National Center on Deaf-Blindness

# Risk List for Combined Vision and Hearing Loss in Infants and Toddlers
## Associated Etiologies with Corresponding ICD-10 Codes

*This list has been developed for use by Part C Service Coordinators* in identifying children who are eligible for referral to their state's DeafBlind project. When one or more of the conditions in the tables below are present in infants and toddlers who have been determined eligible for Part C services, or who are being evaluated for eligibility, it is important that Service Coordinators work closely with families, early intervention providers and medical professionals to obtain accurate vision and hearing evaluations. These evaluations should include functional vision and hearing assessments in addition to medical assessments.

Referring a childbirth through two years of age to the state DeafBlind project begins the process of determining whether the child is considered DeafBlind. It is important to remember that "DeafBlindness" encompasses a wide diversity of children and conditions. The term describes any combination of vision and hearing loss that negatively impacts a child's ability to access environmental information, communicate and interact with others. Only a small percentage of children considered DeafBlind are totally deaf and blind. Most have varying degrees of residual vision and hearing and over 90% have additional disabilities.

Referral to a state DeafBlind project allows for children who qualify as DeafBlind to be counted in an annual National Child Count that is shared with Project Directors from the Office of Special Education (OSEP) Technical Assistance & Dissemination Network. Referral also qualifies early intervention providers and families to receive a variety of technical assistance services, including print and web resources, family support and consultation with experts on effective early intervention practices for children birth through two who have conditions that affect both hearing and vision.

***Please review this list carefully to identify children who may have both vision and hearing loss. For additional information please contact:***


**Florida & Virgin Islands DeafBlind Collaborative**
**University of Florida**
**PO Box 100234**
**Gainesville, FL 32610**

**800-667-4052**
**352-273-8539 fax**
**info@deafblind.ufl.edu**
**https://deafblind.ufl.edu**



The information provided was developed under a grant from the U.S. Department of Education #H326T130013. However, those contents do not necessarily represent the policy of the Teaching Research Institute, nor the US Department of Education, and you should not assume endorsement by the Federal Government. Project Officer, Jo Ann McCann.



*This resource was developed by a collaborative Early Identification & Referral Work Group. Thank you to Diane Haynes (KY Services for Children and Youth Who Are Deaf-Blind), Ruth Ann King (WV SenseAbilities), Sam Morgan (NY Deaf-Blind Collaborative), Gail Olson (IL Hearing/Vision Early Intervention Outreach), Karen Windy (IL Project Reach), and Mark Schalock (NCDB). With special appreciation to Kentucky CHFS OATS MSMP ICD-10 Project Manager.*


*Links checked: March 2010*

NATIONAL CENTER ON DEAF-BLINDNESS

# *Risk List for Combined Vision and Hearing Loss in Infants and Toddlers*
## *Etiologies Associated with Combined Vision and Hearing Loss*

## Section A: Diagnoses Associated with Sensory Loss

| Dual Sensory Impairment | | | |
|---|---|---|---|
| *This code indicates that both vision and hearing or absent or affected, which would qualify a child as DeafBlind. Immediate referral to your state's DeafBlind project is recommended to ensure appropriate intervention strategies and family support.* | | | |
| **ICD Code** | | | |
| Z73.82 | Dual Sensory Impairment | | |
| **Blindness/Vision Impairment** | | **Deafness/Hearing Loss** | |
| **ICD Code** | | **ICD Code** | |
| H54.0 | Blindness (acquired) (congenital) (both eyes) | H91.9 | Deafness (acquired) (complete) (hereditary) (partial) |
| H54.3 | Vision Impairment or Vision Loss (both eyes) | H90.2 | Deafness – conductive |
| H54.8 | Legal blindness (both eyes) (USA definition) | H90.5 | Deafness – congenital or sensorineural |
| H47.619 | Cortical blindness (cortical visual impairment) | H90.8 | Deafness – mixed conductive and sensorineural |
| H35.10 | Retinopathy of prematurity | H90.5 | Hearing Loss or Hearing Impairment – congenital or sensorineural |
| H53.003 | Amblyopia (bilateral( (both eyes) | | |
| E70.3 | Albinism | | |

## Section B: Hereditary/Chromosomal Syndromes and Disorders

These tables include ICD-10-CM Codes

*Presence of a listed etiology does not automatically qualify a child as DeafBlind; rather it indicates further evaluation to determine whether a combined vision and hearing loss exists. Immediate referral to your state's DeafBlind project can help make that determination.*

(Column 1 = NCDB National Child Count Code, Column 2 = Corresponding ICD-10 Code)

| Code | ICD Code | Primary Identified Etiology |
|---|---|---|
| 101 | Q04.0 | Aicardi syndrome |
| 102 | Q87.81 | Alport syndrome |
| 103 | Q87.8 | Alstrom syndrome |
| 104 | Q87.0 | Apert syndrome (Acrocephalosyndactyly, Type 1) |
| 105 | Q87.89 | Bardet-Biedl syndrome (Laurence Moon-Biedl) |
| 106 | E75.4 | Batten disease |
| 107 | Q89.8 | CHARGE Syndrome |
| 108 | Q93.2 | Chromosome 18, Ring 18 |
| 109 | Q87.1 | Cockayne syndrome |
| 110 | H16.32 | Cogan Syndrome |
| 111 | Q87.1 | Cornelia de Lange |
| 112 | Q93.4 | Cri du chat syndrome (Chromosome 5p-syndrome) |
| 113 | E80.5 | Crigler-Najjar syndrome |
| 114 | Q75.1 | Crouzon syndrome (Craniofacial Dysotosis |
| 115 | Q03.1 | Dandy Walker syndrome |
| 116 | Q90.9 | Down syndrome (Trisomy 21 syndrome) |
| 117 | Q87.0. | Goldenhar syndrome |
| 118 | C96.5 | Hand-Schuller-Christian (Histiocytosis X |
| 119 | H35.5 | Hallgren syndrome |
| 120 | B02 | Herpes-Zoster (or Hunt) |
| 121 | E76.1 | Hunter Syndrome (MPS II) |
| 122 | E76.01 | Hurler syndrome (MPS I-H |
| 123 | H49.81 | Kearns-Sayre syndrome |
| 124 | Q76.1 | Klippel-Feil sequence |
| 125 | Q87.2 | Klippel-Trenaunay-Weber syndrome |
| 126 | Q77.8 | Kniest Dysplasia |
| 127 | H35.50 | Leber congenital amaurosis |
| 128 | G31.82 | Leigh Disease |
| 129 | Q87.40 | Marfan syndrome |

(Column 1 = NCDB National Child Count Code, Column 2 = Corresponding ICD-10 Code)

| Code | ICD Code | Primary Identified Etiology |
|---|---|---|
| 130 | Q87.0 | Marshall syndrome |
| 131 | E76.29 | Maroteaux-Lamy syndrome (MPS VI) |
| 132 | Q87.0 | Moebius syndrome |
| 133 | Q93.0 | Monosomy 10p |
| 134 | E76.219 | Morquio syndrome (MPS IV-B)( |
| 135 | Q85.01 | NF1 - Neurofibromatosis (von Recklinghausen disease) |
| 136 | Q85.02 | NF2 - Bilateral Acoustic Neurofibromatosis |
| 137 | H35.5 | Norrie disease |
| 138 | G25.9 | Optico-Cochleo-Dentate Degeneration |
| 139 | Q87.0 | Pfieffer syndrome |
| 140 | Q87.1 | Prader-Willi |
| 141 | Q87.0 | Pierre-Robin syndrome |
| 142 | G60.1 | Refsum syndrome |
| 143 | E76.03 | Scheie syndrome (MPS I-S) |
| 144 | E78.72 | Smith-Lemli-Opitz (SLO) syndrome |
| 145 | Q8709 | Stickler syndrome) |
| 146 | Q85.8 | Sturge-Weber syndrome |
| 147 | Q75.4 | Treacher Collins syndrome |
| 148 | Q91.7 | Trisomy 13 (Trisomy 13-15, Patau syndrome) |
| 149 | Q91.3 | Trisomy 18 (Edwards syndrome) |
| 150 | Q96.9 | Turner syndrome |
| 151 | H35.5 | Usher I syndrome |
| 152 | H35.5 | Usher II syndrome |
| 153 | H35.5 | Usher III syndrome |
| 154 | H20.829 | Vogt-Koyanagi-Harada syndrome |
| 155 | E70.3 | Waardenburg syndrome |
| 156 | Q75.4 | Wildervanck syndrome |
| 157 | Q93.3 | Wolf-Hirschhorn syndrome (Trisomy 4p) |
| 199 | | *Other* |

## Section C: Pre-Natal Complications, Post-Natal Complications and Complications of Prematurity

These tables include ICD-10-CM Codes

| Pre-Natal/Congenital Complications (Column 1 = NCDB National Child Count Code, Column 2 = Corresponding ICD-10 Code) | | | Post-Natal/Non-Congenital Complications (Column 1 = NCDB National Child Count Code, Column 2 = Corresponding ICD-10 Code) | | |
|---|---|---|---|---|---|
| *Presence of a listed etiology does not automatically qualify a child as DeafBlind; rather it indicates further evaluation to determine whether a combined vision and hearing loss exists. Immediate referral to your state's DeafBlind project can help make that determination.* | | | | | |
| **Code** | **ICD Code** | **Primary Identified Etiology** | **Code** | **ICD Code** | **Etiology** |
| 201 | P35.0 | Congenital Rubella | 301 | R09.01 | Asphyxia |
| 202 | A50 | Congenital Syphilis | 302 | H93.19 | Direct Trauma to the eye and/or ear |
| 203 | P37.1 | Congenital Toxoplasmosis | 303 | G04.81 | Encephalitis |
| 204 | B25.9 | Cytomegalovirus (CMV) | 304 | P00.2 | Infections |
| 205 | Q86.0 | Fetal Alcohol syndrome | 305 | G03.9 | Meningitis |
| 206 | G91.9 | Hydrocephaly | 306 | S09.8XXA | Severe Head Injury |
| 207 | P04.49 | Maternal Drug Use | 307 | O99.43 | Stroke |
| 208 | Q02 | Microcephaly | 308 | O48.0 | Tumors |
| 209 | P35.2 | Neonatal Herpes Simplex (HSV) | 309 | E09.22 | Chemically Induced |
| 299 | O99 | Other ____ | 399 | O99 | Other _____ |
| **Related to Prematurity** | | | | | |
| Code | ICD Code | | **Code** | **ICD Code** | |
| 401 | O60.10X0 | Complications of Prematurity | | H35.10 | Retinopathy of prematurity (any stage) |

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TA&D NETWORK

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IDEAs that Work
Office of Special Education Programs
U.S. Department of Education

*Links checked: March 2010*

# ***Etiologies Related to DeafBlindness***

***This is a list of syndromes and conditions that may cause a combined vision and hearing loss. Keep in mind, the majority of causes of DeafBlindness are still unknown.***

***Always a good place to start to learn more:***

**National Consortium on DeafBlindness: http://www.nationaldb.org**

| **SYNDROMES and DISEASES<br>- PRIMARY CHARACTERISTICS -** |
|---|
| **Aicardi Syndrome**<br>• Absence of corpus callosum, either partial or complete (the corpus callosum allows the right side of brain to communicate with the left side).<br>• Infantile spasms<br>• Lesions or lacunae of the retina<br>• Microcephaly (abnormally small head)<br>• Porencephalic cysts (inside the brain tissue)<br>• Only affects females except in males with Klinefelter Syndrome (XXY)<br>**WEBSITE: http://www.aicardisyndrome.org/** |
| **Alport Syndrome**<br>• X-linked disorder<br>• Hereditary kidney damage<br>• Nerve deafness<br>• Congenital eye abnormalities<br>• Ankle, feet, and leg swelling<br>**WEBSITE: http://www.alportsyndrome.org/index.html** |
| **Alstrom Syndrome**<br>• Photophobia (light sensitivity) in infancy<br>• Nystagmus (wobbling of the eyes)<br>• Congestive heart failure (CHF)<br>• Childhood obesity<br>• Blindness from progressive pigmentary retinopathy<br>• Mild to moderate bilateral sensorineural hearing loss<br>• Type II diabetes<br>• Heart failure<br>• Liver disease<br>• Pulmonary fibrosis<br>• Renal failure<br>• Progressive disease<br>• Normal intelligence<br>**WEBSITE: http://www.alstrom.org** |
| **Apert Syndrome**<br>• Prematurely fused cranial structures<br>• A reruded midface<br>• Fused fingers and toes<br>• Various heart defects<br>• Pulmonary atresia<br>• Tracheoesophageal Fistula<br>• Sleep apnea<br>• Ear infections<br>• Severe acne<br>• Increased incidence of eye injuries<br>**WEBSITE: http://www.apert-international.org/** |
| **Bardet-Biedl Syndrome (BBS)**<br>• Obesity |

*Links checked: March 2010*

## SYNDROMES and DISEASES
## - PRIMARY CHARACTERISTICS -

- Pigmentary retinopathy
- Plydactyly
- Hypogonadism
- Renal failure
- Mental retardation

**WEBSITE:** http://mlmorris.com/lmbbs/

**Batten Disease**

- Mental retardation
- Seizures
- Progressive loss of sight
- Progressive loss of motor skills
- Fatal

**WEBSITE:** http://www.bdsra.org/

**CHARGE Syndrome**

- Coloboma of the eye
- Choanal atresia or stenosis
- Cranial nerve dysfunction – lack of smell, swallowing difficulties, facial palsy
- Malformed inner ear
- Significant balance problems
- Cleft lip and/or palate
- Short stature

**WEBSITE:** http://www.chargesyndrome.org/

**Ring 18 Syndrome**

- Mental retardation
- Microcephaly (abnormally small head)
- Hypertelorism
- Speech deficit
- Deafness
- Heart anomalies
- Poor muscle tone

**WEBSITE:** http://www.trisomy18.org/

**Cockayne Syndrome**

- Dwarfism
- Microcephaly (abnormally small head)
- Progressive neurodevelopmental delay
- Unsteady gait
- Sunburns easily
- Retinopathy and/or cataracts
- Progressive hearing loss
- Premature aging

**WEBSITE:** http://cockayne-syndrome.net/

**Cogan's Syndrome**

- Inflammation of the eye
- Hearing problems
- Dizziness
- Progressive disease

**WEBSITE:** http://www.coganssyndrome.info/

**Cornelia de Lange Syndrome (CdLS)**

- Small stature
- Microcephaly (abnormally small head)
- Excessive body hair
- Small hands and feet
- GERD
- Seizures

*Links checked: March 2010*

| SYNDROMES and DISEASES<br>- PRIMARY CHARACTERISTICS - |
|---|
| • Heart defects<br>• Cleft palate<br>• Developmental delays<br>• Missing limbs or portions of limbs<br>**WEBSITE:** http://www.cdlsusa.org/ |
| **Cri-du-Chat Syndrome**<br>• High pitched cry at birth<br>• Low birth weight<br>• Poor muscle tone<br>• Microcephaly (abnormally small head)<br>• Potential medical complications<br>**WEBSITE:** http://www.fivepminus.org/ |
| **Crigler-Najjar Syndrome**<br>• Very rare disorder<br>• Hyperbilirubinemia (must have daily 12 hour exposure to special blue lights)<br>• Jaundiced<br>**WEBSITE:** http://www.criglernajjar.com/ |
| **Crouzon syndrome**<br>• Craniaosynostosis<br>• Hypertelorism<br>• Exophthalmos<br>• Strabismus<br>• Beaked nose<br>• Short upper lip<br>• Hypoplastic maxilla<br>• Upper airway obstruction develops secondary to septal deviation<br>**WEBSITE:** http://www.crouzon.org/ |
| **Cytomegalovirus (CMV)**<br>• most common congenital infection<br>• low birth weight<br>• Microcephaly (abnormally small head)<br>• Seizures<br>• Rash – little red spots under the skin<br>• Enlarged liver and spleen (with jaundice)<br>• Abnormal muscle tone<br>**WEBSITE:** http://www.bcm.edu/pedi/infect/cmv/index.htm |
| **Dandy-Walker Syndrome**<br>• Slow motor development<br>• Progressive enlargement of the skull<br>• Convulsions<br>• Unsteadiness<br>• Lack of muscle coordination<br>• Jerky movements of the eyes<br>**WEBSITE:** http://www.dandy-walker.org/ |
| **Down Syndrome (Trisomy 21)**<br>• Smaller stature along with slower development physically and mentally<br>• Mental retardation<br>• Congenital heart disease<br>• Intestinal abnormalities<br>• Thyroid dysfunctions<br>• Skeletal problems<br>• Obesity in adolescence<br>• Small ear canals<br>**WEBSITE:** http://www.ndss.org/ |

*Links checked: March 2010*

## SYNDROMES and DISEASES
## - PRIMARY CHARACTERISTICS -

**Encephalitis**

- Inflammatory diseases of the membranes that surround the brain and spinal cord and are caused by bacterial or viral infections
- Can cause vision and hearing impairments

**WEBSITE:** http://www.ninds.nih.gov/disorders/encephalitis_meningitis/encephalitis_meningitis.htm

**Fetal Alcohol Syndrome**

- Prenatal exposure to alcohol
- Low birth weight
- Growth deficiencies for weight, height or both
- Face anomalies, including small eye slits, flat mid-face, short upturned nose, thin lips, and a smooth and/or long ridge that runs between the nose and lips
- Neurological damage, including small brain size, tremors, hyperactivity, learning disabilities
- Fine or gross motor problems
- Vision and hearing impairments

**WEBSITE:** http://www.nofas.org/

**Goldenhar Syndrome**

- Facial asymmetry, which may become more pronounced as the child gets older
- Underdevelopment of facial musculature on one side
- Mouth problems such as lack of saliva, problems in tongue shape or use
- Small or misshapen ears, sometimes no outer ear structure
- Skin tags or pits usually in front of the ear in line with the mouth opening
- Usually a unilateral hearing loss
- Speech problems, due to malformation of mouth and jaw, cleft lip and/or palate and facial muscles
- Spinal vertebrae which are small or not completely formed on one side.
- Eye defects, including one eye missing, benign growths on eye
- Cleft lip and/or palate

**WEBSITE:** http://www.goldenharsyndrome.org/

**Hand-Schüller-Christian disease (Histiocytosis)**

- Rare blood disease caused by an excess of white blood cells
- Failure to Thrive (FTT)
- Scaly, waxy rash on scalp
- Abdominal pain and jaundice, vomiting, diarrhea
- Bone pain, lesions on bones
- Limping
- Thirst and frequent urination
- Feeding problems in infants
- Short stature
- Delayed puberty
- Mental deterioration
- Seizures
- Vision problems and increased eyeball protrusion
- Inflamed ear canals, chronically draining ears, rash behind ears or on scalp

**WEBSITE:** http://www.histio.org

**Hallgren Syndrome (see Usher Syndrome or Alstrom Syndrome)**

**Herpes Zoster (Ramsey Hunt syndrome)**

- Reactivation of the dormant varicella-zoster virus (chicken pox)
- Shingles, can travel the affected nerves fibers to the eyes
- Can cause glaucoma, cataract, double vision, and scarring of the cornea and eyelids
- Can cause hearing loss, vertigo (abnormal sensation of movement), and tinnitus (abnormal sounds)
- Loss of taste and dry mouth

**WEBSITE:** http://www.ninds.nih.gov/disorders/ramsay2/ramsay2.htm **AND** http://www.stlukeseye.com/Conditions/HerpesZoster.asp

*Links checked: March 2010*

## SYNDROMES and DISEASES
## - PRIMARY CHARACTERISTICS -

**Hunter Syndrome (Mucopolysaccharidosis Type II or MPS II)**

- Short stature with progressive growth delays
- Joint stiffness
- Thickening of the lips, tongue, and nostrils
- Abnormally large head
- Cloudy corneas
- Progressive hearing loss
- Enlargement of the liver and spleen
- Mental retardation

**WEBSITE:** http://www.mpssociety.org/

**Hydrocephaly**

- Lower than average IQ
- Fine and gross motor problems
- Early puberty
- Blindness due to damage to pressure on the optic nerve
- "Sunset" eye, eyes fixed in a downward position
- Epilepsy

**WEBSITE:** http://www.hydroassoc.org/

**Kearns-Sayre Syndrome**

- Progressive limitation of eye movements until there is complete immobility
- Eyelid droop
- Mild skeletal muscle weakness
- Heart block
- Short stature
- Hearing loss
- Inability to coordinate voluntary movements
- Diabetes
- Impaired cognitive function

**WEBSITE:** http://www.ninds.nih.gov/disorders/kearns_sayre/kearns_sayre.htm

**Klippel-Feil Sequence**

- Short neck
- Low hairline at the nape of the neck
- Limited movement of the head
- Fusion of the cervical vertebrae
- Scoliosis

