Clinical Questionnaire for **Whole Exome/Genome Sequencing**

Patient name: _____ **Date of birth:** _____

Ordering provider signature: _____ Provider NPI: _____ Phone: _____ Date: _____

Ordering Provider: check any boxes that apply to the proband and his/her family (current or previous history of), including the proband's 1st and 2nd degree relatives. Accurate clinical information is critical for exome/genome analysis and interpretation. The prior authorization and laboratory testing processes will not begin until this form has been completed and received.

*For any checked boxes for family members, document who the affected individual(s) is/are in the additional relevant clinical information section below.

For prior authorization only, please fax this form to 844-890-0003; otherwise, include this form along with the sample. For questions, call 866-248-1265.

Prenatal History	**Proband**	**Family***
IUGR		
Prematurity/postmaturity	❍	❍
Poly/oligohydramnios	❍	❍
Cystic hygroma/increased NT	❍	❍
Other:_____	❍	❍

General History	**Proband**	**Family***
Failure to thrive	❍	❍
Growth retardation/short stature	❍	❍
Overgrowth	❍	❍
Fine motor delay	❍	❍
Gross motor delay	❍	❍
Speech delay	❍	❍
Intellectual disability/MR	❍	❍
IQ: _____	❍	❍
Learning disability	❍	❍
Autism/Autism spectrum	❍	❍
Psychiatric disorder	❍	❍
Behavioral disorder	❍	❍

Cardiovascular Findings	**Proband**	**Family***
ASD	❍	❍
VSD	❍	❍
Cardiomyopathy	❍	❍
Arrhythmia/conduction defect	❍	❍
Tetralogy of Fallot	❍	❍
Coarctation of the aorta	❍	❍
Hypoplastic left heart	❍	❍
Aortic root dilation	❍	❍
Other:_____	❍	❍

Craniofacial/Ophthalmologic/ Auditory	**Proband**	**Family***
Blindness	❍	❍
Cataracts	❍	❍
Coloboma	❍	❍
Glaucoma	❍	❍
Retinal disorder	❍	❍
Ptosis	❍	❍
Optic atrophy	❍	❍
Retinitis pigmentosa	❍	❍
Cleft lip/palate	❍	❍
Craniosynostosis	❍	❍
Microcephaly	❍	❍
Macrocephaly	❍	❍
Hearing loss	❍	❍
External ear malformation	❍	❍
Ototoxicity	❍	❍
Dysmorphic facial features	❍	❍
Other:_____	❍	❍

Cancer/Malignancy	**Proband**	**Family***
Tumor type: _____	❍	❍
Location:_____	❍	❍
Age of onset:_____	❍	❍

Affected family members: _____

_____

Dermatologic Findings	**Proband**	**Family***
Alopecia	❍	❍
Angiokeratoma	❍	❍
Café au lait spots	❍	❍
Hypo/hyperpigmentation	❍	❍
Ichthyosis	❍	❍
Rash/dermatitis/eczema	❍	❍
Nail dysplasia	❍	❍
Abnormal connective tissue	❍	❍
Other:	❍	❍

Brain Malformations	**Proband**	**Family***
Agenesis-Corpus Callosum	❍	❍
Holoprosencephaly	❍	❍
Lissencephaly	❍	❍
Cortical dysplasia	❍	❍
Heterotopia	❍	❍
Hydrocephalus	❍	❍
Brain atrophy	❍	❍
Abnormalities of basal ganglia	❍	❍
Other:_____	❍	❍

Endocrine Findings	**Proband**	**Family***
Adrenal abnormality	❍	❍
Diabetes mellitus	❍	❍
Hypothyroidism	❍	❍
Hypoparathyroidism	❍	❍
Pheochromocytoma	❍	❍
Sex hormone abnormality	❍	❍

Gastrointestinal Findings	**Proband**	**Family***
Abnormal liver function	❍	❍
Achalasia	❍	❍
Bile duct proliferation	❍	❍
Biliary atresia	❍	❍
Cirrhosis	❍	❍
Chronic constipation	❍	❍
Chronic diarrhea	❍	❍
Congenital anomaly:_____	❍	❍
Hepatomegaly	❍	❍
Hirschsprung disease	❍	❍
Jaundice	❍	❍
Liver failure	❍	❍
Other:_____	❍	❍

Genitourinary Findings	**Proband**	**Family***
Ambiguous genitalia	❍	❍
Abnormal renal function	❍	❍
Hypospadias	❍	❍
Hydronephrosis	❍	❍
Kidney malformation	❍	❍
Obstructive renal disease	❍	❍
Renal agenesis/hypoplasia	❍	❍
Sex reversal	❍	❍
Undescended testicles	❍	❍
Congenital genital anomalies:	❍	❍

Hematologic/Immunologic	**Proband**	**Family***
Anemia	❍	❍
Allergies:_____	❍	❍
Autoimmune disorder	❍	❍
Enlarged lymph nodes	❍	❍
Immunodeficiency	❍	❍
Neutropenia/pancytopenia	❍	❍
Thymic hypoplasia	❍	❍
Other:_____	❍	❍

Metabolic	**Proband**	**Family***
Elevated alanine	❍	❍
Elevated pyruvate	❍	❍
CPK abnormalities	❍	❍
Ketosis	❍	❍
Lactic acidosis	❍	❍
Organic aciduria	❍	❍
Low plasma carnitine	❍	❍
Other:_____	❍	❍

Neuromuscular	**Proband**	**Family***
Ataxia	❍	❍
Chorea	❍	❍
Dystonia	❍	❍
Exercise intolerance/fatigue	❍	❍
Headaches/migraines	❍	❍
Hypotonia	❍	❍
Hypertonia	❍	❍
Muscle weakness	❍	❍
Neuropathy	❍	❍
Seizures/epilepsy	❍	❍
Stroke/stroke-like episodes	❍	❍
Spasticity	❍	❍
Torticollis	❍	❍
Other:_____	❍	❍

Skeletal/Limb Findings	**Proband**	**Family***
Contractures	❍	❍
Clubfoot	❍	❍
Polydactyly	❍	❍
Syndactyly	❍	❍
Scoliosis	❍	❍
Vertebral anomaly	❍	❍
Other:_____	❍	❍

_____

Consanguinity ❍ **Yes** ❍ **No**

_____

Other Testing/Findings
(Please summarize results or attach them)

Chromosomes _____

FISH _____

Microarray _____

Fragile X _____

Muscle biopsy _____

Other (clinical or research) _____

_____

Ethnicity (check all that apply)	**Maternal**	**Paternal**
Caucasian/White	❍	❍
African American/Black	❍	❍
Hispanic	❍	❍
Asian	❍	❍
Ashkenazi Jewish	❍	❍
Sephardic Jewish	❍	❍
Other:_____	❍	❍

Additional relevant clinical information:

LabCorp

www.LabCorp.com