Established Conditions (Not an exhaustive list)
Genetic and Metabolic Disorders
Albinism
Albright Hereditary
Angelman Syndrome (Happy Puppet Syndrome)
Achondroplasia (dwarfism)
Acrocallosal Syndrome, Schinzel Type (Absence of Corpus Callosum, Schinzel Type, ACS Hallux Duplication, Postaxial Polydactyly, Absence of Corpus Callosum, Schinzel Acrocallosal Syndrome, ACLS)
Adrenoleukodystrophy
Amelia
Antley-Bixler Syndrome (Multisynostotic Osteodysgenesis, Craniosynostosis, Choanal Atresia, Radial Humeral Synostosis, Trapezoidocephaly-Multiple Synostosis Syndrome, ABS, Multisynostotic Osteodysgenesis with Long Bone Fractures)
Apert Syndrome (Acrocephalosyndactyly)
Arthrogryposis Multiplex Congenita
Ataxia
Ataxia-Telangiectasia Syndrome (Louis-Bar Syndrome)
Beals Syndrome (Congential Contractural Arachnodactyly, Hecht-Beals Syndrome)
Beckwith-Wiedemann Syndrome
Canavan Disease
Cardio-Facio-Cutaneo Syndrome
Cerebral Lipdosis
Cerebro-Oculo-Factio-Skeletal (COFS) Syndrome
CHARGE Syndrome/Association
Chromosome 10p+, 11p-, 12p-, 13q-, 13q+, 18q-, 21q-, 22q-, 3q+, 4q-, 4Q+, 5p- Syndromes
Coffin-Lowry Syndrome
Coffin-Siris Syndrome
Cornelia de Lange Syndrome (Brachmann de Lange
Cri-du-chat Syndrome (Deletion 5p Syndrome)
Cystic Fibrosis
Dandy Walker Syndrome
Down Syndrome (Trisomy 21)
Duchenne Muscular Dystrophy
Dyggve-Melchio-Clausen Syndrome (DMC Disease, DMC Syndrome, Smith-McCort Dysplasia)
Fanconi Syndrome
Fragile X Syndrome
Fraser Syndrome (Cryptophthalmos Syndrome, Meyer-Schwickerath's syndrome, Fraser-Francois syndrome, Ullrich-Feichtiger syndrome)
Galactosemia
Gaucher Syndrome (Glucosylceramide storage disease; GSDI)
Glutaric Aciduria Type I & Type II
Glyocen Storage Disease

Hypothyroidism (congenital)
Jeune Syndrome
Joubert Syndrome
Klinefelter Syndrome
Krabbe's disease
Lesch-Nyhan Syndrome
Lissencephaly Syndrome (Miller-Dieker Syndrome, Agyria)
Maple Syrup Urine
Mucolipidosis II, III
Noonan Syndrome
Organic Acidemias
Pelizaeus-Merzbacher disease
Peroxisomal Disorders
Phenylketonuria (PKU)
Phelan-McDermid syndrome
Rubenstein-Taybi Syndrome
Schwartz-Jampel Syndrome
Prader-Willi Syndrome
Steinert Myotonic Dystrophy Syndrome (Curschmann-Batten-Steinert syndrome)
Tay-Sachs disease (Sandhoff)
Treacher-Collins Syndrome
Trisomy 8
Trisomy 9
Tetrasomy 12p
Trisomy 13 (Patau Syndrome)
Trisomy 18 (Edward's Syndrome)
Tuberous Sclerosis Complex
Urea Cycle Defect
Very long chain fatty acid storage diseases
Waardenburg Syndrome, Types I and II
Walker-Warburg Syndrome (XO)
Williams Syndrome
Zellweger Syndrome (Cerebro-Hepato-Renal Syndrome)
Neurological Disorders
Agyria (Miller-Dieker lissencephaly syndrome (MDLS), agyria syndrome, agyria-pachygyria syndrome, classical lissencephaly)
Aicardi Syndrome
Alpers Syndrome/Disease
Apert Syndrome (Acrocephalosyndactyly)
Aphasia
Arachnoid cyst with neuro-developmental delay
Arhinencephaly (Holprosencephaly)
Arnold-Chiari syndrome, type II (Malformation d'Arnold-Chiari)
Ataxia
Cerebral Palsy

CNS Aneurysm with Neuro-Developmental Delay
CNS Tumor with Neuro- Developmental Delay
Encephalopathy, congenital only
Encephalopathy, Static
Erb's Palsy (Brachial Plexus Injury, Perinantal Origin)
Holoprosencephaly
Hypertonia (persistent only)
Hypophosphotasia-Infantile
Lennox-Gastaut Syndrome
Intraventricular hemorrhage (III or IV)
Meningocele (cervical)
Miller-Dieker Syndrome
Mitochondrial Disorder
Multiple anomalies of the brain
Myopathy
Neural Tube Defect
Spinocerebellar Disorders
TAR (Thrombocytopenia-Absent Radii syndrome)
Traumatic Brain Injury (Head Trauma)
Severe Attachment Disorders
Anxiety Disorders of Infancy and Early Childhood
Depression of Infancy and Early Childhood
Infantile Anorexia
Autism Spectrum Disorders
Asperger's Disorder
Autism Spectrum Disorder
Childhood Disintegrative Disorder
Pervasive Developmental Disorder
Rett's Syndrome
Significant Sensory Impairment
Auditory Neuropathy
Blindness ("legal" blindness or 20/200 best acuity with correction)
Cataracts (congenital only)
Glaucoma
Optic Nerve Hypoplasia (DeMorsier's Syndrome, Septo Optic Dysplasia)
Progressive hearing loss as related to syndromes such as neurofibromatosis, osteopetrosis, and Usher's
Sensorineural hearing loss in excess of 25 dB HL
Aural Atresia (bilateral or unilateral)
Other
Hydrocephalus (congenital or acquired)
Low Birth Weight (<1,200 grams at birth)
Fetal Alcohol Syndrome