**WEBSITE:** http://health.groups.yahoo.com/group/klippelfeilsupport/ **AND** http://www.fortunecity.com/millenium/bigears/99/kfs.html

**Kniest Dysplasia**

- Short stature
- Malformed bones and joints
- Round, flat faces with prominent and widely set eyes
- Cleft palate
- Vision problems, especially severe nearsightedness (myopia)
- Hearing loss resulting from recurrent ear infections

**WEBSITE:** http://www.ksginfo.org/kniest.html

**Leber's Congenital Amaurosis**

- Retinal degenerative disease
- Reduced vision
- Nystagmus (shaky eyes)
- Roving eye movements
- Eye poking common
- Photophobia (sensitivity to light)
- Developmental delay
- Epilepsy

*Links checked: March 2010*

## SYNDROMES and DISEASES
## - PRIMARY CHARACTERISTICS -

- Motor skill impairment
- Sensorineural hearing loss

**WEBSITE: http://www.blindness.org/visiondisorders/**

**Leigh Disease**

- Feeding problems
- Vomiting
- Failure to thrive
- Delayed motor and language skills
- Seizures
- Generalized weakness
- Abnormal eye movements
- Droopy eyelids
- Respiratory and kidney problems
- Heart problems

**WEBSITE: http://www.ninds.nih.gov/disorders/leighsdisease/leighsdisease.htm**

**Marfan Syndrome**

- Disease of the connective tissue of the body
- Usually tall, slender, loose jointed
- Vision problems, resulting from disconnected lenses in one or both eyes
- Problems with the heart and blood vessels
- Lung problems (spontaneous collapse of lungs, emphysema)

**WEBSITE: http://www.marfan.org/marfan/**

**Marshall Syndrome**

- Flattened nasal bridge and short upturned nose
- Widely spaced eyes
- Short stature
- Nearsightedness (myopia), cataracts and glaucoma are common
- Hearing loss usually moderate to severe and is sensorineural

**WEBSITE: http://www.healthline.com/galecontent/marshall-syndrome-1**

**Maroteaux Lamy Syndrome**

- Symptoms not usually evident at birth
- Growth retardation – short stature
- Thickening of the nose, lips, and tongue
- Large head
- Joint stiffness
- Vision problems include clouding of the corneas, glaucoma, damage to the optic nerve or retina
- Hearing problems are caused by frequent ear infections
- Dental problems from poor enamel and small, widely spaced teeth

**WEBSITE: http://www.maroteaux-lamy.com/Index.aspx**

**Meningitis**

- Inflammatory diseases of the membranes that surround the brain and spinal cord and are caused by bacterial or viral infections
- Can cause vision and hearing impairments

**WEBSITE: http://www.ninds.nih.gov/disorders/encephalitis_meningitis/encephalitis_meningitis.htm**

**Chromosome 10, Monosomy 10p**

- Severe mental retardation
- Growth delays
- Malformations of the skull and facial region
- Short neck
- Congenital heart defects

**WEBSITE: http://www.peacehealth.org/kbase/nord/nord1030.htm**

**Moebius Syndrome**

- Unable to move facial muscles (to smile, frown, suck, blink)
- Unable to move eyes laterally

*Links checked: March 2010*

## SYNDROMES and DISEASES
## - PRIMARY CHARACTERISTICS -

- High palate, short or deformed tongue
- Feeding, swallowing and choking problems
- Drooling
- Hand and feet anomalies and/or club feet
- Upper body weakness, resulting in motor delays
- Hearing impairments
- Strabismus (crossed eyes)

**WEBSITE:** http://www.moebiussyndrome.com/

**Morquio Syndrome (MPS IV)**

- Short stature
- Coarse facial features
- Macrocephaly (abnormally large head)
- Knock-knees
- Widely spaced teeth
- Bell-shaped chest with ribs flared out at the bottom
- Hypermobile joints
- Compression of the spinal cord
- Cloudy cornea
- Liver enlargement
- Heart murmur

**WEBSITE:** http://www.mpssociety.org/content/4040/MPS_IV/

**Neurofibromatosis**

- Tumors on the nerves anywhere in the body
- Six or more café-au-lait spots
- Optic glioma (tumor of the optic pathway)
- Lisch nodules (benign iris hamartomas)
- Blindness
- Seizures
- Mental retardation
- Macrocephaly (abnormally large head)
- Scoliosis

**WEBSITE:** http://www.nfinc.org/

**Norrie Disease**

- Only males
- Bilateral blindness
- Abnormal development of the retina
- Pupils appear white when light is shone on them
- Mental retardation
- Progressive hearing loss
- Developmental delays in motor skills

**WEBSITE:** http://www.norries.org/ **AND** http://ghr.nlm.nih.gov/condition=norriedisease

**Pfeiffer Syndrome**

- Skull is prematurely fused and unable to grow normally
- Bulging wide-set eyes due to shallow eye sockets
- Underdevelopment of the midface
- Broad, short thumbs and big toes
- Possible webbing of the hands and feet

**WEBSITE:** http://www.faces-cranio.org/Disord/Pfeiffer.htm

**Prader-Willi Syndrome**

- Profound poor muscle tone
- Underdeveloped sex organs
- Short stature
- Retarded bone age
- Developmental delays

*Links checked: March 2010*

| SYNDROMES and DISEASES<br>- PRIMARY CHARACTERISTICS - |
|---|
| • Rapid weight gain between ages 1 and 6 leading to obesity<br>• Obesity with food<br>• Distinctive facial features: narrow face, almond-shaped eyes, small-appearing mouth with thin upper lip and down-turned corners of mouth<br>**WEBSITE: http://www.pwsausa.org/** |
| **Pierre Robin Sequence**<br>• Lower jaw is abnormally small, but usually grows out as individual ages<br>• Tongue is displaced downwards<br>• Cleft Palate<br>• Many ear infections, leading to hearing impairment<br>• Often present with another genetic disorder<br>• Breathing and feeding issues<br>**WEBSITE: http://www.pierrerobin.org/ AND http://www.faces-cranio.org/Disord/PierreRobin.htm** |
| **Infantile Refsum Syndrome (Peroxisomal Biogenesis Disorder: Zellweger and Neonatal Adrenoleukodystrophy)**<br>• Progressive loss of vision from retinitis pigmentosa<br>• Loss of smell<br>• Hearing loss from nerve damage<br>• Heart abnormalities<br>• Nerve disorder causing loss of sensation<br>• Ataxia (balance disorder)<br>• Ichthyosis (dry, scaly skin)<br>• Severe mental retardation<br>**WEBSITE: http://home.pacifier.com/~mstephe/** |
| **Scheie Syndrome**<br>• Corneal clouding<br>• Deafness<br>• Joint stiffness<br>• Coarse facial features<br>• Potential glaucoma<br>• Claw Hands<br>• Carpal tunnel syndrome<br>• Deformed feet<br>**WEBSITE: http://www.mpssociety.org/content/4021/MPS_I/** |
| **Smith-Lemli-Opitz syndrome**<br>• Psychomotor and growth retardation<br>• Cleft palate<br>• Hypospadias<br>• Microcephaly (abnormally small head)<br>• Ptosis<br>• Mental retardation<br>**WEBSITE: http://www.smithlemliopitz.org/** |
| **Stickler Syndrome**<br>• Myopia, cataracts, glaucoma, detached retinas, astigmitism<br>• Stiff joints and over-flexible joints, arthritis<br>• Cleft palate<br>• Flat face with a small nose and little or no nasal bridge<br>• Middle or inner ear hearing loss<br>• Scoliosis<br>• 30-40% also have Pierre Robin sequence<br>**WEBSITE: http://www.sticklers.org/sip2/** |
| **Sturge-Weber syndrome**<br>• Facial birthmark "Port Wine Stain," usually over the eye and forehead region<br>• Seizures, often starting by one year of age |

*Links checked: March 2010*

## SYNDROMES and DISEASES
## - PRIMARY CHARACTERISTICS -

- Weakening or loss of use of one side of the body (hemiparesis), usually on the opposite side of the port wine stain
- Developmental delay
- Glaucoma
- Growth hormone deficiency
- Severe headaches

**WEBSITE:** http://www.sturge-weber.org/

**Treacher Collins Syndrome**

- Cranio-facial birth defect, missing facial bones and muscles
- Hearing problems - underdeveloped, malformed and/or prominent ears
- Breathing problems
- Eating problems
- Down-slanting eyes
- Underdevelopment or absence of cheekbones and the side wall and floor of the eye socket
- Lower jaw is often small and slanting

**WEBSITE:** http://www.faces-cranio.org/Disord/Treacher.htm **AND** http://www.tcconnection.org/

**Patau Syndrome (Trisomy 13)**

- Heart defects (about 80%)
- Microcephaly (abnormally small head)
- Small eyes or absent eye
- Cleft lip and/or cleft palate
- Hearing loss
- Vision impairment
- Sleep apnea
- Gastroesophageal reflux (GERD)
- Seizures
- Developmental disabilities
- Kidney defects

**WEBSITE:** http://www.trisomy.org/trisomy13.php

**Edward Syndrome (Trisomy 18)**

- Congenital heart defects (over 90%)
- Hearing loss
- Spina bifida
- Feeding problems
- GERD
- Developmental disabilities
- Seizures
- Urinary tract infections
- Birth defects to the eye

**WEBSITE:** http://www.trisomy.org/trisomy18.php

**Turner Syndrome**

- Females only
- Short stature
- Lack of ovarian development
- Narrow, high arched palate
- Low set ears, low hair line
- Lazy eye (strabismus)
- Broad chest
- Cardiovascular problems
- Kidney problems
- Thyroid problems
- Scoliosis
- Hearing disturbances from ear infections (otitis media)

**WEBSITE:** http://www.turnersyndrome.org/

*Links checked: March 2010*

## SYNDROMES and DISEASES
## - PRIMARY CHARACTERISTICS -

**Usher Syndrome**

**Usher type I**

- Profoundly deaf from birth
- Severe balance problems from birth
- Vision problems, usually starting with decreased night vision, by age ten

**WEBSITE: http://www.blindness.org/visiondisorders/ AND http://www.familyvillage.wisc.edu/lib_ushe.htm**

**Usher type II**

- Moderate to severe hearing impairment at birth
- Vision loss varies in severity; decreased night vision begins in late childhood or teens
- Normal balance

**WEBSITE: http://www.blindness.org/visiondisorders/ AND http://www.familyvillage.wisc.edu/lib_ushe.htm**

**Usher type III**

- Normal hearing at birth, progressive loss in childhood or early teens
- Vision loss varies in severity; night vision problems often begin in teens
- Normal to near-normal balance, chance of problems later in life

**WEBSITE: http://www.blindness.org/visiondisorders/ AND http://www.familyvillage.wisc.edu/lib_ushe.htm**

**Vogt-Koyanagi-Harada Syndrome**

- Neurological abnormalities
- Auditory abnormalities
- Rapid vision loss
- Eye irritation
- Hearing loss
- Alopecia (hair loss)
- Vitiligo (loss of pigmentation in skin)

**WEBSITE: http://www.emedicine.com/oph/topic459.htm**

**Waardenburg Syndrome**

- Moderate to profound hearing loss
- Changes in hair and skin pigmentation
- White shock of hair or early graying
- Convergent strabismus (lazy eye)
- Microcephaly (abnormally small head)
- Two differently colored eyes – often one bright blue
- Wide space between inner corner of eyes
- Balance problems

**WEBSITE: http://www.nidcd.nih.gov/health/hearing/waard.asp**

**Wildervanck Syndrome**

- Primarily affects females
- Hearing impairment
- Nystagmus
- Fusion of two or more bones in the spinal column within the neck

**WEBSITE: http://children.webmd.com/wildervanck-syndrome**

**Wolf-Hirschhorn Syndrome**

- Severe growth and mental deficiency
- Microcephaly (abnormally small head)
- Wide space between inner corner of eyes
- "Greek Helmet" like noses
- Low set malformed ears
- Cleft lip and/or palate
- Coloboma of the eye
- Heart defects

**WEBSITE: http://www.4p-supportgroup.org/general.html**

*Links checked: March 2010*

**Resources:**
National Consortium on DeafBlindness (NCDB) – http://www.nationaldb.org
Sense - http://www.sense.org.uk/
Texas School for the Blind and Visually Impaired (TSBVI) - http://www.deafblind.com/downmoss.html
Washington State Services for Children with DeafBlindness - Family Leadership Training Series materials

*Links checked: March 2010*