Philadelphia Birth-to-Three Early Intervention Eligibility Determination Guide: Conditions Related to Early Intervention Eligibility

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Philadelphia, Pennsylvania: Birth-to-Three Early Intervention Eligibility Determination Guide

This Guide has been developed to assist early intervention professionals within the Philadelphia early intervention system in identifying conditions and syndromes which have a high probability of resulting in a developmental delay in the birth to three populations.

The syndromes and conditions identified in this guide have been researched by Stephanie L. Ryder, M.Ed., Clinical Assessment Supervisor at the Philadelphia Health Management's ChildLink Early Intervention Service Coordination Program. A multitude of sources were utilized in the identification of which syndromes and conditions to be included, as well as a review of what other states have identified as 'established conditions' in order to make eligibility determinations within the Philadelphia early intervention system.

This Guide was reviewed by the Philadelphia Department of Behavioral Health and Mental Retardation Services and others in the Philadelphia and Pennsylvania birth to three early intervention system.

Special thanks go to Lisa Schneider and Dr. Fabiana Perla (Pennsylvania College of Optometry) for their support during this massive undertaking.

Please direct all questions or recommended updates to Ms. Ryder at 267-765-2335.

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Key:

Items which do not have either a star (*) or a plus (+++) are considered to be 'Established Conditions' within the Philadelphia Early Intervention Systems.

(*) =Require supportive documentation.
(+++)=Special Considerations should be taken when documenting these diagnoses. Contact Eleena Vo, Records Supervisor, ChildLink—215-985-6240 for further clarification.

***Disclaimer: This Guide is not to be considered inclusive of all syndromes and conditions which affect children in the early intervention systems in Pennsylvania. As new information is obtained, updates will be made accordingly**.

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A

Absence of Corpus Callosum

- **Vision impairments, delayed motor development, possible hearing deficits, mental retardation**

Acquired Immune Deficiency Syndrome (AIDS) +++

- **Disorder caused by the human inmmuno-deficiency virus (HIV). Symptoms in children include damaged immune system, recurrent infections, poor growth, and possibly brain disease resulting in developmental delays.**
- **DO NOT WRITE THIS DIAGNOSIS ON ANY DOCUMENTATION.**

Achondrogenesis Syndrome

- **This is a group of severe disorders that affect cartilage and bone development. A small body, short limbs, and other skeletal abnormalities characterize these conditions.**
- **Most infants' die of this disorder due to serious health impairments; or they are born stillborn or die shortly after birth from respiratory failure.**
- **There are documented cases where a small percentage of infants have lived a short time with intensive medical support.**
- **Types: Achondrogenesis Type 1A which is also known as Houston-Harris type; Achondrogenesis Type 1B which is also known as Parenti-Fraccaro Type (most severe form) ; Achondrogenesis Type 2 is which is also known as Langer-Saldino Type.**

Acquired Torticollis

- **See Torticollis**

Agenesis of Corpus Callosum with Infantile Spasms and Ocular Anomalies

- **See Agenesis of Corpus Callosum**

***Agenesis of Corpus Callosum**

- **A birth defect in which the structures that connect the two hemispheres of the brain are partially or completely absent.**
- **In severe cases intelligence may be affected; seizures may be present; spasticity may be present.**
- **Supportive documentation needed to determine severity of diagnosis as well as to document negative impact on intelligence.**

Agenesis of Corticalis

- **A condition in which some of the embryo's brain cells do not grow, causing loss of motor function and severe mental retardation in the infant.**

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Aicardi Syndrome

- **Genetic disorder accompanied by mental retardation, vision problems and seizure activity.**
- **http://www.healthline.com/adamcontent/aicardi-syndrome.**

Albinism

- **Genetic disorder which causes the absence of pigmentation of skin, hair, eyes, or eyes only.**
- **It can also be accompanied by visual impairments.**
- **Request supportive documentation to establish degree of visual impairment.**
- **Request Functional Visional Assessment.**

Allan Herndon Syndrome (a.k.a. Allen-Herndon-Dudley Syndrome; Allan-Herndon-Dudley Mental Retardation)

- **In the severest forms, severe mental retardation.**

Alper's Disease

- **Mental retardation, hypotonia.**

Alport Syndrome

- **An inherited disorder. Presents with congenital hearing loss and eye defects.**
- **Most often affects males, however when it occurs in females, there are no symptoms which are observable.**
- **Child must have either hearing loss or visual impairment or both.**
- http://www.healthline.com/adamcontent/alport-syndrome

Alstrom's Syndrome

- **Observable in infancy due to the light sensitivity that infant displays.**
- **Request supportive documentation to document vision impairment and/or hearing loss.**
- **Request a functional vision assessment**

Andersen-Warburg's Syndrome

- **Congenital blindness; affects males only. Hearing loss may occur.**

Anencephaly

- **Generally these children do not survive; however a small percentage does survive and the child is deaf-blind.**
- **(Good to know what portion of the brain was affected.)**

Angelman Syndrome

- **Congenital mental retardation, abnormal muscle tone, unusual facial features**.

http://medgen.genetics.utah.edu/photographs/pages/angelman.htm

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Aniridia

- **Present at birth, child is born without an iris. Vision problems. Vision fluctuates depending on lighting conditions and glare.**
- **Extremely helpful to know what are the best lighting situations for child and this can be determined by a functional vision assessment.**
- http://www.aniridia.org/conditions
- **Additional forms: Aniridia, Cerebellar Ataxia Mental Deficiency; Aniridia, Partial-Cerebellar Ataxia-Oligophrenia Aniridia-Cereballar Ataxia-Mental Retardation**

Anophthalmia (Microphathalmia) (<Both terms are used interchangeably.) (a.k.a. Small Eye Syndrome)

- **Rare condition; one or both eyes do not form during pregnancy. When both eyes are affected, blindness is the result.**
- **Vision problems; child may have residual vision.**

Apert Syndrome

- **Mental retardation; hearing loss.**

Aphasia (With evidence of brain damage)

- **Affects the child's ability to speak as well as to understanding spoken communication. Intelligence is not affected.**

Arthrogryposis

- **A spectrum of congenital multiple joint contractures.**
- **Severity varies.**

Asperger's Syndrome

- **Child presents with marked social interaction problems; prefers no changes in routine and demonstrates great difficulty coping with changes in routine; may be overly sensitive to light, sounds, smells, tastes.**
- **FYI: Language may appear to be 'normal', even at times considered to be 'rich'.**
- **They have trouble with abstract reasoning; they tend to take what is said literally.**

