VSD
- Coactation of aorta
- Hypoplastic left heart
- Tetralogy of Fallot
- Cardiomyopathy
- Arrhythmia/conduction defect
- Other: _____

Hematologic/Immunologic

- Recurrent fever
- Anemia/neutropenia/pancytopenia
- Immunodeficiency
- Type _____
- Other: _____

Skeletal/Limb abnormalities

- Contractures
- Club foot
- Polydactyly
- Syndactyly
- Scoliosis
- Vertebral anomaly
- Other: _____

Gastrointestinal

- Gastroschisis/omphalocele
- Pyloric stenosis
- Tracheoesophageal fistula
- Delayed gastric emptying
- Eosinophilic esophagitis
- Gastrointestinal reflux
- Recurrent vomiting
- Chronic diarrhea
- Constipation
- Chronic intestinal pseudo-obstruction
- Hirschsprung disease
- Hepatic failure
- Elevated transaminases

Genitourinary abnormalities

- Ambiguous genitalia
- Hypospadias
- Hydronephrosis
- Undescended testis
- Kidney malformation
- Renal agenesis
- Renal tubulopathy
- Polycystic kidneys
- Multicystic kidneys
- Other: _____

Metabolic

- Ketosis
- Lactic acidemia/high CSF lactate
- Elevated pyruvate
- Elevated alanine
- Organic aciduria
- Low plasma carnitine
- Elevated CPK
- Hypoglycemia