

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: _____

Gastrointestinal: Absent Stomach Anal Atresia Duodenal Atresia Echogenic Focus
 Esophageal Atresia Gastroischis 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 dysmorphism, behaviors and other clinical features: