CORIELL INSTITUTE FOR MEDICAL RESEARCH

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

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☐ Leg Anomaly
☐ Polydactyly (fingers or toes)
☐ Syndactyly (fingers or toes)
☐ Mesomelia
☐ Rhizomelia
☐ Vertebral anomaly
☐ Micromelia
☐ Scoliosis
☐ Skeletal Dysplasia
☐ Other:______________________________

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

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

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

Genitourinary:
☐ Ambiguous Genitalia
☐ Hypospadias
☐ Polycystic Kidneys
☐ Cryptorchidism
☐ Kidney Malformation
☐ Renal Agenesis
☐ Hydronephrosis
☐ Megacystis
☐ Urethral Obstruction
☐ Other: ______________________________

Growth/Development:
☐ Failure to Thrive
☐ Gross Motor Delay
☐ Short Stature
☐ Fine Motor Delay
☐ Overgrowth
☐ Speech Delay
☐ Other: ______________________________

Cognitive/Behavioral:
☐ Autism
☐ Mental Retardation: ___ (IQ/DQ)
☐ Obsessive Compulsive Disorder
☐ Learning Disability
☐ Oppositional Defiant Disorder
☐ Pervasive Developmental Delay
☐ Other: ______________________________

Assistive Devices:
☐ Wheelchair
☐ Walker
☐ Braces

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☐ Orthotics ☐ Hearing Aid ☐ Glasses
☐ Service Animal ☐ Communication/Learning Device
☐ Other: ____________________

Treatment and Management:
☐ Physical Therapy ☐ Occupational Therapy ☐ Psychological Therapy
☐ Speech Language Therapy ☐ Special Education Services
☐ Other:____________________
☐ Medication(s):____________________
☐ Surgeries: ____________________
Please describe additional dysmorphology, behaviors and other clinical features: ____________________
____

<u>Once this form is complete</u>:

- Please include form and the General Clinical Data elements form (along with other relevant documents) with the sample in the shipping box.
- You can also email the form/documents to nigms@coriell.org or fax it to 856-437-5638.