***Attention Deficit Hyperactivity Disorder (ADHD)**

- **Often not diagnosed prior to age 4; but there are documented cases prior to age 4.**
- **Behavioral disorder accompanied by short attention span, excessive impulsiveness and inappropriate hyperactivity.**
- **Request supportive documentation from physician, neurodevelopmental pediatrician or psychologist that states child has this diagnosis.**

Autism, Infantile

- **Child presents with communication disorders, maladaptive behaviors, poor social relationships and motor development is frequently delayed.**

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Axenfeld's Anomaly

- **Mental retardation; vision problems.**

B

Baller Gerold Syndrome

- **Mental retardation, physical abnormalities.**

Bannayan-Zonana Syndrome

- **Child presents with macrocephaly, developmental delays, and hypotonia.**

Bardet-Biedl Syndrome

- **See Lawrence-Moon-Biedl Syndrome.**

Batten Disease

- **Initially, no observable signs. Family will be the first to identify that there is something wrong due to the child's increased sensitivity to light, which will prompt the family to take child to doctor, which hopefully, will prompt the doctors to begin testing.**

Batten Turner Syndrome

- **See Muscular Dystrophy**

***Bartter's Syndrome**

- **A group of conditions involving the kidneys.**
- **The diagnosis is not what makes the child eligible, it is the muscle weakness that occurs that may make the child eligible.**
- **Supportive documentation needed.**

***Beals Syndrome**

- **Genetic disorder that causes permanent fixation of joints.**
- **Supportive documentation needed.**

***Beckwith-Weidman Syndrome**

- **Diagnosis itself will not make child eligible, it is the secondary complications that will make child eligible.**
- **Child presents with speech concerns due to large tongue. This diagnosis may impact on the development of speech and language therefore supportive documentation is needed.**

Biedl-Bardet Syndrome

- **Mental retardation, eye abnormalities, extra fingers and toes.**

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Bjornstad Syndrome (with hearing impairment)

- **The diagnosis in and of itself does not make the child eligible, it is the hearing impairment that makes the child eligible.**

Bilateral Cleft Lip/Palate

- **Speech and feeding issues; possible hearing deficits.**

Blindness

- **Congenitally blind children are almost always delayed in motor development.**
- **Request functional vision assessment.**
- **If vision is less than 20/200 in corrected or better eye, child is eligible.**
- **Request supportive documentation, eye report.**

Bonnevie-Ulrich Syndrome

- **See Turner Syndrome**

Bowen Hutterite Syndrome

- **Intrauterine growth delays that continues after birth.**
- **Child may be diagnosed as Failure to Thrive as well.**
- **Prognosis is poor.**

***Brachial Plexus Injuries**

- **Those children who suffer from the *avulsion* and *rupture* forms of this condition would be eligible. (Supportive information is required.)**
- **Request supportive documentation—diagnosis of Avulsion or Rupture form of diagnosis has a high probability of resulting in a developmental delay.**

***Bronze Schilder Disease**

- **Progressive disease.**
- **May have cortical blindness, cortical deafness, and spastic hemiplegia.**

C

C Syndrome (a.k.a Optiz Trigonocephaly Syndrome)

- **Craniofacial abnormalities present, developmental and learning disabilities are common.**

Canavan's Disease

- **Congenital mental retardation present; increased muscle tone, poor head control, and blindness and /or hearing loss.**

Carpenter Syndrome

- **Mental retardation, finger and toe abnormalities, short stature.**

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Cat-Eye Syndrome

- **See Partial Trisomy 22**

Catel-Manzke Syndrome

- **Shares same features as Pierre Robin Syndrome and may have cleft palate, growth retardation, cardiac issues, feeding issues and other developmental issues.**

Caudal Regression Syndrome, Severe

- **Abnormal fetal development of the spine which can lead to a variety of problems including partial absence of the tail bone regions of the spine to major malformations of the lower vertebrate, spine or pelvis.**
- **Severe cases have significant birth defects and neurological impairments.**

Cerebella Agenesis

- **Total or partial part of the brain called the cerebellum does not develop.**
- **Infants will have difficulty with control of muscle movements, inability to coordinate eye movements, demonstrate quivering and jerky movements which are involuntary.**

Cerebral Palsy

- **Brain damage, physical complications vary.**

***Cerevical Dystonia**

- **See Spasmodic Torticollis**

CHARGE Syndrome

- **Mental retardation, vision problems (Colobama), growth retardation, hearing loss, as well as other problems.**

Charcot-Marie-Tooth Disease (a.k.a. Progressive Neuropathic Muscular Atrophy: Hereditary Motor and Sensory Neuropathy)

- **Group of slowly progressive disorders that result from progressive damage to nerves.**
- **Wasting of muscle tissue occurs in feet and legs and then in hands and arms.**
- **May have a floppy gait, foot drop, or foot deformity.**
- **Physical therapy indicated.**

Cleft Palate

- **Usually accompanied by speech and feeding complications.**
- **May occur in combination with other disorders/diagnoses.**

Cleft Lip

- **Usually accompanied by speech and feeding complications.**
- **May occur in combination with other disorders/diagnoses.**

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Club Foot

- **Physical complications may require surgery as well as a need for PT/OT services**

Cockayne Syndrome

- **Inherited disorder, accompanied by mental retardation, sensitivity to sunlight, jerky movements, and problems with walking**.

Coffin-Lowry Syndrome

- **Males: Severe to profound mental retardation.**
- **Females may or may not be affected; therefore, supportive documentation is needed.**

Cogan Syndrome

- **Hearing impairments.**

Cohen Syndrome

- **Developmental delays, mental retardation, hypotonia.**

Coloboma

- **Vision problems.**
- **Functional vision assessment.**

Congenital Rubella

- **Condition occurs when an infant has been exposed prenatally to rubella virus; transmission is mother to child in utero.**
- **Hearing problems, possible vision problems.**
- **May be deaf-blind.**

Congenital Syphilis (+++)

- **Developmental delays may be present as well as Failure to Thrive.**
- **DO NOT WRITE THIS DIAGNOSIS ON ANY DOCUMENTATION.**

Congenital Torticollis

- **See Torticollis**

***Congenital Toxoplasmosis**

- **Child may have blindness, seizures.**
- **Supportive documentation needed to indicate visual impairment.**

***Conradi-Hunerman Syndrome**

- **Mental retardation may be present. Primarily occurs in females with a few rare occurrences in males.**
- **Supportive documentation needed to document mental retardation.**

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Congenital Muscular Dystrophy

- **Affects the muscles of the body, is present at birth and may manifest itself by low muscle tone, poor head control, and facial muscles may be affected.**
- **See Muscular Dystrophy for types.**

Cornelia deLange Syndrome

- **Congenital mental retardation and disorder is generally accompanied by other conditions or diagnoses.**

Cortical Visual Impairment (CVI)

- **Brain disorder, may be permanent or temporary, which affects vision.**
- **Request functional vision assessment which should be completed over several sessions in a variety of lighting situations.**

Costello Syndrome

- **Affects many parts of the body.**
- **Presents with developmental delay, mental retardation, distinctive facial features, and difficulties with feeding.**
- **Growth may be slow.**

http://ghr.nlm.nih.gov/conditions=costellosyndrome.

***Cranial Mononeuropathy VI (with significant vision impairment)**

- **This is a neurological disorder which causes double vision. Can be temporary or permanent.**

Cri du Chat Syndrome

- **Genetic disorder accompanied by mental retardation and physical abnormalities.**

Cystic Fibrosis (CF)

- **Congenital respiratory disease which affects the lungs and digestive system.**

***Cystic Periventricular Leukomalacia (CPL)**

- **Supportive documentation needed because this diagnosis is classified as high risk for developmental abnormalities.**
- **Additional problems include spastic diplegia, problems in motor development, slow mental development, problems with hearing, and problems with vision, seizure activity, learning disabilities, poor eye-hand coordination and behavioral difficulties.**

***Cytomegalovirus (CMV) a.k.a. Cytomegalovirus Inclusion Disease (CID)**

- **May have hearing impairment, mental retardation.**
- **Requires supportive documentation because 1 in 750 children are born w/ or develops permanent disabilities that include mental retardation.**

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D

Dandy Walker Syndrome
- **Congenital mental retardation.**

DeBarsy Syndrome
- **Vision problems, premature aging.**

Deaf-Blindness
- **Request functional vision assessment.**
- **Team must consist of professional who has expertise with deaf-blind population.**

***DeMorsier's Syndrome (a.k.a. Septo-Optic Dysplasis)**
- **Rare disorder, present at birth, in which the optic nerve is underdeveloped and the pituitary gland does not function. A portion of the brain tissue is not formed.**
- **This disorder may cause blindness in one or both eyes and is also accompanied by Nystagumus and various other symptoms.**
- **Some children with this diagnosis are of normal intelligence, while others may be developmentally delayed or mental retarded.**
- **Supportive documentation needed to document the visual impairment and or mental retardation.**

De Santis Cacchione Syndrome
- **Neurological problems, mental retardation, eye disorders.**

DiGeorge Syndrome(a.k.a. 22q11deletion)
- **Accompanied by Cleft Palate and or Cleft Lip, feeding difficulties, hearing loss, microcephaly, and mental retardation.**
- **Not all of the symptoms need to be present to make the diagnosis.**

DOOR Syndrome
- **Deafness, Onychodystrophy, Osteodystrophy and mental retardation.**

Down Syndrome (a.k.a. Trisomy 21, Trisomy G)
- **Congenital mental retardation (varying in severity), delays in motor development, feeding problems, and communication problems.**
- **In addition, there may be vision problems, hearing problems, hypotonia, upward slant to eyes, epicanthal features, flattened back of head, short broad hands.**

Dubowitiz Syndrome
- **Mental retardation (varying in severity), facial features are unusual, may present with hyperactivity, stubbornness, and shyness.**

http://www.peacehealth.org/kbase/nord/nord198.htm.

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Duchenne's Muscular Dystrophy
Additional names:

- **Duchenne's Paralysis**
- **Duchenne-Erb Paralysis**
- **Duchenne-Erb Syndrome**

See Muscular Dystrophy for additional information.

Dwarfism

- **Disproportionate Dwarfism: If body size is disproportionate, some parts of the body will be small in size while other parts of the body will be average or above average in size. Disorders causing disproportionate dwarfism will inhibit the development of bones. Almost all persons who have disproportionate dwarfism are of normal intelligence with the exception of those who have excess fluid on the brain which will result in hydrocephalus.**

- **Proportionate dwarfism: A body size which is proportionately small if all body parts are small to the same degree and appear to be proportioned like a body of average size and stature. Usually, this means that a person has an average-size trunk and very short limbs, but some may have a very short trunk and shortened but disproportionately large limbs.**

  A common characteristic of these disorders is that the head is larger than the rest of the body.

E

Ealses Disease

- **Vision problem, sudden vision loss, affects males.**

Edward's Syndrome

- **Severe mental retardation; digits (fingers) overlap.**

Encephalitis

- **One side of body paralyzed; may have vision problems.**

Encephalitis, Rasmussen's

- **Rare chronic inflammatory disease that usually affects only one hemisphere of the brain.**
- **Characterized by frequent and severe seizures, loss of motor skills and speech, paralysis on one side of the body.**
- **Prognosis varies.**
- **Cognitive and speech deficits.**

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Encephalocele

- **Congenital brain damage, motor delays with weakness and or spasticity, ataxia, seizures and vision problems as well as other birth defects.**

Encephalomalacia (a.k.a: Cerebromalacia)

- **Abnormal softness of the cerebral parenchyma often due to ischemia or infarction.**

Encephalopathy, Congenital

- **Any disorder of the brain.**

Encephalopathy, Hypoxic Ischemic

- **A disorder of the brain.**

Encephalopathy, Static

- **A disorder of the brain.**

Encephalomyelitis

- **Mental retardation, partial paralysis, seizure activity, inflammation of the spinal cord and brain.**

***Epilepsy**

- **Repeated seizures that may result in mental retardation, motor delays.**

***Erb's Palsy**

- **Usually temporary weakness in one of the upper arms; paralysis is possible.**
- **Supportive documentation need to substantiate degree and severity.**

F

***Fahr's Disease**

- **Neurological disorder, progressive deterioration of cognitive and motor skills, may affect vision.**

Farber Disease

- **Neurological disorder; impaired motor, mental abilities, children diagnosed with classic Farber's Disease expires prior to age 2.**

***Familial Dysautonomia (FD)**

- **May present with feeding issues, vision problems (corneal abrasions), and speech and motor delays.**
- **Supportive documentation needed.**

Fetal Alcohol Syndrome (FAS)

- **Mental retardation, motor delays, ADHD, and possible vision problems.**

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***Fetal Valproate Syndrome (FVS)**

- **Supportive documentation needed to determine if developmental milestones are being met.**

***Fetal Hydantoin Syndrome**

- **Supportive documentation needed to determine if developmental milestones are being met.**

FG Syndrome

- **Hearing loss, vision problems, and developmental delays particularly in speech.**

Fountain Syndrome

- **Mental retardation; speech and language concerns.**

Fragile X Syndrome

- **Cognitive problems and varying degrees of mental retardation demonstrated.**

***Frederick's Ataxia (a.k.a. Fredrick's Ataxia, FA, FRDA)**

- **Hearing loss, muscle weakness, loss of coordination, slurred speech, and mental status is not affected.**
- **Although rare, some documented cases occurring prior to age 3.**
- **Supportive documentation needed.**

Fryns Syndrome

- **Congenital abnormalities at birth; varying degrees of mental retardation, Cleft Palate.**

G

Gillespie Syndrome

- **Vision problems: request a functional vision assessment, may have motor problems.**

Goldenhar Syndrome

- **Mild to moderate mental retardation, speech and language issues.**

Goodman Syndrome

- **Malformation of head and face. Mental retardation of varying degrees. A variant of Carpenter's Syndrome.**

Gordon Syndrome

- **Motor problems- permanent stiffness of joints.**

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H

Hallgren Syndrome

- **Vision problem; congenital deafness, neurological problems.**

Hearing Loss (diagnosed with bilateral permanent hearing loss)

- **A permanent mild bilateral hearing loss exists when the diagnosis indicates there is in both ears, a calculated or predicted average pure tone air conduction threshold at 0.5, 1, 2, kHz between 20 and 40 decibels hearing level (dB HL or pure tone air conduction thresholds greater than 25 dB HL at two or more frequencies above 2 kHz. (http://www.cdc.gov/hcbddd/ehdi/documents/unilaterial)**

Hearing Loss (diagnosed with unilateral permanent hearing loss)

- **A permanent unilateral hearing loss exists when the diagnosis indicates there is a calculated or predicted average pure tone air conduction threshold at 0.5, 1, 2 kHz of any level greater than or equal to 20 dB HL or pure tone air conduction thresholds greater than 25 dB HL at two or more frequencies above 2 kHz in the affected ear with an average pure tone air conduction threshold in the good ear less than or equal to 15dB. (http://www.cdc.gov/hcbddd/ehdi/documents/bilaterial)**

Hearing Loss, Mixed

- **Conductive and sensori-neural hearing loss.**

Hemianopia (a.k.a. Heminanopsia)

- **Blindness that affects half of the field of vision.**
- **This condition may be caused by a variety of medical conditions including strokes or brain injury. It can either affect the right or left side of the visual field and is usually permanent.**

***Hemiparesis**

- **Weakness on one side of the body.**
- **Supportive documentation needed to substantiate degree and which side of the body is affected.**

Hemiplegia

- **Total paralysis of arm, leg and trunk on one side of the body.**

Hereditary Deafness and Nephropathy (a.k.a. Alport Syndrome)

Hereditary Nephritis with Sensory Deafness (a.k.a. Alport Syndrome

***Human Immunodeficiency Virus (HIV) (+++)**

- **A serious virus that damages the immune system and attacks the brain, resulting in developmental delays and increased susceptibility to infection. HIV is transmitted**

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when the virus enters the bloodstream, and can be passed to a fetus by his/her mother. HIV causes AIDS.
- **Eligibility will be decided on a case by case basis based on supportive documentation of developmental concerns.**
- **DO NOT WRITE THIS DIAGNOSIS ON ANY DOCUMENTATION.**

Hirschsprung's Disease
- **Mental retardation, failure to thrive.**

Holoprosencephaly
- **Mental retardation, Cleft Palate, Cleft Lip, progressive hearing loss, deafness. Severest form of this diagnosis, expire before birth.**
- **Those with less severe form of this diagnosis generally have vision problems.**
- **See Trisomies 13, 18, and 13Q syndromes.**

Houston-Harris Type
- **See Achondrogenesis.**

Hunter Syndrome
- **An inborn error of metabolism with mental retardation (varying in severity).**
- **Accompanied by skeletal deformities, facial deformations, macrocephaly, and progressive hearing loss**.

Hurler Syndrome
- **Inborn error of metabolism accompanied by macrocephaly and severe mental retardation.**
- **Additional characteristics include course facial features, crouched stance, thickened digits (fingers and toes) and protuberant abdomen.**

Hydranencephaly
- **Vision problems; seizures; motor problems**

Hydrocephaly, Hydrocephalic
- **Characterized by a progressive loss of acquired motor coordination if not corrected.**
- **May present with mental and physiological problems.**
- **This diagnosis can accompany other conditions or diagnoses.**

I

Infantile Cerebral Palsy
- **The brain is involved and it's ability to control muscles.**
- **There are difficulties in mobility and communication.**
- **There may also be associated problems with cognitive abilities, behavioral responses, and vision.**

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Infantile Spinal Muscular Atrophy

- **See Werdnig Hoffman Syndrome**

***Intraventricular Hemorrhage (IVH)**

- **Grades 3 and 4 are the only two that would be eligible.**
- **Supportive documentation needed to substantiate grade.**

J

Jervell and Lange-Nielsen Syndrome

- **Cardiac problem; sudden death.**

Johanson-Blizzard Syndrome

- **Motor problems, hearing impairment, mental retardation of varying degrees.**

Joubert Syndrome

- **Rare brain malformation.**
- **Common features are abnormally rapid breathing, jerky eye movements, mental retardation and the inability to coordinate voluntary muscle movements.**
- **Physical abnormalities that may also be present include extra digits (fingers or toes), Cleft Lip/Palate, abnormal tongue, and seizures.**

Juberg-Marsidi Syndrome

- **Full manifestation in males; severe mental retardation, motor problems.**

K

Kabuki Syndrome

- **Mental retardation, mild to moderate, behavioral issues, hearing impairments, autistic like behaviors, etc.**

Kanner Syndrome

- **Alternate name for autism**

KBG Syndrome

- **Mental retardation of varying degrees**

Kearns Sayre Syndrome

- **A neuromuscular disorder.**
- **Progressive limitation of eye movements until there is complete immobility, accompanied by eye lid droop.**
- **Also associated with this disorder is abnormal accumulation of pigmented material on the membrane lining the eyes, which leads to vision problems.**

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- **Additional characteristics include: mild skeletal muscle weakness, cardiac conduction defect, and short stature, hearing loss, inability to coordinate voluntary movements, impaired cognitive functions, and diabetes.**
- **Prognosis varies; progression of disorder is slow.**

Keratitis Ichthyosis Deafness Syndrome

- **Rare congenital disorder characterized by vascularizing Keratitis, sensorineural hearing loss.**
- **Due to the Keratitis, visual problem leading to blindness.**

Kernicterus

- **Brain damage that causes athetoid cerebral palsy, hearing loss and sometimes mental retardation.**
- **Supportive documentation needed only if cerebral palsy, hearing loss or mental retardation is not evident, but suspected.**

***Klinefetters Syndrome**

- **Males are affected and may have learning difficulties. Females unaffected.**
- **Supportive documentation needed.**

Krabbe Disease

- **Progressive loss of motor and mental abilities.**
- **Infantile Krabbe diagnosis fatal prior to age 2.**

Kugelberg-Welander Syndrome

- **See Spinal Muscular Atrophy**
- **Motor abilities affected neuromuscular disorder.**

L

Landau Kleffner Syndrome

- **Sudden or gradual inability to understand or use spoken language. Normal intelligence.**

Langer-Saldino Type:

- **See Achondrogenesis**

Langer-Giedion Syndrome (a.k.a. Trichorhinophalangeal Syndrome, Type II and Type I)

- **This is a multi system disorder involving the deletion of at least 2 genes on the long arm of chromosome 8.**
- **Craniofacial abnormalities which include: large laterally protruding ears, broad nasal bridge and bulbous nose, elongated upper lip with thin upper vermillion border, broad eyebrows, sparse hair, mild microcephaly.**

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- **Additional characteristics which may or may not be present include: mental retardation, short stature, overly flexible joints, and excess folds of skin.**

Lawrence Moon Biedl Syndrome

- **Mental retardation, may have Retinitis Pigmentosa and other vision problems, there may also be paralysis of the legs.**

www.familyvillage.wisc.edu/lib_lmbb.htm

Leber Congenital Amarosis (LCA)

- **Severe visual problems and possible central nervous system abnormalities.**
- **May be accompanied by other diagnoses and mental retardation.**

Legal Blindness

- **A level of visual impairment that has been defined by law to determine eligibility for benefits.**
- **It refers to central visual acuity of 20/200 or less in the better eye with the best possible correction as measured on a Snellen vision chart, or a visual field of 20 degrees or less.**

Leigh's Disease

- **Inborn error of metabolism, mental retardation.**

Lesch-Nyhan Syndrome

- **Inborn error of metabolism accompanied by neurological problems, mental retardation and self injurious behaviors. This diagnosis is frequently misdiagnosed as cerebral palsy.**
- http://www.ninds.nih.gov/disorders/lesch-nyhan/lesch-nyhan.htm?css=print

Lennox Gastaut Syndrome

- **Severe form of epilepsy.**

Levy-Yeboa Syndrome

- **Multi-system disorder.**

Lissencephaly

- **Delayed psychomotor development, physiological abnormalities, failure to thrive.**

Lobster Claw

- **Deformity of the extremities which cause deep clefts in the anterior of the hand. May have webbing of extremities. Physical therapy is indicated.**

Locked In Syndrome

- **Complete paralysis of all voluntary muscles in all parts of the body except those that control eye movements.**

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Low Vision

- **Diagnosis of mild to moderate low vision.**
- **Low vision is visual acuity of between 20/70 to 20/200 in better eye with correction.**

Lowe Syndrome

- **Inborn error of metabolism; mental retardation, vision problems.**

M

***Macrocephaly**

- **Sometimes accompanied by mental retardation.**
- **Supportive documentation needed**

Maple Syrup Urine Disease

- **Inborn error of metabolism accompanied by mental retardation, feeding issues, lethargy, coma and seizures may develop**.

Marfan Syndrome

- **Bruises easily, has spider-like fingers, is tall and thin in stature, and has under developed muscles.**
- **May also have vision problems which include retinal detachment, displaced lens**.
- **Supportive documentation needed**.

Marinesco Sjogren Syndrome

- **Vision problems, ataxia, cognitive delays**

Marshall Syndrome

- **Mild to moderate hearing loss**

Menkes Syndrome (a.k.a. "Kinky Hair Syndrome")

- **Inborn error of metabolism, mainly affects males. May be accompanied by Failure to Thrive.**

Moebius Syndrome

- **Congenital abnormalities of the face, hands and feet that may benefit from physical therapy.**

Microcephaly

- **Mental retardation**, **small head, sometimes Cerebral Palsy and seizures occur.**

Mohr's Syndrome

- **Neuromuscular disturbances, mental disturbances, Cleft Lip/Palate**

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Muscular Dystrophy (MD)

- **Becker's Muscular Dystrophy: Muscle weakness in legs and pelvis which is associated with loss of muscle mass. Muscle weakness occurs in arms, neck, and other areas but not as severely as the lower half of the body. There may be cognitive issues. Age of onset is between 2 and 16 years of age.**
- **Congenital Muscular Dystrophy: Symptoms include general muscle weakness and possible joint deformities; disease progresses slowly; shortened life span. Present at birth.**
- **Duchenne's Muscular Dystrophy: Rapidly progressive form of Muscular Dystrophy. Loss of muscle function which begins in the lower limbs. There may be frequent falls. Progression of muscle weakness of the legs and pelvis. Age of onset is between two and 6 years of age.**
- **There are additional forms of this disorder however they are not evident in the zero to three populations.**

N

Nephritis and Nerve Deafness, Hereditary

- **See Alport Syndrome**

Neurological Visual Impairment(a.k.a. Cortical Visual Impairment)

- **See Cortical Visual Impairment**

www.sfsu.edu/cadbs/Eng022.html

Neuroblastoma

- **This is the most common form of cancer found in infants and young children.**
- **It is sometimes found prenatally, however, in most instances it is found after birth.**
- **Child may experience developmental delays due to numerous hospitalizations.**

Neurofibromatosis (NFI)

- **Orthopedic problems and possible hearing problems.**
- **Supportive documentation needed.**

Niemann-Pick Disease

- **Affects the body's metabolism. Four types: Type A: Severe brain damage; Type B: No brain damage; Type C: Severe brain damage; Type D: Only occurs in families who have a forefather who came from Nova Scotia at the start of 1700.**
- **Supportive documentation required to determine the type.**
- http://www.azornet.com/genetics/niemann-pick.asp
- http://www.ninds.nihds.nih.gov/disorders/niemann/niemann.htm?css=print
- **Types A and C result in severe brain damage and are eligible diagnoses.**

Noonan Syndrome

- **Hearing is affected**

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- **Supportive documentation needed to determine degree of hearing loss.**

Norrie's Syndrome

- **Blindness in both eyes (congenital), may have varying degrees of mental retardation, and hearing impairment.**

O

Ocular Albinism

- **Affects retina and iris.**
- **Functional vision assessment necessary.**

***Optic Nerve Atrophy**

- **Degeneration of the optic nerve which carries vision information from the eye to the brain.**
- **There may be dimmed or blurred vision as well as reduced field of vision.**

***Optic Nerve Hypoplasia**

- **Present at birth. Optic nerve is underdeveloped so there is not adequate information carried from the eye to the brain.**
- **There is a broad range from little to no affect on vision to total blindness.**
- **This condition can affect one or both eyes.**
- **No cure.**
- **Because of the range of effects to vision, supportive documentation is needed which would include medical reports and assessments from vision professionals.**

Oral Facial Digital Syndrome

- **See Mohr's Disease**

Osteogenesis Imperfecta (a.k.a "Brittle Bone Syndrome", "Glass House Syndrome")

- **Frequent fractures which may or may not be present at birth. Fractures can occur during a sneeze or coughing. Often mistaken for child abuse.**
- **Hearing deficits may be present.**

P

Pallister Killian Mosaic Syndrome

- **Mental retardation**
- **Sensory Hearing Loss**

Pallister W Syndrome

- **Mental retardation, cleft palate**

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Patau's Syndrome (a.k.a Trisomy 13)

- **Severe mental retardation, deafness, Cleft Plate/Lip, visual problems.**

Parenti-Fraccaro Type

- **See Achondrogenesis**

Partial Trisomy 6p

- **Mental retardation, multiple facial abnormalities and other physical concerns**.

Partial Trisomy 22, (a.k.a. Cat Eye Syndrome)

- **Severe mental retardation, vision problems**

Penta X Syndrome

- **May or may not have mental retardation; hearing loss.**
- **Supportive documentation needed to determine mental retardation. If child presents with a hearing loss of moderate loss or greater, supportive documentation is not needed.**

Peter's Anomaly

- **Vision problems and can occur in conjunction with other disorders including other vision disorders.**
- **May present with developmental delays, hearing loss, and Cleft Plate/Lip**.

Periventricular Leukomalacia (PVL)

- **Premature infants at greatest risk, although there are no outward signs, but at risk for motor delays, mental/cognitive delays, vision problems, hearing problems.**
- **Supportive documentation needed to document mental, cognitive delays.**
- **If hearing or vision problem is evident; supportive documentation is not needed.**

***Phenylketonuria (PKU)**

- **Only untreated or poorly treated PKU is eligible therefore supportive documentation is necessary.**
- **Inborn error of metabolism**

Phocomelia Syndrome

- **Severe birth defects especially of the upper limbs. Bones of the arms and in some cases other appendages may be extremely shortened or absent.**

Pierre Robin Syndrome

- **Cleft soft plate, possible Failure to Thrive, Dysphagia and apneic spells, and may occur in conjunction to other diagnoses.**

Pompe Disease

- **Rare and inherited disease. Prognosis is poor with death occurring during first year of life.**

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- **Will exhibit muscular weakness, respiratory problems, tonal issues, feeding difficulties, poor weight gain.**

Prader-Willi Syndrome

- **Mental retardation, visual problems, abnormal development of hands and fingers, hypotonia, and Failure to Thrive.**
- http://medgen.genetics.utah.edu/photographs/pages/praderwilli.htm

R

Rasmussen Encephalitis

- **Mental retardation, motor problems**

Retinoblastoma

- **Vision problems (Intraocular Retinoblastoma, Extraocular Retinoblastoma, and Recurrent Retinoblastoma.)**
- **Functional vision assessment needed**

Retinitis Pigmentosa (RP)

- **Eye disease which causes vision problems.**

***Retinopathy of Prematurity (ROP), a.k.a. Terry Disease; a.k.a. Retrolental Fibroplasia**

- **Stage 5 is eligible, child has no functional vision**
- **Supportive documentation is needed to determine stage of ROP**

Rett's Syndrome

- **Primarily seen in females.**
- **Between 6 and 18 months regression of skill acquisition decreases and regression occurs.**

Reye's Syndrome

- **Mental retardation, motor problems**
- **Supportive documentation needed**

Rieger Anomaly (Rieger Syndrome)

- **Vision problems, mental/cognitive issues may be present**
- **Functional assessment needed**

Robert's Syndrome

- **Mental retardation**

Rubella, congenital

- **Mental retardation , deaf-blindness, hearing impairment**
- **Vision professional must be on team**

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Rubinstein-Taybi Syndrome

- **Mental retardation of varying degrees, eye problems**

Russell Silver Syndrome

- **Growth deficiencies; may have learning disabilities.**

Rosenberg Chutorian Syndrome

- **Hearing loss, degeneration of the optic nerve, neurological abnormalities.**

Roussy Levy Syndrome

- **Motor problems; inability to coordinate motor abilities.**

***Ruvalcaba Syndrome**

- **Characterized by excessive growth before and after birth; head may be extremely large.**
- **May have mental retardation, speech and language issues as well as other concerns.**
- **Request supportive documentation to document mental retardation, speech and language concerns.**

S

***Saethre Chotzen Syndrome**

- **A craniosynostotic condition.**
- **Intelligence is usually normal; however there have been documented cases where mild to moderate mental retardation has been found as well as communication disorders, emotional problems,. Often confused with other diagnoses.**
- **Supportive documentation needed to document secondary complications which include hearing loss and or learning disabilities.**

Sandhoff's Disease

- **Mental retardation, central nervous system problems**

***Santavuori Disease**

- **A group of progressive degenerative neurometabolic diseases.**
- **Normal development up until 9 to 19 months, regression of skills occurs; developmental problems will be noted in motor area as well as in cognitive areas.**
- **Supportive documentation of diagnosis and regression needed.**

***Sandfilippo Syndrome**

- **May have communication problems, cognitive problems, hyperactivity**
- **Supportive documentation needed**

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Sensorineural Hearing Loss

- **Request speech evaluation, audiological assessment results.**

Schizencephaly

- **Mental retardation**
- **Cleft in skull**

***Shaken Baby Syndrome a.k.a Sudden Impact Syndrome**

- **Request medical reports to substantiate injuries and severity of injuries.**
- **MDE will determine eligibility.**

Sly Syndrome

- **Vision problems, motor problems, cognitive concerns, and other problems.**

Sotos Syndrome

- **Intellectual impairment, behavioral problems, tone problems—particularly in infancy. May also have hearing and vision problems.**

Smith-Lemlin-Opitz Syndrome

- **Mental retardation; sometimes autistic-like tendencies; behavior problems.**

Smith-Magenis Syndrome

- **Mental retardation, delayed speech, behavioral problems.**

***Spasmodic Torticollis (ST) (a.k.a. Cervical Dystonia, Torticollis)**

- **Spasmodic Torticollis is a painful and debilitating neurological movement disorder.**
- **This disorder is caused by a dysfunction of the brain.**
- **Symptoms are caused by intermittent or sustained contractions of the muscles around the neck which control the position of the head. This causes the head to lean to one side or be pulled forward or backward.**
- **There is no cure.**
- **Can be more severe during periods of stress or anxiety.**

***Spina Bifida with Hydrocephaly**

- **Degree of physical impairment varies depending on location of the hydrocele (cervical, dorsal, or lumbar).**
- **Supportive documentation needed.**

***Spina Bifida**

- **Congenital, incomplete closure of the spinal cord. Surgery and orthopedic devices and physical therapy are indicated in moderate to severe conditions.**

***Spinal Muscular Atrophy**

- **Atrophy in limbs and trunk resulting in a slumped forward posture. There is impaired motor function (uncoordinated gait). Vision may be affected.**

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Stargardt Disease

- **Most common form of inherited juvenile macular degeneration.**
- **Characterized by a reduction of central vision with a preservation of peripheral vision.**
- **Infants/children should wear protective sunglasses to preserve their vision during sun exposure.**
- **Vision problems.**
- **Functional vision assessment needed.**

Stickler Syndrome

- **Hearing, vision, Cleft Palate**

***Stroke**

- **Because of the various types of strokes which can affect many different parts of the brain and body, supportive documentation is needed.**

***Struge-Weber Syndrome**

- **Progressive seizure activity often accompanied by mental retardation, hemiparesis or hemiplegia and impaired vision.**
- **There is a port-wine stain around the face/eyes that is present at birth.**
- **Supportive documentation needed**

Struge-Kalischer-Weber Syndrome

- **See Struge-Weber Syndrome**

T

Tay Sachs Disease

- **Inborn error of metabolism**

Timothy Syndrome

- **Physical characteristics include webbing of fingers and toes. Child has an abnormal heart rhythm and abnormal amount of calcium in body.**
- **Have autistic like tendencies; speech is impaired.**

***Torticollis**

- **See Spasmodic Torticollis (ST)**
- **Supportive documentation needed to determine severity of condition.**

Townes Brocks Syndrome

- **Hearing loss**

Trisomy 16-18

- **Severe mental retardation**

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Trisomy 18p
- **Physical abnormalities**

Trisomy 21 (a.k.a. Down Syndrome, Trisomy G)
- **Mental retardation.**

Trisomy G
- **Mental retardation.**

Trisomy 8
- **Mental retardation ranging from mild to moderate.**

Trisomy 9p
- **Genetic disorder accompanied by mental retardation and other physical abnormalities.**

Trisomy 10q
- **Severe mental retardation; cleft plate.**

Trisomy 13q
- **Genetic disorder accompanied by severe mental retardation, brain abnormalities, midline anomalies, cleft lip/palate, polydactyly (extra digits), cardiac defects, microcephaly and deafness.**

Trisomy 13-15
- **See Trisomy 13**

Trisomy 18
- **Mental retardation as well as physical anomalies, difficulties with feeding, failure to thrive and hypotonia.**

U

Unilateral Blindness

Usher's Syndrome
- **Vision problems (retinitis pigmentosa, decreased visual acuity, depth perception problems, spotty vision, photophobia, reduced visual fields, cataracts, myopia), hearing impairment.**

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V

VACTERL with Hydrocephalus

- **Is an acronym for vertebral anomalies, anal atresia, congenital cardiac disease, tracheoesophageal fistula, renal anomalies, radial Dysplasia and other limb defects.**
- **This is an extremely rare genetic disorder.**
- **Those with this diagnosis often have bilateral and symmetrical radial ray abnormalities especially radial aplasia, imperforate anus and genital anomalies but other anomalies may occur. Cleft palate, pancreatic hypoplasia and agenesis of the corpus callosum have been observed in some cases.**
- **Few have survived beyond infancy and those who do are developmentally delayed and are physically delayed.**

Visual Impairment

- **Any visual impairment that is not correctable with treatment, surgery, glasses, and contact lenses.**
- **Low Vision (vision acuity of 20/70 to 20/200 in better eye with correction**).
- **Legally blind (visual acuity of 20/200 or less or field restriction of 20 degrees or less in the better eye with correction.**
- **Light perception only**
- **Totally Blind**

http://www.ed.gov.nl.ca/edu/pub/vi/Appe_.pdf

W

WAGR Syndrome

- **Eye abnormalities; mental retardation.**

Walker Warburg Syndrome

- **Considered to be a form of muscular dystrophy. See muscular dystrophy.**

Waardenburg Syndrome

- **Inherited disorder accompanied by varying degrees of hearing loss and changes in skin and hair pigmentation.**
- http://www.emedicine.com and http://www.devbio.com

Weaver Syndrome (a.k.a. Weaver-Smith Syndrome; Weaver-Like Syndrome, Included.)

- **Accelerated growth and osseous maturation, unusual craniofacial appearance, hoarse and low pitched cry, hypotonia, psychomotor retardation, exaggerated reflexes, slow development of voluntary movements.**

***Weil Marchesani Syndrome**

- **Vision impairments of varying degrees.**

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- **Supportive documentation.**
- **Functional vision assessment needed.**

***West Syndrome**

- **Form of epilepsy. Sometimes mental retardation is present.**
- **Supportive documentation needed.**

Williams Syndrome

- **Mild to moderate mental retardation.**

Werding-Hoffman Syndrome

- **Severest form of Muscular Atrophy.**
- **Infants who are symptomatic at birth may have respiratory distress and are unable to feed.**
- **Severe hypotonia and generalized weakness.**
- **May have feeding problems.**

Wolf-Hirchhorn Syndrome (Trisomy 4p)

- **Genetic disorder may have Microcephaly**

Z

Zellweger Syndrome

- **This disease is one of four related diseases called Peroxisome Biogenesis Disorders (PBD) which are part of a larger group of diseases know as Leukodystrophies. These are inherited conditions that damage the white matter of the brain and also affect how the body metabolizes particular substances in the blood and organ tissues.**
- **This form is the most severe of the PBD's.**
- **What will be noted in individuals with this disease is high levels of iron and cooper which builds up in the blood tissue, enlarged liver, facial deformities (high forehead), under developed eyebrow ridges, deformed ear lobes, neurological abnormalities, mental retardation and seizures.**
- **Infants with this disease lack muscle tone sometimes to the point that they are unable to move, swallow or suck.**
- **Vision and hearing problems may be observed.**

4p Syndrome

- **See Wolf-Hirschhorn Syndrome**

5p Syndrome

- **See Cri du Chat Syndrome**

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11q Syndrome

- **Genetic disorder accompanied by mental retardation and physical abnormalities.**

13q Syndrome

- **Genetic disorder accompanied by mental retardation and physical abnormalities.**

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Philadelphia, Pennsylvania: Birth-to-Three Early Intervention Eligibility Determination Guide

Ocular Manifestations

These syndromes and or conditions have a visual component. This listing was taken in part from: *Understanding and Managing Vision Deficits: A Guide for Occupational Therapists, 2nd edition*: Mitchell Scheiman, 2002, 212-213. This list is not inclusive of all ocular manifestations of syndromes with multiple impairments.

- **Absence of Corpus Callosum**
- **Aicardi Syndrome**
- **Apert's Syndrome**
- **Alstrom's Syndrome**
- **Andersen-Warburg's Syndrome**
- **Anophthalmia (Microphathalmia)**
- **Axenfeld's Anomaly**
- **Batten Disease**
- **Bronze Schilder Disease**
- **Canavan's Disease**
- **Cerebral Palsy**
- **CHARGE Syndrome/Association**
- **Cockayne Syndrome**
- **Coloboma**
- **Cornelia deLange Syndrome**
- **Cortical Visual Impairment (CVI)**
- **Cranial Mononeuropathy VI**
- **Crouson's Syndrome**
- **Cytomegalovirus (congenital)**
- **Dandy Walker Syndrome**
- **DeBarsy Syndrome**
- **DeSantis Cacchione Syndrome**
- **Ealses Disease**
- **Encephalocele**
- **Familial Dysautonomia**
- **Fetal Alcohol Syndrome**
- **FG Syndrome**
- **Fragile X Syndrome**
- **Gillespie Syndrome**
- **Hallgren Syndrome**
- **Hallerman Streiff Syndrome**
- **Hemianopia (Heminanopsia)**
- **Hydranencephaly**
- **Hydrocephaly/Hydrocephalus**
- **Joubert's Syndrome**
- **Kearns Sayre Syndrome**
- **Keratitis Ichthyosis Deafness**

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- **Kernicterus**
- **Laurence Moon Syndrome**
- **Leber Congenital Amarosis (LCA)**
- **Lowe Syndrome**
- **Marinesco Sjogren Syndrome**
- **Neurological Visual Impairment**
- **Norrie's Syndrome**
- **Ocular Albinism**
- **Optic Nerve Atrophy**
- **Optic Nerve Hypoplasia**
- **Patau's Syndrome**
- **Partial Trisomy 22 (Cat Eye Syndrome)**
- **Peter's Syndrome/ Peter's Anomaly**
- **Pierre Robin Syndrome**
- **Periventricular Leukomalacia (PVL)**
- **Rieger's Syndrome**
- **Retinoblastoma**
- **Retinitis Pigmentosa**
- **Retinopathy of Prematurity/Terry's Disease (only stage impacts on vision)**
- **Rieger Anomaly (Rieger Syndrome)**
- **Rubella (congenital)**
- **Rubinstein-Taybi Syndrome**
- **Septo-Optic Dysplasia (de Morsier's Syndrome)**
- **Shaken Baby Syndrome**
- **Sly Syndrome**
- **Spina Bifida**
- **Sotos Syndrome**
- **Stargardt Disease**
- **Stickler's Syndrome**
- **Sturge Webber Syndrome**
- **Toxoplasmosis (Congenital)**
- **Treacher Collins Syndrome**
- **Trisomy 18**
- **Trisomy 21 (Down's Syndrome)**
- **Usher's Syndrome**
- **WAGR Syndrome**
- **Weil Marchesani Syndrome**

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These conditions and/or syndromes <u>may be</u> accompanied by a visual problem:

- **Congenital Rubella**
- **Congenital Toxoplasmosis**
- **DeMorsier's Syndrome**
- **Down's Syndrome/Trisomy 21/ Trisomy G**
- **Encephalitis**
- **Fahr's Disease**
- **Fetal Alcohol Syndrome**
- **Infantile Cerebral Palsy**
- **Lawrence Moon Biedl Syndrome**
- **Marfan Syndrome**
- **Optic Nerve Hypoplasia (This diagnosis may cause significant visual problems or can cause no visual problems.)**
- **Periventricular Leukomalacia (PVL)**
- **Soto's Syndrome**
- **Spinal Muscular Atrophy**
- **Zellwegger Syndrome**

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The following diagnoses affect hearing abilities:

- **Absence of Corpus Callosum**
- **Alstrom's Syndrome**
- **Andersen-Warburg's Syndrome**
- **Anencephaly**
- **Apert Syndrome**
- **Bjornstad Syndrome (with hearing impairment)**
- **Bilateral Cleft Palate/Lip**
- **Canavan's Disease**
- **CHARGE Syndrome**
- **Cogan Syndrome**
- **Congenital Rubella**
- **Cytomegalovirus/ Cytomegalovirus Inclusion Disease (CID)**
- **DiGeorge Syndrome/Trisomy 22q11deletion**
- **Down's Syndrome/Trisomy 21/Trisomy G**
- **FG Syndrome**
- **Frederick's Ataxia/Fredrick's Ataxia, FA, FRDA**
- **Hallgren Syndrome**
- **Holoprosencephaly**
- **Hunter Syndrome**
- **Johnson-Blizzard Syndrome**
- **Kearns Sayre Syndrome**
- **Keratitis Ichthyosis Deafness Syndrome**
- **Kernicterus**
- **Marshall Syndrome**
- **Neurofibromatosis (NFI)**
- **Noonan Syndrome**
- **Norrie's Syndrome**
- **Osteogenesis Imperfecta/Brittle Bone Syndrome/Glass House Syndrome**
- **Pallister Killian Mosaic Syndrome**
- **Penta X Syndrome**
- **Periventricular Leukomalacia (PVL)**
- **Rosenberg Chutorian Syndrome**
- **Saethre Chotzen Syndrome**
- **Stickler Syndrome**
- **Townes Brocks Syndrome**
- **Trisomy 13q**
- **Usher Syndrome**
- **Zellweger Syndrome**

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Websites

The websites identified below were all functioning as of July 1st, 2008. These websites are either university based websites or government based websites. Please report any non functioning websites so that I might contact the web provider and remove website from future editions of this guide.

1. **Genetic and Rare Conditions Site**
http://www.kumc.edu/gec/support

This website is an excellent resource which houses various syndromes and genetic conditions as well as links to resources that may be helpful.

2. **National Center for Biotechnology Information**
http://www.ncbi.nlm.nih.gov

3. **DNA Learning Center**
http://www.yourgenesyourheath.org/ygyh/mason/index

4. **Genetic Alliance**
http://www.genticalliance.org

5. **Mayo Clinic**
http://www.mayoclinic.com

6. **Gene Gateway-Exploring Genes and Genetic Disorders**
http://www.ornl.gov

7. **National Human Genome Research Institute**
http://www.genome.gov

8. Genetics Home Reference: Your Guide to Understanding Genetic Conditions
http://ghr.nlm.nih.gov/condition

9. The Deafness and Family Communication Center at the Children's Hospital of Philadelphia
www.raisingdeafkids.org

10. The National Association for Parents with Visual Impairments
www.napvi.org

11. The Pennsylvania Training and Technical Assistance Network
www.pattan.org

12. The Parent Education and Advocacy Leadership Center
www.pealcenter.org

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13. American Federation of the Blind
www.afb.org

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