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 dysmorphology, behaviors and other clinical features:

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