Early ACCESS

Iowa Early ACCESS Diagnosed Conditions Eligibility List

List adapted with permission from Early Intervention Colorado

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Medical Diagnosis Name	Other Names for the Diagnosis and Additional Diagnosis Information	Is this diagnosis automatically eligible for Early ACCESS?
Achondrogenesis I	Parenti-Fraccaro	Yes
Achondrogenesis II	Langer-Saldino	Yes
Acrocallosal syndrome, Schinzel Type	Schinzel Acrocallosal syndrome; ACLS; ACS; Hallux duplication, postaxial polydactyly, and absence of the corpus callosum	Yes
Acrodysostosis	Acrodysplasia; Arkless-Graham syndrome; Maroteaux-Malamut syndrome; Nasal hypoplasia-peripheral dysostosis-intellectual disability syndrome; Peripheral dysostosis-nasal hypoplasia-intellectual disability (PNM) syndrome	Yes
Adrenoleukodystrophy	ALD; AMN; X-ALD; Addison disease and cerebral sclerosis; Adrenomyeloneuropathy; Siemerling-creutzfeldt disease; Bronze schilder disease; Schilder disease; Melanodermic Leukodystrophy; sudanophilic leukodystrophy; Pelizaeus-Merzbacher disease	Yes
Agenesis of Corpus Callosum	Absence of the corpus callosum; Hypogenesis of the corpus callosum; Dysplastic corpus callosum	Yes
Aicardi syndrome	Agenesis of Corpus Callosum and Chorioretinal Abnormality; Agenesis of Corpus Callosum With Chorioretinitis Abnormality; Agenesis of Corpus Callosum With Infantile Spasms And Ocular Anomalies; Chorioretinal Anomalies with Agenesis	Yes
Alexander Disease		Yes
Allan Herndon syndrome	Allan-Herndon-Dudley syndrome; AHDS	Yes
Alper Disease	Alper's Diffuse Degeneration of Cerebral Gray Matter with Hepatic Cirrhosis; Alpers Progressive Infantile Poliodystrophy; Christensen's disease; Christensen-Krabbe disease; Diffuse Cerebral Degeneration in Infancy; Progressive Cerebral Poliodystrophy	Yes
Amputation of leg at hip		Yes
Anencephaly		Yes
Angelman syndrome	AS	Yes
Aniridia Cerebellar Ataxia Mental Deficiency	Aniridia Cerebellar Ataxia MD; Gillespie syndrome; Partial-Cerebellar Ataxia-Oligophrenia; Aniridia-Cerebellar Ataxia-Intellectual Disability; Partial-Cerebellar Ataxia-Mental Retardation	Yes
Anophthalmia, bilateral		Yes
Apert syndrome	ACS 1; ACS I; Acrocephalosyndactyly, Type I; Syndactylic Oxycephaly	Yes
APGAR score 3 or less @ 20 min	APGAR score of 3 or less at 20 minutes	Yes
Aphasia with brain damage	Aphasia with evidence of brain damage; aphemia with evidence of brain damage	Yes

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Argininosuccinic aciduria	Arginino Succinase Deficiency; ASA Deficiency; Argininosuccinate Lyase Deficiency; ASL Deficiency	Yes
Arthrogryposis	arthrogryposis multiplex congenita; AMC; multiple congenital contractures	Yes
Asphyxia w brain damage	Asphyxia (with evidence of brain damage within the first couple days of the event)	Yes
Ataxia Telangiectasia	AT; Cerebello-Oculocutaneous Telangiectasia; Immunodeficiency with Ataxia Telangiectasia	Yes
ATR-16 syndrome	Familial Mental Retardation ATR-16 syndrome	Yes
Auditory Neuropathy	Auditory neuropathy/auditory dyssynchrony; Auditory neuropathy/auditory dys-synchrony; AN/AD	Yes
Aural Atresia, Bilateral or Unilateral	congenital aural atresia; CAA (malformation of the outer ear)	Yes
Autism Spectrum Disorder	Infantile Autism; Autisitc Disorder; Asperger syndrome; Asperger's Disorder; Pervasive Developmental Disorder; PDD; Pervasive Developmental Disorder-Not Otherwise Specified; PDD-NOS	Yes
Baller Gerold syndrome	Craniosynostosis with Radial Defects; Craniosynostosis-Radial Aplasia syndrome	Yes
Bannayan Riley Ruvalcaba	Bannayan Riley Ruvalcaba syndrome; BRRS; Bannayan-Zonana syndrome (BZS); Riley-Smith syndrome; Ruvalcaba-Myhre-Smith syndrome	Yes
Bardet-Biedl syndrome	Biedl-Bardet syndrome; Laurence Moon-Biedl	Yes
Bartter syndrome	Hypokalemic Alkalosis with Hypercalciuria	Yes
Batten Disease	Neuronal Ceroid Lipofuscinoses: CLN; NCL;Vogt-Spielmeyer-Sjogren Disease; Kufs disease (adult onset)	Yes
Borhing-Opitz syndrome	Bohring syndreom; BOS syndrome	Yes
Borjeson syndrome	BORJ; Borjeson-Forssman-Lehmann syndrome; BFLS	Yes
Brain Tumor	brain cancer; brain teratoma; glioma; astrocytoma; glioblastoma multiforme; ependymoma; oligodendroglioma; medulloblastoma; meningioma; Schwannoma; acoustic neuroma; craniopharyngioma; germ cell tumor of the brain; germinoma	Yes
Bronchopulmonary Dysplasia (BPD)	chronic lung disease in premature infants	Yes- if supplemental oxygen is required at discharge from the neonatal intensive care unit
C syndrome	C syndrome; Opitz Trigonocephaly syndrome; Trigonocephaly "C" syndrome; Trigonocephaly syndrome	Yes
Cardiofaciocutaneous syndrome	CFC syndrome; Cardio-facial-cutaneous syndrome; Facio-cardio-cutaneous syndrome	Yes
Cerebellar agenesis	Cerebellar Aplasia; Cerebellar Hemiagenesis; Cerebellar Hypoplasia; Cerebellar Atrophy	Yes
Cerebral atrophy	Cerebral atrophy	Yes

