

NIGMS HUMAN GENETIC CELL REPOSITORY CHROMOSOMAL ABNORMALITY CLINICAL DATA ELEMENTS

Sample ID#: _____ **Karyotype (Current ISCN nomenclature):** _____

Test Methodology (FISH, aCGH, etc): _____

Age at Diagnosis: _____ **Age at onset of symptoms:** _____

Diagnosed by: Geneticist Genetic Counselor Maternal Fetal Medicine /Obstetrician Pediatrician
 Primary Care Physician Specialist: _____

Clinical Information (Please check all that apply)

Pregnancy:

Abnormal Serum Screen Advanced Maternal Age Fetal Abnormality (*indicate below*)
 IUGR Oligohydramnios Polyhydramnios Increased Nuchal Translucency Cystic Hygroma
 Hydrops (unknown or infection) 2 Vessel Cord Premature Delivery: __weeks Prior Affected Pregnancy
 Other: _____

Neurological:

Abnormal Gyri (lissencephaly) Agenesis of the Corpus Callosum Ataxia Cerebellar Hypoplasia
 Cerebral Palsy Chorea Dandy Walker Decreased Fetal Motion Dystonia Holoprosencephaly
 Hydrocephalus Hypotonia Neural Tube Defect Seizures Spasticity Structural Brain Anomaly
 Ventriculomegaly
 Other: _____

Craniofacial:

Cleft Lip Cleft Palate Coloboma Craniosynostosis Dysmorphic Facial Features Ear Malformation
 Hemangioma Hemifacial Microsomia Hypertelorism Hypotelorism Macrocephaly: __cm
 Microcephaly: __cm Micrognathia Plagiocephaly
 Other: _____

Cutaneous:

Hyperpigmentation Hypopigmentation Other: _____

Musculoskeletal:

Acromelia Arm Anomaly Brachydactyly (fingers or toes) Clinodactyly Contractures Club Foot
 Leg Anomaly Mesomelia Micromelia Polydactyly (fingers or toes) Rhizomelia Scoliosis
 Syndactyly (fingers or toes) Vertebral anomaly Skeletal Dysplasia
 Other: _____

Cardiac:

Aortic Atresia ASD AV Canal Defect Coarctation of Aorta Dextrocardia Double Outlet Right Ventricle
 Ebstein Anomaly Echogenic Intracardiac Focus Hypoplastic Left Heart Hypoplastic Right Heart
 Interrupted Aortic Arch Pulmonary Valve Atresia Supravalvular Aortic Stenosis Transposition of Great Vessels
 Tetralogy of Fallot Truncus Arteriosus VSD
 Other: _____

Pulmonary:

CCAM Diaphragmatic Hernia Eventration of Diaphragm Pleural Effusion Pulmonary Sequestration
 Other: _____

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Gastrointestinal:

- Absent Stomach Anal Atresia Duodenal Atresia Echogenic Focus Esophageal Atresia
 Gastroischisis Hepatosplenomegaly Hirschsprung's Disease Meconium Ileus Omphalocele
 Pyloric Stenosis Tracheoesophageal Fistula
 Other: _____

Genitourinary:

- Ambiguous Genitalia Cryptorchidism Hydronephrosis Hypospadias Kidney Malformation
 Megacystis Polycystic Kidneys Renal Agenesis Urethral Obstruction
 Other: _____

Growth/Development:

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

Cognitive/Behavioral:

- Autism Learning Disability Mental Retardation: ____ (IQ/DQ) Oppositional Defiant Disorder
 Obsessive Compulsive Disorder Pervasive Developmental Delay
 Other: _____

Please describe additional dysmorphology, behaviors and other clinical features: _____
