

CLINICAL HISTORY FORM

Patient Name (Last, First MI): _____

MRN: _____

DOB (MM/DD/YYYY): _____

ICD-10 code(s): _____

Please check and complete **all** that apply. This is not a substitute for submitting clinical records.

FAMILY HISTORY

- Consanguinity (parents related)
- Parents with 2 or more miscarriages
- Other relatives with similar clinical history
Please explain: _____

PERINATAL

- Cystic hygroma/Increased NT
- IUGR
- Oligohydramnios
- Polyhydramnios
- Prematurity
- Teratogenic Exposures: _____
- Other: _____

GROWTH

- Height: _____ %ile Weight: _____ %ile HC: _____ %ile
- Failure to Thrive
 - Macrocephaly: _____ %ile
 - Microcephaly _____ %ile
 - Obesity/Overgrowth
 - Short Stature
 - Tall Habitus
 - Other: _____

DEVELOPMENT

- Normal
- Developmental Regression
- Fine Motor Delay
- Global Developmental Delay
- Gross Motor Delay
- Speech Delay
- Other: _____

COGNITIVE / BEHAVIORAL

- Asperger Syndrome
- Autism Spectrum Disorder (ASD)
- Cognitive Impairment/Intellectual Disability
- Learning Disability
- Obsessive-Compulsive Disorder (OCD)
- Psychiatric Symptoms
- Stereotypic Behavior
- Other: _____

NEUROLOGICAL

- Ataxia
- Chorea
- Dystonia
- Exercise Intolerance
- Hypertonia
- Hypotonia
- Infantile Spasms
- Muscle Weakness
- Muscular Dystrophy
- Neural Tube Defect
- Recurrent Headaches/Migraines
- Seizures
- Spasticity
- Stroke
- Other: _____

HEMATOLOGY / ONCOLOGY

- Hematologic Abnormality
Specify: _____
- Solid Tumor
Specify: _____

IMMUNOLOGIC

- Bone Marrow Failure
- Immunodeficiency
Specify: _____
- Other: _____

PREVIOUS STUDIES: MRI/CT: Yes No
 Karyotype analysis _____
 DCN: 3-200.1

ENDOCRINE

- Congenital Adrenal Hyperplasia
- Delayed Puberty
- Diabetes
- Hyperinsulinism
- Hypoglycemia
- Hypoparathyroidism
- Hypothyroidism
- Precocious Puberty
- Rickets
- Other: _____

METABOLIC

- CPK Abnormalities
- Elevated Alanine
- Elevated Pyruvate
- Ketosis
- Lactic Acidemia/High CSF Lactate
- Low Plasma Carnitine
- Plasma Amino Acidopathy
- Urine Organic Aciduria

CUTANEOUS / HAIR

- Abnormal Connective Tissue
- Abnormal Hair
- Abnormal Nails
- Blistering
- Hyperpigmented Lesions
- Hypopigmented Lesions
- Ichthyosis
- Linear Streaks
- Skin Hyperelasticity
- Skin Tumors/Malignancies
- Other: _____

MUSCULOSKELETAL

- Club Foot
- Contractures
- Joint Hypermobility
- Lower Limb Anomaly
- Marfan/Other Connective Tissue Disorder
- Osteogenesis Disorder
- Polydactyly: ___ Feet ___ Hands
- Scoliosis
- Skeletal Dysplasia
- Syndactyly: ___ Fingers ___ Toes
- Upper Limb Anomaly
- Vertebral Anomaly
- Other: _____

STRUCTURAL BRAIN MALFORMATION

- Abnormalities of the Basal Ganglia
- Agenesis of Corpus Callosum
- Brain Atrophy
- Cerebellar Hypoplasia
- Cortical Dysplasia
- Dandy-Walker Malformation
- Hemimegalencephaly
- Heterotopia
- Holoprosencephaly
- Hydrocephalus
- Lissencephaly
- Pachygyria
- Periventricular Leukomalacia
- Polymicrogyria
- Other: _____

OTHER CLINICAL DESCRIPTIONS (Please explain):

EKG: Yes No
 Microarray analysis _____

CRANIOFACIAL/OPHTHALMOLOGIC/AUDITORY

- Blindness
- Cataracts
- Cleft Lip
- Cleft Palate
- Coloboma: ___ Iris ___ Retinal
- Craniosynostosis
- Dysmorphic Facial Features
- Ear Malformation
- External Ear Malformation
- Hearing Loss
- Ophthalmoplegia
- Optic Atrophy
- Optic Nerve Hypoplasia
- Ototoxicity (aminoglycoside exposure)
- Ptosis
- Retinitis Pigmentosa
- Other: _____

CARDIAC

- Aortopathy
- Arrhythmia
- ASD
- AV Canal Defect
- Bicuspid Aortic Valve
- Cardiomyopathy: ___ Dilated ___ Hypertrophic
- Coarctation of Aorta
- Heterotaxy
- Hypoplastic Left Heart
- Tetralogy of Fallot
- VSD
- Other: _____

PULMONARY

- Ciliary Dyskinesia
- Hypoventilation
- Lung Malformation
- Other: _____

GASTROINTESTINAL

- Chronic Diarrhea
- Chronic Intestinal Pseudo-Obstruction
- Constipation
- Delayed Gastric Emptying
- Diaphragmatic Hernia
- Elevated Transaminases
- Eosinophilic Esophagitis
- Gastroschisis
- Hepatic Failure
- Hirschsprung
- Omphalocele
- Pyloric Stenosis
- Tracheoesophageal Fistula
- Recurrent Vomiting
- Reflux
- Other: _____

GENITOURINARY

- Ambiguous Genitalia
- Cryptorchidism
- Hydronephrosis
- Hypospadias
- Kidney Failure
- Kidney Malformation
- Polycystic Kidneys
- Polycystic Ovaries
- Renal Agenesis
- Renal Tubulopathy
- Undescended Testes
- Urethral Obstruction
- Other: _____

Echo: Yes No
 Molecular analysis _____