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Cerebral Dysgenesis		Yes
Cerebral Palsy	Ataxic Cerebral Palsy; Athetoid Cerebral Palsy; Congenital Cerebral Palsy; Diplegia of Cerebral Palsy; Hemiparesis of Cerebral Palsy; Hemiplegia of Cerebral Palsy; Postnatal Cerebral Palsy	Yes
Cerebro Oculo Facio Skeletal syndrome	COFS syndrome; Cerebrooculofacioskeletal syndrome; Cockayne syndrome type II; Pena Shokeir II syndrome; Pena Shokeir syndrome Type II	Yes
Cerebromalacia	Encephalomalacia	Yes
CHAMP 1	Champ 1 gene mutation	Yes
CHARGE syndrome	CHARGE Association, Hall-Hittner syndrome	Yes
Chromosome 1p36 Deletion syndrome	Chromosome 1p36 deletion syndrome; Distal monosomy 1p36; Donosomy 1p36 syndrome	Yes
Chromosome 2q24 microdeletion	2q24.1 microdeletion; 2q24.2 microdeletion; 2q24.3 microdeletion	Yes
Chromosome 2q32 Deletion		Yes
Chromosome 2q37 Deletion		Yes
Chromosome 3, Monsomy 3p2	Chromosome 3, Deletion of Distal 3p; Chromosome 3, Distal 3p Monosomy; Monosomy 3p2; Partial Deletion of Chromosome 3	Yes
Chromosome 3, Trisomy 3q2	Chromosome 3, Distal 3q2 Duplication; Chromosome 3, Distal 3q2 Trisomy; Partial Duplication 3q syndrome; Partial Trisomy 3q syndrome	Yes
Chromosome 3q29 microdeletion syndrome	3qter deletion; Del(3)(q29); Monosomy 3qter; 3q subtelomere deletion syndrome; 3q29 deletion; Monosomy 3q29; 3q29 deletion syndrome	Yes
Chromosome 4 Ring	Ring 4; Ring 4, Chromosome; r4	Yes
Chromosome 4, Monosomy 4q	Interstitial Deletion of 4q; Proximal Deletion of 4q; Terminal Deletion of 4q; Chromosome 4Q minus micro deletion	Yes
Chromosome 4, Monosomy Distal 4q	4q Deletion syndrome, Partial; Chromosome 4, 4q Terminal Deletion syndrome; Chromosome 4, Partial Monosomy 4q; Del(4q) syndrome, Partial; Distal 4q Monosomy; Distal 4q- syndrome; Chromosome 4Q minus micro deletion	Yes
Chromosome 4, Partial Trsmy Distal 4q	Chromosome 4, Partial Trisomy 4q (4q2 and 4q3); Chromosome 4, Partial Trisomy 4q (4q21-qter to 4q32-qter), Distal 4q Trisomy, Duplication(4q) syndrome; Partial Duplication 4q syndrome; Partial Partial Trisomy 4q syndrome	Yes
Chromosome 4, Trisomy 4p	Chromosome 4 (Partial Trisomy 4p); Duplication(4p) syndrome; Duplication 4p syndrome	Yes
Chromosome 5, Trisomy 5p	Chromosome 5, (Trisomy 5p, Partial, Included); Duplication(5p) syndrome; Duplication 5p syndrome	Yes

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Chromosome 6 Ring	Ring 6; Ring 6, Chromosome; r6	Yes
Chromosome 6, Partial Trisomy 6q	6q+ syndrome, Partial; Chromosome 6, Trisomy 6q2; Distal Duplication 6q; Distal Trisomy 6q; Duplication 6q, Partial; Trisomy 6q syndrome, Partial; Trisomy 6q, Partial	Yes
Chromosome 6p Partial Monosomy	Partial Deletion of Chromosome 6p	Yes
Chromosome 6q 14.1 to 6q15 deletion		Yes
Chromosome 6q duplication	Duplication 6q; Trisomy 6q; 6q duplication; 6q trisomy; Partial trisomy 6q	Yes
Chromosome 6q terminal deletion syndrome		Yes
Chromosome 7, Partial Monosomy 7p	Chromosome 7, 7p Deletion syndrome, Partial; Chromosome 7, Partial Deletion of Short Arm; Del(7p) syndrome, Partial; Interstitial 7p Monosomy; Partial 7p Monosomy; Terminal 7p Monosomy; Terminal 7p Monosomy	Yes
Chromosome 7p Partial Duplication syndrome	7p Duplication syndrome	Yes
Chromosome 7q duplication	7q22.3-7q36.1 duplication	Yes
Chromosome 7q Partial Monosomy	Chromosome 7q Deletion; Chromosome 7q Partial Deletion; Chromosome 7q11.22 deletion; Chromosome 7q34-36.1 deletion; Monosomy Chromosome 7q; Partial Monosomy Chromosome 7q; Monosomy Chromosome 7q34-36.1	Yes
Chromosome 8, Monosomy 8p2	8p- syndrome, Partial; Chromosome 8, 8p Deletion syndrome, Partial Chromosome 8, Partial Deletion of Short Arm; Chromosome 8, Partial Monosomy 8p2; Del (8p) syndrome, Partial; Distal 8p Monosomy; Partial 8p Monosomy; Monosomy 8p23.1	Yes
Chromosome 8p inverted duplication/deletion syndrome	8p inverted duplication and deletion; Inverted 8p duplic	Yes
Chromosome 8q21.11 microdeletion syndrome		Yes
Chromosome 9 Ring	r9; Ring 9; Ring 9, Chromosome	Yes
Chromosome 9, Complete Trisomy 9P	Complete Trisomy 9P; Partial Trisomy 9; Chromosome 9, Partial Trisomy 9P; Trisomy 9P syndrome (Partial); Duplication 9p syndrome; Duplication(9p) syndrome; Chromosome 9, Trisomy 9pter-q11-13	Yes
Chromosome 9, Tetrasomy 9p	Chromosome 9, Tetrasomy 9p Mosaicism; Mosaic Tetrasomy 9p; Tetrasomy 9p; Tetrasomy, Short Arm of Chromosome 9	Yes
Chromosome 9, Trisomy Mosaic	Trisomy 9 Mosaic; Trisomy 9 Mosaicism; Trisomy 9 Mosaicism syndrome	Yes

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Chromosome 9q Partial Monosomy	Deletion 9q; Monosomy 9q; 9q deletion; 9q monosomy; Partial monosomy 9q	Yes
Chromosome 10, Monosomy 10p	10p Deletion syndrome (Partial); Chromosome 10, 10p- Partial; Chromosome 10, Partial Deletion (short arm)	Yes
Chromosome 10q25 and/or 10q26 deletion syndrome	10qter deletion; chromosome 10q26 deletion syndrome; distal 10q deletion syndrome; distal deletion 10q; distal monosomy 10q; monosomy 10qter; telomeric deletion 10; terminal chromosome 10q26 deletion syndrome	Yes
Chromosome 11, Partial Trisomy 11p14.3		Yes
Chromosome 11, Partial Monosomy 11q	11q- syndrome, Partial; Deletion 11q syndrome, Partial; Distal 11q Monosomy; Distal 11q- syndrome; JBS; Jacobsen syndrome; Monosomy 11q, Partial; Partial Monosomy of Long Arm of Chromosome 11; 11q14 deletion; 11q21 deletion; 11q14-21 deletion	Yes
Chromosome 11, Partial Trisomy 11q	11q Partial Trisomy, Chromosome 11; Partial Trisomy 11q13-qter, Chromosome 11; Partial Trisomy 11q21-qter, Chromosome 11; Partial Trisomy 11q23-qter; Distal Trisomy 11q; Partial Trisomy 11q; Trisomy 11q, Partial	Yes
Chromosome 12 Deletion	Deletion 12q; Monosomy 12q; 12q deletion; 12q monosomy; Partial monosomy 12q	Yes
Chromosome 12 Partial Trisomy	Duplication 12p; Trisomy 12p; 12p duplication; 12p trisomy; Partial trisomy 12p	Yes
Chromosome 12p duplication	Duplication 12p; Trisomy 12p; 12p duplication; 12p triso	Yes
Chromosome 13, Partial Monosomy 13q	13q- syndrome, Partial; Deletion 13q syndrome, Partial; Monosomy 13q, Partial; Partial Monosomy of the Long Arm of Chromosome 13	Yes
Chromosome 14 Deletion	Deletion 14q; Monosomy 14q; 14q deletion; 14q monosomy; Partial monosomy 14q	Yes
Chromosome 14 Ring	Ring 14; Ring Chromosome 14; r14	Yes
Chromosome 14, Trisomy Mosaic	Trisomy 14 Mosaic; Trisomy 14 Mosaicism syndrome; Trisomy 14 syndrome	Yes
Chromosome 15 Ring	Ring 15; Ring 15 Chromosome; Ring 15 Chromosome (mosaic pattern); r15	Yes
Chromosome 15, Distal Trisomy 15q	Chromosome 15, Trisomy 15q2; Distal Duplication 15q; Partial Duplication 15q syndrome	Yes
Chromosome 15q11-q13 Dup	Isodicentric 15; Inverted duplication 15	Yes
Chromosome 15q24 microdeletion syndrome	Del(15)(q24); Monosomy 15q24	Yes
Chromosome 16p11.2 deletion syndrome		Yes
Chromosome 16p12.2 deletion syndrome		Yes

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Chromosome 16 Duplication	Duplication 16p; Trisomy 16p; 16p duplication; 16p trisomy; Partial trisomy 16p	Yes
Chromosome 17p 13.2 duplication	Duplication 17p; Trisomy 17p; 17p duplication; 17p trisomy; Partial trisomy 17p; Duplication(17p)	Yes
Chromosome 17p13.1 and/or 17p13.2 microdeletion	17p13.1 deletion syndrome; Distal 17p13.1 microdeletion syndrome; Distal Deletion(17)(p13.1)	Yes
Chromosome 17q12 duplication	Chromosome 17q12 duplication syndrome; 17q12 microduplication syndrome; Trisomy 17q12; Recurrent duplication of 17q12; 17q12 microduplication; Duplication(17)(q12)	Yes
Chromosome 18 Ring	Ring 18; Ring Chromosome 18; r18	Yes
Chromosome 18, Tetrasomy 18p	Tetrasomy, Short Arm of Chromosome 18	Yes
Chromosome 19p duplicaton	Duplication 19p; Trisomy 19p; 19p duplication; 19p trisomy; Partial trisomy 19p	Yes
Chromosome 19p13.11 deletion syndrome		Yes
Chromosome 20q Trisomy	Chromosome 20q Duplication; Partial Trisomy 20q; Trisomy 20q11.2; Chromsome 20q11.2 Duplication; Trisomy 20	Yes
Chromosome 21q Partial Deletion syndrome	21q22 Deletion; not including small deletions of only 21q22.3	Yes
Chromosome 22 Ring	Ring 22; Ring 22, Chromosome; r22	Yes
Chromosome 22, Trisomy Mosaic	Trisomy 22 Mosaic; Trisomy 22 Mosaicism syndrome	Yes
Chromosome 22q11.2 duplication	22q11.2 duplication; 22q11.2 microduplication syndrome; Chromosome 22q11.2 duplication syndrome	Yes
Chromosome Xp deletion		Yes
Chromosome Xp22 duplication		Yes
Chromosome Xq26.2 duplication	duplication of the distal portion of the long arm of the X chromosome; chromosome X duplication; chromosome Xq duplication	Yes
Cleft Palate		Yes- up to one year after repair operation
Closed Head Injury	Closed Head Injury with neuroradiological evidence of intracranial injury (e.g.; subarachnoid hemorrhage; or intracranial hemorrhage; or swelling); Traumatic Brain Injury; TBI	Yes
Cockayne syndrome	CS; Deafness-Dwarfism-Retinal Atrophy; Dwarfism with Renal Atrophy and Deafness; Neill-Dingwall syndrome; Progeroid Nanism	Yes
Coffin-Lowry syndrome	Coffin syndrome	Yes
Coffin-Siris syndrome	Dwarfism-Onychodysplasia; Fifth Digit syndrome; Short Stature-Onychodysplasia	Yes
Cohen Synd	Pepper syndrome	Yes

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Colpocephaly		Yes
Cong/acquired absence of limb	severe limb deficiency; severe deficiency of extremity; severe form of Fibular Hemimelia; bilateral foot amputations at the ankle; complete absence of the fibula; tibular fibular hemimelia	Yes
Congenital Myopathy	Batten Turner congenital myopathy, all subdivisions (nemaline myopathy, central core myopathy, multiminicore myopathy, centronuclear myopathy, congenital fiber type disproportion myopathy, myotubular myopathy)	Yes
Congenital Toxoplasmosis		Yes
Connexin 26	Connexin 26 gene mutation (with mutation on both copies and a diagnosed unilateral or bilateral hearing loss)	Yes
Cornelia de Lange syndrome	BDLS; Brachmann-de Lange syndrome; CdLS; de Lange syndrome	Yes
Cortical Dysplasia		Yes
Cortical Hearing Impairment		Yes
Cortical Visual Impairment	CVI	Yes
Costello syndrome	FCS syndrome; Faciocutaneoskeletal syndrome	Yes
CTNNBI syndrome		Yes
Cystinosis		Yes
Cytomegalovirus	CMV; Cytomegalic Inclusion Disease; Giant Cell Inclusion Disease (CID); Human Cytomegalovirus Infection; Salivary Gland Disease, CMV Type	Yes
Dandy Walker syndrome	Dandy-Walker Cyst; Dandy-Walker Deformity; Hydrocephalus, Internal, Dandy-Walker Type; Hydrocephalus, Noncommunicating, Dandy-Walker Type; DWM; Luschka-Magendie Foramina Atresia; Heterozygous ZIC1	Yes
De Barsy syndrome	Cutis Laxa-Growth Deficiency syndrome; De Barsy-Moens-Diercks syndrome; Progeroid syndrome of De Barsy	Yes
De Sanctis Cacchione syndrome	Xerodermic Idiocy	Yes
Dejerine Sottas Disease	Hereditary Motor Sensory Neuropathy Type III; HSMN Type III; Hypertrophic Interstitial Neuritis; Hypertrophic Interstitial Neuropathy; Hypertrophic Interstitial Radiculoneuropathy, Onion-Bulb Neuropathy	Yes
Deletion 18p Synd	Chromosome 18, Monosomy 18p; 18p Deletion syndrome; 18p- syndrome; Del(18p) syndrome; Monosomy 18p syndrome; Short Arm 18 Deletion syndrome; Partial Deletion of Chromosome 18	Yes

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Deletion 18q Synd	Chromosome 18q- syndrome; 18q Deletion syndrome; 18q- syndrome; Chromosome 18 Long Arm Deletion syndrome; Chromosome 18, Monosomy 18Q; Del(18q) syndrome; Monosomy 18q syndrome; Partial Deletion of Chromosome 18	Yes
Deletion 22q11.2 syndrome	DiGeorge syndrome; velocardiofacial syndrome; Shprintzen syndrome	Yes
Deletion 5p syndrome	Cri du chat syndrome; Chromosome 5p-syndrome; Cat's Cry syndrome; Chromosome 5, Monosomy 5p; Chromosome 5p-syndrome; Le Jeune syndrome; Partial Deletion of the Short Arm of Chromosome 5 syndrome	Yes
Deletion 9p syndrome	Chromosome 9, Partial Monosomy 9p; 9p Partial Monosomy; 9p- syndrome, Partial; Chromosome 9, Partial Monosomy 9p22; Chromosome 9, Partial; Monosomy 9p22-pter; Del(9p) syndrome, Partial; Deletion 9p syndrome, Partial	Yes
Depression: Type I-Major Depression	as defined within DC:0-5; and diagnosed by specially-qualified professional as noted, the person making the diagnosis must be a practicing medical or mental/behavioral health professional	Yes
Deprivation/Maltreatment Disorder	as defined within DC:0-5; and diagnosed by specially-qualified professional as noted, the person making the diagnosis must be a practicing medical or mental/behavioral health professional	Yes
Diastrophic Dysplasia	DD; DTD; Diastrophic Dwarfism; Diastrophic Nanism syndrome	Yes
Diencephalic syndrome	Diencephalic syndrome of Childhood; Diencephalic syndrome of Emaciation; Paramedian Diencephalic syndrome; Russell's Diencephalic Cachexia; Russell's syndrome	Yes
DiGeorge syndrome		Yes
DOOR syndrome	DOOR(S) syndrome; Deafness, Onychodystrophy, Osteodystrophy,and Mental Retardation	Yes
Down syndrome	Trisomy 21 syndrome; Chromosome 21, Mosaic 21 syndrome; Chromosome 21, Translocation 21 syndrome; Trisomy G syndrome	Yes
Dravet syndrome	severe myoclonic epilepsy of infancy; SMEI	Yes
Dubowitz syndrome	Intrauterine Dwarfism	Yes
Duplication 10q syndrome	Chromosome 10, distal trisomy 10q; Chromosome 10, Partial Trisomy 10q24-qter; Chromosome 10, Trisomy 10q2; Distal Duplication 10q; Distal Trisomy 10q syndrome; Duplication(10q) syndrome	Yes
Dyggve Melchior Clausen syndrome	DMC Disease; DMC syndrome; Smith-McCort Dysplasia	Yes

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Dystonia Musculorum Deformans	Torsion Dystonia	Yes
Emanuel syndrome	Emanual syndrome; Derivative 22; der(22) chromosome; Supernumerary der(22) syndrome	Yes
Encephalitis, Herpes Simplex	HSE; Herpes Encephalitis; Herpetic Brainstem Encephalitis; Herpetic Meningoencephalitis	Yes
Encephalitis, Rasmussen's	Chronic Encephalitis and Epilepsy; Chronic Localized (Focal) Encephalitis; Epilepsy, Rasmussen's syndrome	Yes
Encephalocele	Bifid Cranium, Cephalocele, Cranial Meningoencephalocele, Craniocele, Cranium Bifidum	Yes
Encephalopathy, Hypoxic Ischemic	HIE; subacute hypoxic injury	Yes
Encephalopathy, Neonatal		Yes
Encephalopathy, Static		Yes
Epilepsy	Infantile Myoclonic Seizures, Infantile Spasm; Hypsarrhythmia	Yes
Epilepsy, Progressive Myoclonus	Progressive Myoclonus Epilepsy	Yes
Fahr's Disease	Cerebrovascular Ferrocalcinosis; Fahr Disease; Idiopathic Basal Ganglia Calcificationl; IBGC; Nonarteriosclerotic Cerebral Calcifications; SPD Calcinosis; Striopallidodentate Calcinosis	Yes
Farber Disease	Farber's lipogranulomatosis; ceramidase deficiency	Yes
Feingold syndrome		Yes
Fetal Alcohol syndrome	FAS	Yes
Fetal Hydantoin syndrome	Dilantin Embryopathy; Phenytoin Embryopathy	Yes
FG syndrome	FGS, Opitz-Kaveggia syndrome, OKS	Yes
Fiber Type Disproportion, Congenital	Atrophy of Type I Fibers; CFTD; CFTDM; Myopathy of Congenital Fiber Type Disproportion; Myopathy, Congenital, With Fiber-Type Disproportion	Yes
Fibrodysplasia Ossificans Progressiva	FOP; Myositis Ossificans Progressiva	Yes
Filippi syndrome	Syndactyly Type I with Microcephaly and Intellectual Disability	Yes
Floating Harbor syndrome	FHS; Pelletier-Leisti syndrome	Yes
Fountain syndrome		Yes
Fragile X syndrome	fra(X) syndrome; FRAXA syndrome; FXS; marker X syndrome; Martin-Bell syndrome	Yes
Friedreich's ataxia	FRDA	Yes
FRRS1L mutation		Yes
Fryns syndrome	FRNS	Yes
Fucosidosis	Alpha-L-Fucosidase Deficiency	Yes
Galactosemia	Classic Galactosemia; GALT Deficiency; Galactose-1-Phosphate Uridyl Transferase Deficiency	Yes

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Gaucher Disease, Type II	Gaucher Disease, Type 2; Glucocerebrosidase Deficiency; Glucosylceramidase Deficiency	Yes
Gaucher Disease, Type III	Gaucher Disease, Cardiovascular Form; Gaucher Disease, Type 3	Yes
Generalized Anxiety Disorder	as defined within DC:0-5; and diagnosed by specially-qualified professional as noted, the person making the diagnosis must be a practicing medical or mental/behavioral health professional	Yes
Glut One Deficiency syndrome	De Vivo Disease; glucose transporter protein syndrome; Glut-1 deficiency syndrome; Glut1-DS	Yes
Glycinemia	glycine encephalopathy; nonketotic hyperglycinemia; transient neonatal hyperglycinemia	Yes
Gm1Gangliosidosis	Beta-Galactocidase-1 Deficiency; GLB1 Deficiency; Galactocidase, Beta-1; GLB1; Morquio Disease, Type B; Elastin-Binding Protein	Yes
Griscelli syndrome, type 1		Yes
Hallervorden-Spatz Disease	Pantothenate kinase-associated neurodegeneration; PKAN	Yes
Hearing Loss (any degree of loss- bilateral-unilateral loss)		Yes
Hemimegalencephaly		Yes
HIV (confirmed)	HIV (where the child's status of the HIV infecton has been confirmed)	Yes
Holoporencephaly	Alobar Holoprosencephaly; Arhinencephaly; Familial Alobar Holoprosencephaly; HS; Holoprosencephaly Malformation Complex; Holoprosencephaly Sequence; Lobar Holoprosencephaly; Semilobar Holoprosencephaly	Yes
Homocystinuria		Yes
Human HOXA1 syndromes	Athabaskan Brainstem Dysgenesis syndrome (ABDS); Navajo Brainstem syndrome; Bosley-Salih-Alorainy syndrome; BSAS	Yes
Hunter syndrome	MPSII, MPS Disorder II, Mucopolysaccharidosis Type II	Yes
Hurler syndrome	Mucopolysaccharidosis Type I; MPS I-H; MPS1	Yes
Hydranencephaly		Yes
Hydrocephalus, Congenital	Benign Hydrocephalus, Communicating Hydrocephalus, Internal Hydrocephalus, Non-Communicating Hydrocephalus, Normal Pressure Hydrocephalus, Obstructive Hydrocephalus	Yes
Hydrocephalus, Post-hemorrhagic	Post-hemorrhagic Hydrocephalus	Yes
Hypomelanosis of Ito	Incontinentia pigmentosa acromians; Incontinentia pigmentosa	Yes

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Hypothyroidism, Congenital	Congenital Hypothyroidim; Infancy-onset Hypothyroidism	Yes
I Cell Disease	GNPTA; Inclusion Cell Disease; Leroy Disease; ML Disorder, Type II; ML II; Mucolipidosis II; N-Acetylglucosamine-1-Phosphotransferase Deficiency	Yes
Ichthyosis, Sjogren Larsson syndrome	Sjogren Larsson syndrome; SLS	Yes
Infantile Anorexia	as defined within DC:0-5 and diagnosed by specially-qualified professional as noted, the person making the diagnosis must be a practicing medical or mental/behavioral health professional	Yes
Infantile Neuroaxonal Dystrophy	INAD; Seitelberger Disease	Yes
IUGR/SGA 35 wks & < 1,700 g	Intrauterine Growth Restriction/Small for Gestational Age, qualifies children under two years only - evidence shows the child was born at 35 weeks gestational age and weighing 1,700 grams (3 pounds, 12 ounces) or less at birth or shortly after birth	Yes
IUGR/SGA 36 wks & < 1,875 g	Intrauterine Growth Restriction/Small for Gestational Age, qualifies children under two years only - evidence shows the child was born at 36 weeks gestational age and weighing 1875 grams (4 pounds, 2 ounces) or less at birth or shortly after birth	Yes
IUGR/SGA 37-40 wk & < 2,000 g	Intrauterine Growth Restriction/Small for Gestational Age, qualifies children under two years only - evidence shows the child was born between 37 and 40 weeks gestational age and weighing less than 2,000 grams (4 pounds, 5 ounces) or less at birth or shortly after birth	Yes
Intraventricular Hemorrhage, Grade III	Grade 3 IVH; Grade 3 periventricular hemorrhage; Grade III PVH	Yes
Intraventricular Hemorrhage, Grade IV	IVH; periventricular hemorrhage; PVH) Grade IV	Yes
Jervell & Lange-Nielsen syndrome	JLNS	Yes
Johanson-Blizzard syndrome		Yes
Joubert syndrome		Yes
Juberg-Marsidi syndrome		Yes
Kabuki syndrome	Kabuki makeup syndrome; KMS; Niikawa-Kuroki syndrome	Yes
KBG syndrome		Yes
Keratitis Ichthyosis Deafness syndrome	Ichthyosiform Erythroderma, Corneal Involvement, and Deafness syndrome; KID syndrome	Yes
Kernicterus	Bilirubin Encephalopathy	Yes
Klinefelter syndrome	XXY syndrome	Yes
Koolen De Vries syndrome	Koolen syndrome	Yes
Kugelberg Welander syndrome	Wohlfart-Kugelberg-Welander syndrome; mild SMA	Yes

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L1 syndrome	CRASH syndrome	Yes
Lambert-Eaton Myasthenic syndrome	LEMS	Yes
Landau Kleffner syndrome	LKS	Yes
Langer-Giedion syndrome	Tricho-Rhino-Phalangeal syndrome Type II; TRP II	Yes
Laurence Moon syndrome	Adipogenital-Retinitis Pigmentosa syndrome; Laurence syndrome; LM syndrome	Yes
Lead Encephalopathy	lead poisoning encephalopathy	Yes
Lead level greater than or equal to 20 micrograms per deciliter (mcg/dL)	Documented lead level of 20 mcg/dL or greater	Yes
Leber Congenital Amaurosis	LCA; Congenital absence of the rods and cones; Congenital retinal blindness	Yes
Leigh's Disease	infantile subacute necrotizing encephalopathy	Yes
Lennox Gastaut syndrome	LGS	Yes
Lenz Microphthalmia syndrome	Microphthalmia syndromic 1; MCOPS1; Lenz dysplasia	Yes
Lesch-Nyhan syndrome	LNS	Yes
Leukodystrophy	Types of Leukodystrophy: metachromatic leukodystrophy; Krabbé disease; adrenoleukodystrophy; Pelizaeus-Merzbacher disease; Canavan disease; Childhood Ataxia with Central Nervous System Hypomyelination; CACH; Vanishing White Matter Disease, Alexander disease, Refsum disease, cerebrotendinous xanthomatosis	Yes
Levy-Yeboa syndrome		Yes
Ligase IV syndrome	Ligase IV Deficiency; LIG4 syndrome	Yes
Linear Sebaceous Nevus syndrome	Linear Sebaceous Nevus Sequence; Sebaceous Nevus syndrome; Linear Epidermal Nevus syndrome; LEN syndrome; Jadassohn nevus phakomatosis; JNP	Yes
Lipodystrophy, generalized	Berardinelli Lipodystrophy; Berardinelli Lipodystrophy syndrome; Congenital Generalized Lipodystrophy	Yes
Lissencephaly		Yes
Locked In syndrome	cerebromedullospinal disconnection	Yes
Lowe syndrome	Oculocerebrorenal syndrome; phosphatidylinositol-4,5-bisphosphate-5-phosphatase deficiency	Yes
Macrocephaly-capillary malformation	macrocephaly-capillary malformation; macrocephaly-cutis marmorata telangiectatica congenita	Yes
Malan syndrome	Malan overgrowth syndrome	Yes
Maple Syrp Urine Disease, untreated	Maple Syrup Urine Disease (where the diagnosis is late, or there is no or inadequate treatment); BCKD Deficiency, Branched Chain Alpha-Ketoacid Dehydrogenase Deficiency, Branched Chain Ketonuria I, Classical Maple Syrup Urine Disease	Yes
Marden Walker syndrome	MWS	Yes

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Marinesco Sjogren syndrome	Garland-Moorhouse syndrome	Yes
Marshall Smith syndrome		Yes
Maxillofacial Dysostosis		Yes
Meckel-Gruber syndrome	Meckel syndrome (w/ skull defect); Dysencephalia Splanchnocystica	Yes
Megalocornea Intellectual Disability syndrome	Neuhauser syndrome	Yes
MELAS syndrome	Myopathy, Mitochondrial-Encephalopathy-Lactic Acidosis-Stroke	Yes
Meningitis		Yes
Meningomyelocele	myelomeningocele; MMC (severe form of spina bifida)	Yes
Menkes syndrome	Menkes Disease; Kinky Hair Disease	Yes
MERRF syndrome		Yes
MHBD Deficiency	2-methyl-3-hydroxybutyryl-CoA Dehydrogenase Deficiency	Yes
Microcephaly		Yes
Microdeletion 15q13.3 syndrome		Yes
Moebius syndrome	Moebius Sequence; MBS	Yes
Motor Neuron Disease		Yes
Mowat-Wilson syndrome		Yes
Mucolipidosis IV		Yes
Mucopolysaccharidosis (except for type IV)		Yes
Multiple Sulfatase Deficiency	Austin syndrome	Yes
Multisystem Developmental Disorder	MSDD; as defined within DC:0-5; and diagnosed by specially-qualified professional as noted, the person making the diagnosis must be a practicing medical or mental/behavioral health professional	Yes
Muscular Dystrophy, Duchenne	Childhood Muscular Dystrophy; DMD; Muscular Dystrophy (Classic X-linked Recessive); Progressive Muscular Dystrophy of Childhood; Pseudohypertrophic Muscular Dystrophy	Yes
Muscular Dystrophy, Fukuyama Type	Cerebromuscular Dystrophy, Fukuyama Type; Congenital Muscular Dystrophy, Fukuyama Type; FCMD; Micropolygyria With Muscular Dystrophy	Yes
Myasthenia Gravis	Myasthenia Gravis (familial infantile type)	Yes
Myhre syndrome	LAPS syndrome	Yes
Myotonic dystrophy type 1	Curschmann-Batten-Steinert syndrome; Steinert disease; dystrophia myotonia; myotonia atrophica	Yes
Myotubular Myopathy		Yes
Necrotizing Enterocolitis (NEC)		Yes- if surgery is required
Neonatal Herpes Simplex	HSV	Yes

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Neu Laxova syndrome	NLS	Yes
Neurocutaneous Melanosis		Yes
Neuropathy, Ataxia & Retinitis	Neuropathy, Ataxia and Retinitis Pigmentosa	Yes
Neuropathy, Congenital Hypomyelinatn	Charcot-Marie-Tooth Type 4E; CHN; CMT4E; Congenital Dysmyelinating Neuropathy; Congenital Hypomyelinating Polyneuropathy	Yes
Neuropathy, Giant Axonal		Yes
Neuropathy, Hered Sens Type I	HSAN1; HSN1; Hereditary Sensory and Autonomic Neuropathy Type 1	Yes
Neuropathy, Hered Sens Type II		Yes
Neuropathy, Hered Sens Type IV	Familial Dysautonomia, Type II; Hereditary Sensory and Autonomic Neuropathy IV; HSAN IV; HSN IV	Yes
Neuropathy, Peripheral		Yes
Nevus Sebaceus Syndrome	Schimmelpenning syndrome; Jadassohn nevus phacomatosis; Jadassohn sebaceous nevus syndrome; linear sebaceous nevus sequence; Schimmelpenning-Feuerstein-Mims syndrome; epidermal nevus syndrome	Yes
NF1-Neurofibromatosis	NF1; Von Recklinghausen Disease	Yes
NF2-Bilateral Acoustic Neurofibromatosis	Neurofibromatosis Type 2; NF2	Yes
Niemann-Pick Disease	Niemann-Pick Disease (Classic Infantile and Juvenile)	Yes
Nonketotic Hyperglycinemia	glycine encephalopathy	Yes
Noonan Syndrome		Yes
Norrie's syndrome	Anderson-Warburg syndrome; Atrophia Bulborum Hereditaria; Episkopi Blindness; Fetal Iritis syndrome; ND; NDP; Norrie syndrome; Whitnall-Norman syndrome	Yes
Oculocerebral syndrome with Hypopigmentation	Cross syndrome; Kramer syndrome	Yes
Oculocerebrocutaneous syndrome	Delleman syndrome; Delleman-Oorthuys syndrome; OCC syndrome; OCCS	Yes
Ohtahara syndrome	early infantile epileptic encephalopathy 1	Yes
Olivopontocerbellar Atrophy	Olivopontocerebellar Atrophy, Hereditary; Hereditary OPCA	Yes
Opitz G/BBB syndrome	Opitz syndrome; BBBG syndrome; Hypertelorism with Esophageal Abnormalities and Hypospadias; Hypertelorism-Hypospadias syndrome; Hypospadias-Dysphagia syndrome	Yes
Opsoclonus-Myoclonus syndrome	OMS; Kinsbourne syndrome	Yes
Optico-Cochleo-Dentate Degeneration		Yes
Oral-Facial-Digital syndrome	OFD syndrome; Orofaciodigital syndrome	Yes
Ornithine Transcarbamylase Deficiency		Yes

