

Established Conditions (Not an exhaustive list)	
Genetic and Metabolic Disorders	ICD10 Code
Albinism	E70.30
Albright's Hereditary Osteodystrophy	E20.1
Angelman Syndrome (Happy Puppet Syndrome)	Q93.5
Adrenoleukodystrophy	E71.529
Antley-Bixler Syndrome (Multisynostotic Osteodysgenesis, Craniosynostosis, Choanal Atresia, Radial Humeral Synostosis, Trapezoidocephaly-Multiple Synostosis Syndrome, ABS, Multisynostotic Osteodysgenesis with Long Bone Fractures)	Q87.5
Apert Syndrome (Acrocephalosyndactyly)	Q87.0
Arthrogryposis Multiplex Congenita	Q74.3
Ataxia-Telangiectasia Syndrome (Louis-Bar Syndrome)	G11.3
Canavan Disease	E75.29
Cardio-Facio-Cutaneo Syndrome	Q87.89
Cerebral Lipidosis	E75.6
Cerebro-Oculo-Facio-Skeletal (COFS) Syndrome	Q87.8
CHARGE Syndrome/Association	Q89.8
Chromosome Syndromes 10p+, 13q+, 3q+, 4Q+	Q92.5
Chromosome Syndromes 11p- (this one also called Jacobsen syndrome), 12p-, 13q-, 18q-, 21q-, 22q-, , 4q-, (this is also Wolf-Hirschhorn syndrome) 5p- (already below as cri-du-chat syndrome)	Q93.89
Coffin-Lowry Syndrome	Q89.8
Coffin-Siris Syndrome	Q03.1
Cornelia de Lange Syndrome (Brachmann de Lange)	Q87.1
Cri-du-chat Syndrome (Deletion 5p Syndrome)	Q93.4
Cystic Fibrosis	E84.0
Dandy Walker Syndrome	Q03.1
Down Syndrome (Trisomy 21)	Q90.9
Duchenne Muscular Dystrophy	G71.0
Dyggve-Melchior-Clausen Syndrome (DMC Disease, DMC Syndrome, Smith-McCort Dysplasia)	Q77.7
Fragile X Syndrome	Q99.2
Fraser Syndrome (Cryptophthalmos Syndrome, Meyer-Schwickerath's syndrome, Fraser-Francois syndrome, Ullrich-Feichtiger syndrome)	Q87.0
Galactosemia	E74.21
Gaucher Syndrome (Glucosylceramide storage disease; GSDI)	E75.22
Glutaric Aciduria	
Type I	E72.3
Type II	E71.313
Glycogen Storage Disease	E74.00
Jeune Syndrome	Q77.2
Joubert Syndrome	Q03.1
Krabbe's disease	E75.23
Lesch-Nyhan Syndrome	E79.1
Lissencephaly Syndrome (Miller-Dieker Syndrome, Agyria)	Q93.88
Maple Syrup Urine	E71.0

Mucopolysaccharidosis II, III	E77.0
Organic Acidemias	E71.121
Pelizaeus-Merzbacher disease	E75.29
Peroxisomal Disorders	E71.5
Phenylketonuria (PKU)	E70.0
Phelan-McDermid syndrome	Q93.0
Rubenstein-Taybi Syndrome	Q87.2
Schwartz-Jampel Syndrome	G71.13
Prader-Willi Syndrome	Q87.1
Steinert Myotonic Dystrophy Syndrome (Curschmann-Batten-Steinert syndrome)	G71.11
Tay-Sachs disease (Sandhoff)	E75.02
Trisomy 8	Q92.9
Trisomy 9	Q92.9
Tetrasomy 12p	Q99.8
Trisomy 13 (Patau Syndrome)	Q91.7
Trisomy 18 (Edward's Syndrome)	Q91.3
Tuberous Sclerosis Complex	Q85.1
Urea Cycle Defect	E72.20
Very long chain fatty acid storage diseases	E71.310
Walker-Warburg Syndrome (XO)	G71.2
Williams Syndrome	Q99.8
Zellweger Syndrome (Cerebro-Hepato-Renal Syndrome)	Q87.89
Neurological Disorders	
Agyria (Miller-Dieker lissencephaly syndrome (MDLS), agyria syndrome, agyria-pachygyria syndrome, classical lissencephaly)	Q04.3
Aicardi Syndrome	Q04.0
Alpers Syndrome/Disease	G31.81
Aphasia	R47.01
Arachnoid cyst with Neuro-Developmental Delay	G93.0
Arhinencephaly (Holoprosencephaly)	Q04.1 Q04.2
Arnold-Chiari Syndrome, type II (Malformation d'Arnold-Chiari)	Q07.00
Ataxia	R27.0
Cerebral Palsy	G80.9
Cerebral Aneurysm with Neuro-Developmental Delay	I67.1
CNS Tumor with Neuro- Developmental Delay	D49.7
Encephalopathy, Congenital Only	G93.40
Encephalopathy, Static	G93.40
Erb's Palsy (Brachial Plexus Injury, Perinatal Origin)	P14.0
Extracorporeal Membrane Oxygenation (ECMO)	Z92.81
Holoprosencephaly	Q04.2
Hypertonia (persistent only)	P94.1
Hypoxic Ischemic Encephalopathy (HIE)	P91.60
Lennox-Gastaut Syndrome	G40.812
Intracranial Calcifications	93.89
Intraventricular Hemorrhage	
Grade 3	P52.21
Grade 4	P52.22
Meningocele (cervical)	Q05.5

Microcephaly	Q02
Miller-Dieker Syndrome	Q93.88
Mitochondrial Disorder	E88.40
Multiple Anomalies of the Brain	Q04.9
Myopathy	G72.89
Neonatal/Perinatal Asphyxia (5 minute Apgar score of 6 or less, Cord PH < 7, Evidence of Central Nervous System involvement, Organ failure, Resuscitation)	P84
Periventricular Leukomalacia (PVL)	91.2
Spina Bifida	Q05.9
Spinocerebellar Disorders	G11.8
Severe Attachment Disorders	
Anxiety Disorders of Infancy and Early Childhood	F41.1
Depression of Infancy and Early Childhood	F33.40
Infantile Anorexia	R63.0
Autism Spectrum Disorders	
Asperger's Disorder	F84.5
Autism Spectrum Disorder	F84.0
Pervasive Developmental Disorder	F84.9
Rett's Syndrome	F84.2
Significant Sensory Impairment	
Auditory Neuropathy	H93.299
Aural Atresia (bilateral or unilateral)	Q16.1
Blindness ("legal" blindness or 20/200 best acuity with correction)	H54.8
Optic Nerve Hypoplasia (De Morsier's Syndrome)	H47.039
• Septo Optic Dysplasia	Q04.4
Retinopathy of Prematurity Stage III and/or IV (ROP)	
• Stage 3 unspecified	H35.149
○ Bilateral	H35.143
○ Left eye	H35.142
○ Right eye	H35.141
• Stage 4 unspecified	H35.159
○ Bilateral	H35.153
○ Left eye	H35.152
○ Right eye	H35.151
• Stage 5 unspecified	H35.169
○ Bilateral	H35.163
○ Left eye	H35.162
○ Right eye	H35.161
Sensorineural hearing loss in excess of 25 dB HL	H90.5
Other	
Fetal Alcohol Syndrome	Q86.0
Hydrocephalus (congenital or acquired)	G91.9
Lead Poisoning	R78.71
Low Birth Weight (<1,200 grams at birth)	P07.00
Zika Confirmed Congenital with Symptoms	A92.5
Zika Confirmed Congenital No Symptoms	A92.5
Zika Probable Congenital with Symptoms	A92.5
Zika Probable Congenital No Symptoms	A92.5