

Smith-Magenis Syndrome 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
 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:
