Diagnosis List
Early Support for Infants and Toddlers (ESIT)

Click on the Letters Below to Jump to that Section, or Hit the "Ctrl" Key and Letter "F" on Your Keyboard to Search the Document by Keywords

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A

Absence of limb	Adams Oliver Syndrome	Alper Disease	Anoxic Insult to Brain	Arthropathy
Absent Septum Pellucidum	Adrenoleukodystrophy	Alport Syndrome	Anticonvulsant exposure	Asphyxia w brain damage
Abuse	Agenesis of Corpus Callosum	Alstrom Syndrome	Anxiety Disorder Not Otherwise Specified (NOS)	Ataxia Telangiectasia
Achondrogenesis I	Aicardi Syndrome	Amniotic Band Syndrome	Apert Syndrome	Atrioventricular canal defect
Achondrogenesis II	Alagille Syndrome	Amputation of leg at hip	APGAR score 3 or less @ 20 min	Auditory Neuropathy Spectrum Disorder
Achondroplasia	Albinism	Anencephaly	APGAR score of five or less at five minutes	Aural Atresia, Bilateral or Unilateral
Acrocallosal Syndrome, Schinzel	Alcohol-Related Neurodevelopmental Disorder	Angelman Syndrome	Aphasia	Autism Spectrum Disorder
Acrodysostosis	Alexander Disease	Aniridia	Argininosuccinic aciduria	
Acute Lymphoid Leukemia	Alexia and dyslexia	Anophthalmia, bilateral	Arnold-Chiari Malformation	
Acute Stress Disorder	Allan Herndon Syndrome	Anophthalmia, unilateral	Arthrogryposis	

B

Baller Gerold Syndrome	Batten Disease	Biliary Atresia	Brachial Plexus Palsy	Bronchopulmonary Dysplasia (BPD)
Bannayan Riley Ruvalcaba	Beals Syndrome	Bjornstad Syndrome	Brain Tumor	
Bardet-Biedl Syndrome	Beckwith Wiedemann Syndrome	Blackfan-Diamond Syndrome	Branchio-oculo-facial Syndrome	
Bartter Syndrome	Benign Enlargement of the Subarachnoid Space in infancy (aka: BESS)	Bloch-Sulzberger Syndrome	Branchio-Oto-Renal (BOR) Syndrome	

C

C Syndrome	Chromosome 2q32 Deletion	Chromosome 9q Partial Monosomy	Chromosome 17p 13.2 duplication	Coarctation of aorta
Cancer	Chromosome 3, Monsomy 3p2	Chromosome 9p Deletion Syndrome	Chromosome 17p13.1	Cockayne Syndrome
Caravan disease	Chromosome 3, Trisomy 3q2	Chromosome 10, Monosomy 10p	Chromosome 17p13.2 microdeletion	Coffin-Lowry Syndrome
Cardiac anomaly, major	Chromosome 3q26.1-3q26.2 deletion	Chromosome 10q duplication Syndrome	Chromosome 17q12 Deletion	Coffin-Siris Syndrome
Cardiofaciocutaneous Syndrome	Chromosome 4 Ring	Chromosome 10q25 deletion Syndrome	Chromosome 17q12 duplication	Cogan Syndrome
Cardiomyopathy	Chromosome 4, Monosomy 4q	Chromosome 10q26 deletion Syndrome	Chromosome 18 Trisomy	Cogan-Reese Syndrome
Carpenter Syndrome	Chromosome 4, Monosomy Distal 4q	Chromosome 11, Partial Monosomy 11q	Chromosome 18 Ring	Cohen Syndrome
Cat Eye Syndrome	Chromosome 4, Partial Trisomy Distal 4q	Chromosome 11, Partial Trisomy 11p14.3	Chromosome 18, Tetrasomy 18p	Coloboma
Cataracts, Congenital	Chromosome 4, Trisomy 4p	Chromosome 11, Partial Trisomy 11q	Chromosome 18q Deletion Syndrome	Colpocephaly
Catel-Manzke Syndrome	Chromosome 5, Trisomy 5p	Chromosome 12 Deletion	Chromosome 18p Deletion Syndrome	Complex congenital heart disease
Caudal Regression Syndrome	Chromosome 5p Deletion Syndrome	Chromosome 12 Partial Trisomy	Chromosome 19p duplicaton	Congestive Heart Failure
Central auditory processing disorder	Chromosome 5q minus Syndrome	Chromosome 12p duplication	Chromosome 20q Trisomy	Conjoined twin
Cerebellar agenesis	Chromosome 6 Ring	Chromosome Trisomy 13	Chromosome 21q Partial Deletion Syndrome	Connexin 26

Cerebellar Ataxia	Chromosome 6, Partial Trisomy 6q	Chromosome 13, Partial Monosomy 13q	Chromosome 22 Ring	Connexin 30 gene mutation
Cerebral atrophy	Chromosome 6p Partial Monosomy	Chromosome 14 Deletion	Chromosome 22, Trisomy Mosaic	Conradi-Hunermann Syndrome
Cerebral Bleed	Chromosome 7, Partial Monosomy 7p	Chromosome 14 Ring	Chromosome 22q11.2 duplication	Cortical Dysplasia
Cerebral Dysgenesis	Chromosome 7p Partial Duplication	Chromosome 14, Trisomy Mosaic	Chromosome 22Q Deletion Syndrome	Cortical Visual Impairment
Cerebral Palsy	Chromosome 7q duplication	Chromosome 15 Ring	Chromosome Trisomy 8	Costello Syndrome
Cerebro Oculo Facio Skel Syndrome	Chromosome 7q Partial Monosomy	Chromosome 15, Distal Trisomy 15q	Chromosome Xp deletion	Crainosystosis
Cerebromalacia	Chromosome 8, Monosomy 8p2	Chromosome 15q11-q13 Dup	Chronic respiratory failure	Cranio-facial anomalies
CHARGE Syndrome	Chromosome 9 Ring	Chromosome 15q24 microdeletion	Cleft Lip	CranioSyndromeostosis without Radial Defects
Childhood onset fluency disorder	Chromosome 9, Complete Trisomy 9P	Chromosome 15q13.3 Microdeletion	Cleft Palate	Crigler-Najjar Syndrome
Choanal Atresia (unilateral or bilateral)	Chromosome 9, Tetrasomy 9p	Chromosome Partial Deletion of 16q	Clinodactyly	Crouzon Syndrome
Chromosome 1p36 Deletion Syndrome	Chromosome 9, Trisomy Mosaic	Chromosome Partial Deletion of 16p	Closed Head Injury	Cystic Fibrosis
Chromosome 2q37 Deletion		Chromosome 16 Duplication	Club Foot	Cytomegalovirus

D

Dandy Walker Syndrome	Developmental Apraxia of Speech	Down Syndrome	Duchenne Muscular Dystrophy	Dysphagia,pharyngeal phase
De Barsy Syndrome	Developmental Coordination disorder	Dravet Syndrome	Dyggve Melchior Clausen Syndrome	Dysphagia,unspecified
De Sanctis Cacchione Syndrome	Diaphragmatic Hernia	Drug Exposure, Prenatal	Dysarthria	Dystonia Musculorum Deformans
Deaf-blind	Disc pigmentation Syndrome, Congenital	Duane Syndrome	Dysphagia, oral	
Dejerine Sottas Disease	DOOR Syndrome	Dubowitz Syndrome	Dysphagia, oropharyngeal phase	

E

Encephalits, Japanese	Encephalopathy Congenita	Encephalopathy, Static	Epilepsy	Esophogeal Atresia
Encephalocele	Encephalopathy, Hypoxic Ischemic	Epidermal Nevus Syndrome	Epstein's Syndrome	Expressive language disorder

F

Fabry Disease	Familial exudative vitreoretinopathy	Fetal Hydantoin Syndrome	Filippi Syndrome	Freeman Sheldon Syndrome
Facial Palsy	Familial MR Syndrome	Fetal Valproate Syndrome	Floating Harbor Syndrome	Friedreich's ataxia
Facioscapulohumeral Muscular Dystrophy	Feeding difficulties and mismanagement	FG Syndrome	Forbes Disease	Fukuyama Type Congenital Muscular Dystrophy
Fahr's Disease	Femoral Facial Syndrome	Fiber Type Disproportion	Fountain Syndrome	
Failure to Thrive	Fetal Alcohol Spectrum Disorder	Fibrodysplasia Ossificans Progressiva	Fracture of Vertebral Column with Spinal Cord Injury	
Familial Dysautonomia	Fetal Alcohol Syndrome	Fibromatosis, Congenital Generalized	Fragile X Syndrome	

G

Galactosemia	Gastroschisis	Glaucoma	Goldenhar Syndrome	Guillain Barre Syndrome
Galloway-Mowat Syndrome	Gaucher Disease Types I, II, & III	Global Developmental Delay	Gordon Syndrome	
Gangliosidosis	Generalized Anxiety Disorder	Glycinemia	Gorlin-Chaudhry-Moss Syndrome	

H

Hallgren Syndrome	Hemihyperplasia	Holoporencephaly	Hydrocephalus, Congenital	Hypoplastic Left Heart Syndrome
Hand-Schuller-Christian	Hemimegalencephaly	Homocystinuria	Hydrocephalus, Post-hemorrhagic	Hypotonia, Congenital, Non-Benign Form
Hearing Loss (permanent conductive)	Hemophilia	Hunter Syndrome	Hydrops fetalis	
Hearing Loss (sensorineural)	Heterotopia	Huntington's Disease	Hypertension pulmonary	
Heart transplant	Hirschsprung Disease	Hurler Syndrome	Hypertonia	
Hemifacial microsomia	HIV	Hydranencephaly	Hypopituitarism, Congenital	

I

I Cell Disease	Infantile Anorexia	Infantile Neuroaxonal Dystrophy	Intrauterine Growth Restriction (IUGR)	Intraventricular Hemorrhage (IVH), all grades
Infant of mother with untreated PKU	Infantile Botulism	Infantile Spasms, Epilepsy	Intraventricular Cyst	

J

Jansen Type Metaphyseal Chondrodysplasia	Jervell & Lange-Nielsen Syndrome	Johanson-Blizzard Syndrome	Joubert Syndrome	Juvenile Myelomonocytic Leukemia	Juvenile Rheumatoid Arthritis

K

Kabuki Make-up Syndrome	Keratitis Ichthyosis Dfns Syndrome	Klinefelter Syndrome	Klippel-Trenaunay Syndrome	Krabbe disease
Kearns-Sayre Syndrome	Kernicterus	Klippel-Feil Syndrome	Kniest Dysplasia	Kugelberg Welander Syndrome

L

Lambert-Eaton Myasthenic Syndrome	Lead encephalopathy	Lesch-Nyhan Syndrome (LNS)	Lipodystrophy	Lymphoma
Landau Kleffner Syndrome	Lead level =5 mcg/dL, blood	Leukemia	Lissencephaly	Lysosomal Storage Disorders
Langer-Giedion Syndrome	Leber Congenital Amaurosis	Leukodystrophy	Locked In Syndrome	
Language delay	Legg Calve Perthes Disease	Levy-Yeboa Syndrome	Low Birth Weight <1500 grams	
Laryngotracheoesophageal cleft	Leigh's Disease	Ligase IV Syndrome	Lowe Syndrome	
Laurence Moon Syndrome	Lennox Gastaut Syndrome	Linear Sebaceous Nevus Sequence	Lumbosacral Agenesis	

M

Macrocephaly	McCune Albright Syndrome	Microphthalmia	Mucopolysaccharidosis (not IVB)	Myoclonus, General
Macrocephaly cutis marmorata telangiectatica congenita syndrome (M-CMTC)	Meckel-Gruber Syndrome	Miller Syndrome	Mucopolysaccharidosis VII	Myopathy, Batten Turner
Malignant neoplasm of the kidney	MELAS Syndrome (Mitochondrial	Missing Fingers	Multiple Sulfatase Deficiency	Myopathy, Congenital

	encephalomyopathy, lactic acidosis, and stroke-like episodes)			
Maple Syrup Urine Disease, untreated	Meningitis	Mitochondrial myopathy	Muscular Dystrophies, Limb Girdle	Myopathy, Desmin Storage
Marcus Gunn Phenomenon	Menkes Syndrome	Mixed receptive expressive language disorder	Muscular Dystrophy, Becker	Myopathy, Scapuloperoneal
Marden Walker Syndrome	MERRF Syndrome (myoclonic epilepsy with ragged red fibers)	Moebius Sequence	Muscular Dystrophy, Emery Dreifuss	Myositis Ossificans Progressiva
Marfan Syndrome	Metaphyseal Chondrodysplasia, Schmid Type	Morquio Syndrome	Muscular Dystrophy, Oculogastrointestinal	Myotonia Congenita
Maroteaux Lamy Syndrome	Metatarsus adductus	Motor Neuron Disease	Musculoskeletal anomalies, Congenital	Myotonic dystrophy type 1
Marshall Syndrome	Microcephaly	Mowat-Wilson Syndrome	Myasthenia Gravis	Myotubular Myopathy
Maxillofacial Dysostosis	Micrognathia	Moyamoya disease	Myelodysplasia	
MCADD (Medium-chain acyl-coenzyme A dehydrogenase deficiency)	Micromelia	Mucolipidosis IV	Myoclonic Encephalopathy of Childhood	

N

Nager Syndrome	Neonatal Abstinence Syndrome	Neurofibromatosis	Neuropathy, Hereditary Sensory Type I, II, & IV	Norrie Disease
Nail Patella Syndrome	Neonatal Cerebral Depression	Neuromyotonia	Neuropathy, Peripheral	Nystagmus, Congenital
Necrotizing Enterocolitis (NEC)	Neonatal Herpes Simplex (HSV)	Neuropathy, Ataxia & Retinitis	Niemann-Pick Disease	
Neglect	Neonatal Seizures	Neuropathy, Congenital Hypomyelination	Nonketotic Hyperglycinemia	
Nemaline Myopathy	Neuroblastoma	Neuropathy, Giant Axonal	Noonan Syndrome	

O

Opsoclonus-Myoclonus Syndrome	Organ Failure	Orthopedically Impaired	OSMED, Homozygous	Osteosarcoma
Optic atrophy	Ornithine Transcarbamylase Deficiency	Oseogenesis imperfecta	Osteodystrophy, Congenital	Other

Optic nerve hypoplasia	Orocraniodigital Syndrome	OSMED, Heterozygous	Osteogenesis Imperfecta	Otitis Media -> 3 months, unresolved
Oral-Facial-Digital Syndrome				

P

Pachygyria	ParentInfRel-GAS of 40 or less (Parent Infant Relationship Global Scale DC:0-5)	Phenylketonuria (Untreated)	Porencephaly	Progressive Osseous Heteroplasia (POH)
Paget's Disease	Partial Fetal Alcohol Syndrome (pFAS)	Pica	Posterior Uveitis	Prolonged Bereavement/Grief Reaction
Pallister Hall Syndrome	Pendred Syndrome	Pierre-Robin Sequence	Posttraumatic Stress Disorder	Propionic acidema
Pallister Killian Mosaic Syndrome	Pentalogy of Cantrell	Plagiocephaly	Potocki-Lupski Syndrome	Proteus Syndrome
Pallister W Syndrome	PEPCK Deficiency	Poland Syndrome	Potter sequence	Prune Belly Syndrome
Panhypopituitarism	Perisylvian Syndrome, Congenital Bilateral	Polychondritis	Prader-Willi Syndrome	PTEN Hamartoma Tumor Syndrome (PHTS)
Papillitis	Periventricular Leukomalacia	Polydactyly (all types: postaxial, preaxial, and central)	Premature closure of the sutures	Ptosis
Papillon Lefevre Syndrome	Perrault Syndrome	Polymicrogyria, bilateral	Prematurity <37 weeks (up to 24 months chronological age)	Pyloric stenosis
Paramyotonia Congenita	Pervasive Developmental Disorder	Polymyalgia Rheumatica	Progressive Cystic Encephalomalacia	
Paraneoplastic Neurologic Syndromes	Peters Anomaly	Polymyositis	Progressive Multifocal Leukoencephalopathy	
Paraplegia, Hereditary Spastic	Pfeiffer Syndrome Type I	Pompe Disease	Progressive Myoclonus Epilepsy	

Q

N/A

R

Radial club hand	Retinal Hemorrhage, bilateral	Rett Syndrome	Rhombencephalosynapsis	Roussy Levy Syndrome
Refsum Syndrome	Retinoblastoma	Reye Syndrome	Rieger Syndrome	Rubella, Congenital

Regulatory Disorder of Sensory Processing	Retinopathy of Prematurity Stages 3, 4, & 5	Rh incompatibility	Roberts Syndrome	Rubinstein Taybi Syndrome
Retinal disorder, other	Retinoschisis	Rhabdomyosarcoma	Robinow Syndrome	

S

Saethre Chotzen Syndrome	Scoliosis	Sleep-Onset Disorder (Sleep-Onset Protodyssomnia)	Spinal cord injury	Subacute Sclerosing Panencephalitis
Sandhoff Disease	Scott Craniodigital Syndrome	Smith-Lemli-Opitz Syndrome	Spinal Muscular Atrophy	Subdural Hematoma
Sandifer Syndrome	Seckel Syndrome	Smith-Magenis Syndrome	Spinal Stenosis	Sydenham Chorea
Sanfilippo Syndrome	Seizures	Social Anxiety Disorder	Split Hand/Split Foot Malformation	Symbolic dysfunction, other
Santavuori Disease	Separation Anxiety Disorder	Sotos Syndrome	Spondyloepiphyseal Dysplasia Tarda	Symbolic dysfunction, unspecified
Schindler Disease	Septo-optic Dysplasia	Specific delays in development, other	Sprengel Deformity	Syndromeactyly
Schinzel Giedion Syndrome	Shaken Baby Syndrome	Specific Phobia	Stickler Syndrome	Syphilis, Congenital
Schizencephaly	Skeletal Dysplasia	Speech Disorder	Stroke	
Scleroderma	Sleep Behavior Disorder	Speech disturbance, other	Structural ocular abnormality	

T

Tay Sachs Disease	Third Nerve Palsy	Toxoplasmosis, Congenital	Transverse Myelitis	Triploidy Syndrome
Tethered Spinal Cord Syndrome	Timothy Syndrome	Tracheoesophageal Fistula	Treacher Collins Syndrome	Truncus Arteriosus
Tetralogy of Fallot	TORCH infections, prenatal	Tracheomalacia	Trichorhinophalangeal Syndrome Type I & III	Turner Syndrome
Thalamic Syndrome (Dejerine Roussy)	Torticollis	Tracheostomy	Trigger Finger	Twin-Twin Transfusion Syndrome
The child has no diagnosis	Tourette Syndrome	Transpositon of great vessels	Triple X Syndrome	

U

Unbalanced Chromosome Translocation	Urea Cycle Defects (untreated)	Usher Syndrome, Types, I, II, & III

V

VACTERL Association	Vascular Malformations of the Brain	Ventricular Septal Defects	Vertebral anomalies	Vogt Koyanagi Harada Syndrome

Van der Knapp Syndrome	Ventilator dependence	Ventriculomegaly	Visual Impairment, bilateral	Von Hippel-Lindau Syndrome

W

Waardenburg Syndrome	Werdnig Hoffman Disease	Whipple Disease	Wilson Disease	Wolfram Syndrome
WAGR Syndrome	Wernicke-Korsakoff Syndrome	Wildervanck Syndrome	Wiskott-Aldrich Syndrome	
Walker Warburg Syndrome	West Syndrome	Williams Syndrome	Wolf-Hirchhorn Syndrome	

X

Xeroderma Pigmentosum	X-linked creatine deficiency	XXXXX Syndrome	XXYY Syndrome	XYY Syndrome

Y

N/A

Z

Zellweger Syndrome