1. **Congenital Anomalies**
   1.1. **Central Nervous System**
       - Agenesis of the Corpus Callosum
       - Holoprosencephaly
       - Hydrocephalus w/o Spina Bifida
       - Microcephalus
       - Spina Bifida w/o Anencephaly
   1.2. **Eye, Ear, Face and Neck**
       - Anopthalmos/Micropthalmos
       - Anotia/Microtia
       - CHARGE Syndrome
       - Congenital Cataract
       - Pierre Robin Sequence
       - Treacher Collins
   1.3. **Heart and Circulatory System**
       - Aortic Valve Atresia & Stenosis
       - Coarctation of Aorta
       - Hypoplastic Left Heart
       - Patent Ductus Arteriosus (PDA)
       - Tetralogy of Fallot
   1.4. **Respiratory System**
       - Choanal Atresia
       - Lung Agenesis/Hypoplasia
   1.5. **Cleft Lip & Palate**
       - Cleft Palate w/o Cleft Lip
       - Cleft Lip w/ and w/o Cleft Palate
   1.6. **Digestive System**
       - Esophageal Atresia/Tracheoesophageal Fistula
       - Hirschsprung’s Disease
       - Pyloric Stenosis
   1.7. **Genital & Urinary Organs**
       - Hypospadias and Epispadias
       - Renal Agenesis
   1.8. **Musculoskeletal System**
       - Achondroplasia
       - Arthrogryposis
       - Congenital Hip Dislocation
       - Lower Limb Reduction Deformities
       - Upper Limb Reduction Deformities
       - Other Congenital Anomalies of the Musculoskeletal system
   1.9. **Other and Unspecified**
       - Bardet-Beidl Syndrome
       - Fragile X Syndrome
   2. **Chromosomal Anomalies**
       - Angelman Syndrome
       - Cri-du-Chat
       - DiGeorge Syndrome (Velo-Cardial-Facial Syndrome)
       - Kleinfelter Syndrome
       - Prader—Willi Syndrome
       - Trisomy 21 (Down Syndrome)
       - Trisomy 13 (Patau Syndrome)
       - Trisomy 18 (Edwards Syndrome)
       - Turner Syndrome
       - Williams Syndrome
   3. **Infectious Conditions**
       3.1. **Congenital Infections**
           - HIV / AIDS
           - Syphilis
           - TORCH:
             - Toxoplasmosis
             - Rubella
             - Cytomegalovirus
             - Herpes
       3.2. **Acquired Infections**
           - Bacterial Meningitis
           - Encephalitis
           - Poliomyelitis
           - Viral Meningitis
   4. **Endocrine/Metabolic Disorders**
       4.1. **Mucopolysaccharidosis**
           - Hunter Syndrome
           - Maroteaux-Lamy Syndrome
           - Sanfilippo Syndrome
           - Scheie Syndrome
           - Sly Syndrome
       4.2. **Enzyme Deficiency**
           - Biotinidase Deficiency
           - Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)
           - Oculocerebrorenal Syndrome (Lowe Syndrome)
   4.3. **Abnormalities of Amino Acid Metabolism**
       - Argininosuccinic Aciduria
       - Citrullinemia
       - Homocystinuria
       - Infant Phenylketonuria (PKU)
       - Maple Syrup Urine Disease
       - Methylmalonic Acidemia (MMA)
       - Ornithine Transcarbamylase Deficiency
   4.4. **Abnormalities of Carbohydrate Metabolism**
       - Galactosemia
       - Glycogen Storage Disease
   4.5. **Abnormalities of Lipid Metabolism**
       - Gaucher Disease
       - Niemann Pick Disease
   4.6. **Abnormalities of the Purine/Pyrimidine Metabolism**
       - Lesch Nyhan Syndrome
   4.7. **Abnormalities of the Parathyroid**
       - Untreated Hyperparathyroidism
       - Untreated Hypoparathyroidism
   4.8. **Abnormalities of the Pituitary**
       - Hyperpituitary
       - Hypopituitary
   4.9. **Abnormalities of Adrenocortical Function**
       - Congenital Adrenal Hyperplasia
       - Hyperadrenocortical Function
       - Hypoadrenocortical Function
   4.10. **Hemoglobinopathies**
        - Sickle Cell Disease
        - Thalassemia (major and minor)
   4.11. **Abnormalities of the Thyroid Hormone**
        - Congenital Hypothyroidism
   4.12. **Peroxisomal Disorders**
        - Adrenoleukodystrophy (ADL)
        - Cerebrohepatorenal Syndrome (Zellweger Syndrome)
        - Rhizomelic Chondrodysplasia Punctata

Note: The Endocrine/Metabolic Disorders Category also includes all disorders tested for in the Michigan Newborn Screening Pro-

www.1800EarlyOn.org

TK January 2016
5. **Other Disorders/Diseases**

### 5.1. Neurological Disorders
- **Neuromotor/Muscle Disorders**
  - Cerebral Palsy
  - Congenital Myasthenia
  - Kernicterus
  - Muscular Dystrophies
  - Paralysis
  - Periventricular Leukomalacia
  - Torticollis
  - Werdnig Hoffman Disease

### 5.2. Vision Impairment
- Amblyopia
- Cortical Visual Impairment (CVI)
- Low Vision (20/700)
- Nystagmus
- Retinopathy of Prematurity (ROP)
  - (Stage 3 - Stage 5)
- Visual Field Loss

### 6. Hearing Deficiency
- **Auditory Neuropathy**
  - Bilateral or Unilateral hearing loss of
  - ≥ 25 dB at 2+ frequencies between
  - 500-4000 Hz.
  - Mixed Hearing Loss
  - Permanent Conductive Hearing Loss
  - Sensorineural Hearing Loss
- Waardenburg Syndrome

### 7. Other Fetal/Placental Anomalies
- Twin to Twin Transfusion Syndrome
- Umbilical Cord Prolapse

### 8. Exposures Affecting Fetus/Child

#### 8.1. Prenatal
- Fetal Alcohol Spectrum Disorders - Diagnosed
- Fetal Drug Exposure - Diagnosed
- Maternal PKU

#### 8.2. Postnatal
- Lead – Venous Blood Lead level at or
  - above reference value recommended
  - by the CDC (currently 5 µg/dL, Jan. 2016).
- Mercury – for recent exposure, blood
  - level of more than 2 micrograms per
  - deciliter (>2 µg/dL); for chronic
  - exposure, urine level of more than
  - 5 micrograms per deciliter (> 5 µg/dL)

### 9. Chronic Illness

#### 9.1. Medically Fragile
- Renal Insufficiency

### 9.2. Medical Illness
- Bronchopulmonary Dysplasia
- Cancer
- Chronic Hepatitis
- Connective Tissue Disorders
- Cystic Fibrosis
- Diabetes
- Immune Disorders
  - (ex. Juvenile Arthritis)
- Organic Failure to Thrive
- Renal Failure
- Very Low Birth Weight
  - (<1500 grams or 3.3 lbs.)
- Chronic Asthma – moderate to severe
- Intrauterine Growth Retardation (IUGR)
- Small for Gestational Age
  - (<10% weight for age) (SGA)

### 10. Developmental Delay

#### 10.1. Pervasive Developmental Disorders
- Autism Spectrum Disorder
- Childhood Disintegrative Disorder
- Pervasive Developmental Disorders
  - (NOS)

#### 10.2. Rett's Disorder

#### 10.3. Regulatory Disorders of Sensory Processing
- Hyposensitive / Hypersensitive
- Sensory-Seeking/Impulsive

### 11. Mental Health Conditions

#### Adjustment Disorders
- Depression of Infancy and
  - Early Childhood

#### Maltreatment/Deprivation Disorder
- (A diagnosis of Reactive Attachment
  - Disorder should be cross-walked to this
  - diagnosis which is listed in the DC:0-3R)

#### Disorders of Affect
- Mixed Disorders of Emotional
  - Expressiveness

#### Post Traumatic Stress Disorder (PTSD)
- Regulatory Disorders**

** Difficulties in regulating physiological, attentional, motor or affective processes, and in organizing a calm, alert or affectively positive state. These disorders affect the child’s daily routines and interpersonal relationships. Must be diagnosed by a qualified professional.
  - (Greenspan, 1992)