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Orocraniodigital syndrome	Juberg Hayward syndrome; Cleft Lip/Palate with Abnormal Thumbs and Microcephaly; Cranio-Oro-Digital syndrome; Digital-Oro-Cranio syndrome	Yes
Otopalatodgtl syndrome,Type I & II	Taybi syndrome; Cranioorodigital syndrome; FPO; Faciopalatoosseous syndrome; OPD syndrome	Yes
Pachygyria	Macrogyria; Broad gyri of cerebrum; Large gyri of cerebrum	Yes
Pallister Killian Mosaic syndrome	tetrasomy 12p; Killian/Teschler-Nicola syndrome	Yes
Pallister W syndrome	W syndrome	Yes
Paraplegia; Hereditary Spastic		Yes
Parent Infant Relationship-Global Assessment Scale (PIR-GAS) of 40 or less	as defined within DC:0-5,and diagnosed by specially-qualified professional as noted, the person making the diagnosis must be a practicing medical or mental/behavioral health professional	Yes
Partial Deletion of Chromosome 16p	Partial monsomy 16p	Yes
Partial Deletion of Chromosome 16q	partial monosomy 16q	Yes
Pediatric Feeding Disorder	PFD	Yes
Perisylvian syndrome, Congenital Bilateral	Polymicrogyria, bilateral; Congenital Bilateral Perisylvian syndrome; CBPS	Yes
Periventricular Leukomalacia	PVL	Yes
Pervasive Developmental Disorder	as defined within DC:0-5 and diagnosed by specially-qualified professional as noted, the person making the diagnosis must be a practicing medical or mental/behavioral health professional	Yes
Phelan-McDermid syndrome	22q13 deletion syndrome	Yes
Phenylketonuria (Untreated)	PKU, untreated	Yes
Phosphoglycerate Kinase Deficiency	Anemia, Hemolytic with PGK Deficiency; Erythrocyte Phosphoglycerate Kinase Deficiency; PGK; Phosphoglycerokinase	Yes
Pick Disease	Pick's Disease	Yes
Pitt Hopkins syndrome		Yes
Pompe Disease		Yes
Porencephaly		Yes
Posttraumatic Stress Disorder	PTSD, as defined within DC:0-5 and diagnosed by specially-qualified professional as noted, the person making the diagnosis must be a practicing medical or mental/behavioral health professional	Yes
Potocki-Lupski syndrome	Chromosome 17p11.2 Duplication	Yes
Prader-Willi syndrome	PWS	Yes
Preterm birth less than 32 weeks	Documentation shows the child was born at 32 weeks gestational age or earlier	Yes
Progressive Cystic Encephalomalacia		Yes

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Progressive Multifocal Leukoencephalopathy		Yes
Pseudo Hurler Polydystrophy	Mucolipidosis IIIA	Yes
Pyruvate Carboxylase Deficiency		Yes
Pyruvate Dehydrogenase Deficiency		Yes
Recombinant Chromosome 8 syndrome	Rec8 syndrome; San Luis Valley syndrome	Yes
Refsum syndrome	DOC 11 (Phytanic Acid Type); Disorder of Cornification 11 (Phytanic Acid Type) ; Heredopathia Atactica Polyneuritiformis; Hypertrophic Neuropathy of Refsum; Phytanic Acid Storage Disease	Yes
Regulation Disorders of Sensory Processing: Hypersensitive; Type A : Fearful/Cautious	as defined within DC:0-5 and diagnosed by specially-qualified professional as noted, the person making the diagnosis must be a practicing medical or mental/behavioral health professional	Yes
Regulation Disorders of Sensory Processing: Hyposensitive/Underresponsive	as defined within DC:0-5 and diagnosed by specially-qualified professional as noted, the person making the diagnosis must be a practicing medical or mental/behavioral health professional	Yes
Regulation Disorders of Sensory Processing: Sensory Stimulation-Seeking/Impulsive	as defined within DC:0-5 and diagnosed by specially-qualified professional as noted, the person making the diagnosis must be a practicing medical or mental/behavioral health professional	Yes
Retinoic Acid Embryopathy	Accutane Embryopathy; Accutane (Fetal Effects of); Isotretinoin Embryopathy; Isotretinoin Teratogen syndrome; Isotretinoin (Fetal Effects of); Fetal Retinoid syndrome)	Yes
Rett syndrome	Rett Disorder	Yes
Rhombencephalosynapsis	RES	Yes
Roberts syndrome	Roberts SC-Phocomelia syndrome; Roberts Tetraphocomelia syndrome; Phocomelia syndrome; Pseudo-thalidomide syndrome	Yes
Rosenberg Chutorian syndrome		Yes
Roussy Levy syndrome	Charcot-Marie-Tooth Disease (Variant); Charcot-Marie-Tooth-Roussy-Levy Disease; Hereditary Areflexic Dystasia	Yes
Rubella, Congenital		Yes
Rubinstein Taybi syndrome	Michail-Matsoukas-Theodorou-Rubinstein-Taybi syndrome; RSTS; Rubinstein Taybi (RTS) Broad Thumb-Hallux syndrome; Rubinstein syndrome	Yes
Sandhoff Disease		Yes
Sanfilippo syndrome	Mucopolysaccharidosis Type III	Yes

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Santavuori Disease	Infantile Finnish Type Neuronal Ceroid Lipofuscinosis; Balkan Disease; Infantile Neuronal Ceroid Lipofuscinosis; Infantile Type Neuronal Ceroid Lipofuscinosis	Yes
Schindler Disease	Alpha-N-Acetylgalactosaminidase Deficiency, Schindler Type; Alpha-NAGA Deficiency, Schindler Type	Yes
Schinzel Giedion syndrome	Schinzel-Giedion Midface-Retraction syndrome	Yes
Schizencephaly		Yes
Schwartz Jampel syndrome	Schwartz-Jampel-Aberfeld syndrome; SJA syndrome; SJS	Yes
Scott Craniodigital syndrome	Scott Craniodigital syndrome	Yes
Seckel syndrome	Seckel Type Dwarfism; Seckel Type Primordial Dwarfism	Yes
Separation Anxiety Disorder	as defined within DC:0-5 and diagnosed by specially-qualified professional as noted, the person making the diagnosis must be a practicing medical or mental/behavioral health professional	Yes
Shaken Baby syndrome	Shaken Impact syndrome; Shaken Infant syndrome	Yes
SHORT syndrome		Yes
Simpson Dysmorphia syndrome	Bulldog syndrome; DGSX Golabi-Rosen syndrome; Dysplasia Gigantism syndrome; X-Linked; SDYS; SGB syndrome; Simpson-Golabi-Behmel syndrome	Yes
Singleton Merten syndrome	Merten Singleton syndrome	Yes
Sirenomelia Sequence	Sirenomelia syndrome	Yes
Smith-Lemli-Opitz syndrome	SLO	Yes
Smith-Magenis syndrome	Chromosome 17p11.2 deletion syndrome	Yes
Social Anxiety Disorder	as defined within DC:0-5 and diagnosed by specially-qualified professional as noted, the person making the diagnosis must be a practicing medical or mental/behavioral health professional	Yes
Sotos syndrome	Sotos sequence	Yes
Specific Phobia	as defined within DC:0-5 and diagnosed by specially-qualified professional as noted, the person making the diagnosis must be a practicing medical or mental/behavioral health professional	Yes
Spina Bifida	(except for spina bifida occulta; in which the spinal cord is not exposed; but the vertebral bones aren't completely closed)	Yes
Spinal Muscular Atrophy	all types including Werdnig Hoffman Disease (SMA 1)	Yes
Spondyloepiphyseal Dysplasia, Congenital		Yes

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Stroke	acute neurologic syndrome; congenital stroke syndrome; neonatal stroke syndrome; brain attack; basal ganglia hemorrhage - changes in ischemia; Vein of Galen ischemia; perinatal stroke; Cerebral Venous Sinus Thrombosis; Cerebral Venous Thrombosis	Yes
Sturge-Weber syndrome		Yes
Subacute Sclerosing Panencephalitis	Dawson's Disease; Dawson's Encephalitis	Yes
Succinic Semialdehyde Dehydrogenase Deficiency	SSADH deficiency	Yes
Sydenham Chorea		Yes
Tay Sachs Disease	Cerebromacular Degeneration; GM2 Gangliosidosis, Type 1; Hexoaminidase Alpha-Subunit Deficiency (Variant B); Infantile Cerebral Ganglioside	Yes
Thanatophoric dysplasia	Dwarfism thanatophoric; TD; Thanatophoric Dwarfism	Yes
Timothy syndrome	TS	Yes
TORCH syndrome		Yes
Transverse Myelitis	Cervical Transverse Myelitis	Yes
Triphosphate Isomerase Deficiency	TPI deficiency; Triose phosphate-isomerase deficiency	Yes
Triple X syndrome	Trisomy X; 47,XXX; Triplo X syndrome	Yes
Triploidy syndrome	Triploid syndrome	Yes
Trisomy 13	Trisomy 13 - 15; Patau syndrome	Yes
Trisomy 18	Edwards syndrome	Yes
Trisomy 8	Trisomy 8 syndrome; Trisomy 8 mosaic; Trisomy 8 mosaicism	Yes
TTF-1 deletion	NKX2 deletion	Yes
Tuberous Sclerosis	Bourneville disease; Bourneville phakomatosis; cerebral sclerosis	Yes
Turner syndrome	45X syndrome; XO syndrome	Yes
Urea Cycle Defects (untreated)	where the diagnosis is late; or there is no or inadequate treatment	Yes
Usher's syndrome	deafness-retinitis pigmentosa syndrome; Graefe-Usher syndrome; retinitis pigmentosa-deafness syndrome; Hallgren syndrome	Yes
VACTERL w Hydrocephalus	VACTERL Association with Hydrocephalus; VACTERL-H Association; VATER Association with Hydrocephalus; VACTERL-H Association; VATER Association with Hydrocephalus	Yes
Van der Knapp syndrome	Megalencephalic leukoencephalopathy with subcortical cysts	Yes
Very Low Birth Weight (VLBW <1,500 grams, 3 pound 5 ounces)	Documented birth weight for an infant born at any gestational age is less than 1,500 grams or less than 3 pound 5 ounces	Yes

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Visual Impairment, Bilateral (not correctable)	bilateral vision impairment bilateral vision loss; bilateral blindness, not correctable with treatment; surgery; glasses; or contact lenses	Yes
WAGR syndrome		Yes
Walker Warburg syndrome	WWS	Yes
Watson syndrome	Pulmonic Stenosis with Cafe-Au-Lait Spots; Cafe-Au-Lait Spots with Pulmonic Stenosis	Yes
Weaver syndrome		Yes
Weill Marchesani syndrome	Marchesani syndrome; WMS	Yes
West syndrome		Yes
Wieacker syndrome	Wieacker-Wolff Syndrome; WRWF	Yes
Wiedemann Rautenstrauch syndrome	Neonatal Progeroid syndrome	Yes
Williams syndrome	Williams-Beuren syndrome	Yes
Wilson Disease		Yes
Wolf-Hirchhorn syndrome	Partial Monsomy 4p; WHS; Wolf syndrome; Chromosome 4p syndrome	Yes
Wolfram syndrome		Yes
X-linked creatine deficiency		Yes
Xeroderma Pigmentosum	DeSanctis-Cacchione syndrome; XP	Yes
XXXXX syndrome	Penta X syndrome	Yes
XXYY syndrome	XXYY syndrome	Yes
XYY syndrome	XYY syndrome	Yes
Zellweger syndrome	Bowen syndrome; Cerebrohepatorenal syndrome	Yes