

NIGMS HUMAN GENETIC CELL REPOSITORY

SMITH-MAGENIS SYNDROME CLINICAL DATA ELEMENTS FORM

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
 Breech
 Decreased Fetal Movement
 Other: _____

Neurological: Structural findings

- Microcephaly
 Macrocephaly
 Holoprosencephaly
 Hydrocephalus
 Abnormal Gyri (lissencephaly)
 Agenesis of the Corpus Callosum
 Cerebellar Hypoplasia
 Stroke
 Delayed Myelination or other White Matter Change
 Neural Tube Defect
 Other Structural Brain Anomaly
 Ventriculomegaly
 Dandy Walker
 Other Brain Findings: _____

Neurological: Clinical findings

- Absent or Sparse Speech
 Defective Vision
 Pupil Abnormality
 Blindness
 Strabismus
 Hearing Defect
 Facial Asymmetry
 Weakness
 Hypotonia
 Hypertonia
 Dystonia
 Ataxia
 Cerebral Palsy
 Chorea
 Dystonia
 Seizures
 Spasticity
 Absent Reflexes
 Brisk Reflexes
 Peripheral Neuropathy
 Central Sleep Apnea
 Decreased Pain Sensitivity
 Other: _____

Craniofacial:

- Cleft Lip
 Cleft Palate
 Coloboma
 Craniosynostosis
 Dysmorphic Facial Features
 Ear Malformation
 Hemangioma
 Hemifacial Microsomia
 Hypertelorism
 Hypotelorism
 Macrocephaly: __cm __%tile
 Microcephaly: __cm __%tile
 Micrognathia
 Plagiocephaly
 Brachycephaly
 Midface Retrusion (Hypoplasia)
 Dental Abnormalities
 Velopharyngeal Insufficiency
 Obstructive Sleep Apnea
 Hoarse Voice
 Other: _____

Cutaneous:

- Hyperpigmentation
 Hypopigmentation
 Dry skin____
 Other: _____

Musculoskeletal:

- Acromelia Arm Anomaly Brachydactyly (fingers or toes) Clinodactyly Contractures Club Foot
 - Leg Anomaly Pes Planus Pes Cavus 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
- Pneumothorax Other: _____

Gastrointestinal:

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

Genitourinary:

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

Growth/Development:

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

Cognitive/Behavioral:

- Autism (test) ____ Autism Spectrum Disorder Learning Disability Intellectual Disability: ____ (IQ/DQ)
 - ADHD Oppositional Defiant Disorder Obsessive Compulsive Disorder Sleep Disturbance
 - Repetitive Behaviors Self-injurious Behaviors Anxiety Disorder Mood Disorder Disruptive Behavior Disorder
- Other: _____

Other: Immunologic Abnormalities Type 2 Diabetes High Total Cholesterol

Please describe additional dysmorphism, behaviors and other clinical features:
