

CLINICAL INFORMATION

Clinical information is critical for best interpretation of data. Other relevant medical records, genetic testing results, and/or family medical health history (pedigree) are encouraged to be included. Patient clinical information can also be sent via Face2Gene by sharing with our lab.

PRIMARY INDICATION

- Developmental Delay Multiple Congenital Anomalies Neuromuscular
 Dysmorphic Features Neurological Other _____

ADDITIONAL CLINICAL INFORMATION *(optional - check all that apply)*

PERINATAL HISTORY

- Cystic Hygroma
 Hydrops Fetalis
 Increased Nuchal Translucency (NT)
 Intrauterine Growth Restriction (IUGR)
 Oligohydramnios
 Polyhydramnios
 Prematurity
 Other _____

GROWTH AND DEVELOPMENT

- Failure to Thrive
 Fine Motor Delay
 Gross Motor Delay
 Overgrowth
 Short Stature
 Other _____

COGNITION AND BEHAVIOR

- ADHD
 Autism
 Global Developmental Delay
 Intellectual Disability
 Mild
 Moderate
 Severe
 Learning Disability
 Obsessive-Compulsive Disorder
 Speech Delay
 Other _____

MUSCULOSKELETAL

- Club Foot / Feet
 Contractures
 Diaphragmatic Hernia
 Joint Hypermobility
 Kyphosis
 Limb Anomaly
 Pes Planus
 Polydactyly
 Pterygium
 Scoliosis
 Syndactyly
 Vertebral Anomaly
 Other _____

CARDIOVASCULAR

- Arrhythmia
 ASD
 Cardiomyopathy
 Coarctation of Aorta
 Tetralogy of Fallot
 VSD
 Other _____

SKIN, HAIR, AND NAILS

- Hyperpigmentation (describe) _____
 Hypopigmentation (describe) _____
 Unusual Scarring
 Connective Tissue Abnormality (describe) _____
 Ichthyosis
 Rash
 Blistering
 Lipoma (or other skin tumors)
 Hair Abnormality (describe) _____
 Nail Abnormality (describe) _____
 Other _____

HEMATOLOGIC AND IMMUNOLOGIC

- Anemia
 Thrombocytopenia
 Neutropenia
 Pancytopenia
 Immunodeficiency
 Other _____

NEUROLOGICAL AND MUSCULAR

- Ataxia
 Chorea
 Seizures / Epilepsy
 Encephalopathy
 Hypotonia
 Hypertonia
 Spasticity
 Dystonia
 Muscle Weakness / Atrophy
 Exercise Intolerance
 Structural Brain Abnormalities / Abnormal Brain Imaging (describe) _____
 Other _____

CRANIOFACIAL

INCLUDING HEARING & VISION

- Cleft Lip
 Cleft Palate
 Craniosynostosis
 Dysmorphic Features (describe) _____
 Ear Malformation (describe) _____
 Microcephaly
 Macrocephaly
 Cataracts
 Coloboma (of eye)
 Chronic Progressive External Ophthalmoplegia
 Ptosis
 Abnormal Vision (describe) _____
 Optic Atrophy
 Retinitis Pigmentosa
 Abnormal Eye Movement
 Abnormal Hearing (describe) _____

GASTROINTESTINAL

- Gastroschisis
 Omphalocele
 Pyloric Stenosis
 Anal Atresia
 Tracheoesophageal Fistula
 Chronic Diarrhea
 Chronic Constipation
 Gastroesophageal Reflux
 Recurrent Vomiting
 Hirschsprung Disease
 Chronic Intestinal Pseudoobstruction
 Other _____

GENITOURINARY

- Ambiguous Genitalia
 Cryptorchidism
 Hydronephrosis
 Hypospadias
 Kidney Malformation
 Renal Agenesis or Dysgenesis
 Renal Tubulopathy
 Other _____

ENDOCRINE

- Diabetes Mellitus
 Type I
 Type II
 Hypothyroidism
 Hyperthyroidism
 Hypoglycemia
 Hypoparathyroidism
 Hyperparathyroidism
 Other _____

METABOLIC

- Abnormal Acylcarnitine Profile (describe) _____
 Abnormal CPK
 Abnormal Urine Organic Acids (describe) _____
 Abnormal Plasma Amino Acids (describe) _____
 Hyperammonemia
 Ketosis
 Lactic Acidosis
 Metabolic Acidemia
 Other _____

CANCER / TUMORS

- Tumor (describe) _____
 Age of Onset _____
 Other _____

ADDITIONAL TESTING

- ATTACH COPIES OF RESULTS, IF AVAILABLE
 Chromosomes (Karyotype), result: _____
 Chromosomal Microarray (CMA), result: _____
 Newborn Screening, result: _____
 Other Molecular Studies, results: _____

FAMILY HISTORY

ATTACH PEDIGREE, IF AVAILABLE

- Consanguinity, degree of relationship: _____