



Fact Sheet

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 Deaf-Blindness: <http://www.nationaldb.org/ISLibrary.php>

Family Village Library: <http://www.familyvillage.wisc.edu/Specific.htm>

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -
<p><u>Aicardi Syndrome</u></p> <ul style="list-style-type: none"> • Absence of corpus callosum, either partial or complete (the corpus callosum allows the right side of brain to communicate with the left side). • Infantile spasms • Lesions or lacunae of the retina • Microcephaly (abnormally small head) • Porencephalic cysts (inside the brain tissue) • Only affects females except in males with Klinefelter Syndrome (XXY) <p>WEBSITE: http://www.aicardisyndrome.org/</p>
<p><u>Alport Syndrome</u></p> <ul style="list-style-type: none"> • X-linked disorder • Hereditary kidney damage • Nerve deafness • Congenital eye abnormalities • Ankle, feet, and leg swelling <p>WEBSITE: http://www.alportsyndrome.org/index.html</p>
<p><u>Alstrom Syndrome</u></p> <ul style="list-style-type: none"> • Photophobia (light sensitivity) in infancy • Nystagmus (wobbling of the eyes) • Congestive heart failure (CHF) • Childhood obesity • Blindness from progressive pigmentary retinopathy • Mild to moderate bilateral sensorineural hearing loss • Type II diabetes • Heart, liver, and renal failure • Pulmonary fibrosis • Progressive disease • Normal intelligence <p>WEBSITE: http://www.alstrom.org</p>
<p><u>Apert Syndrome</u></p> <ul style="list-style-type: none"> • Prematurely fused cranial structures

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -

- A rudimentary midface
- Fused fingers and toes
- Various heart defects
- Pulmonary atresia
- Tracheoesophageal Fistula
- Sleep apnea
- Ear infections
- Severe acne
- Increased incidence of eye injuries

WEBSITE: <http://www.apert-international.org/>

Bardet-Biedl Syndrome (BBS)

- Obesity
- Pigmentary retinopathy
- Polydactyly
- 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

- Cognitive Disability
- 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

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -

- 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
- Gastroesophageal reflux disease (GERD)
- Seizures
- Heart defects
- Cleft palate
- Cognitive Disability
- Missing limbs or portions of limbs

WEBSITE: <http://www.cdlsusa.org/>

Cri-du-Chat Syndrome

- High pitched cry at birth
- Low birth weight
- Poor muscle tone
- Microcephaly (abnormally small head)
- Potential medical complications

WEBSITE: <http://www.fivepminus.org/>

Crigler-Najjar Syndrome

- Very rare disorder
- Hyperbilirubinemia (must have daily 12 hour exposure to special blue lights)
- Jaundiced

WEBSITE: <http://www.criglernajjar.com/>

Crouzon syndrome

- Craniaosynostosis
- Hypertelorism
- Exophthalmos
- Strabismus
- Beaked nose
- Short upper lip
- Hypoplastic maxilla
- Upper airway obstruction develops secondary to septal deviation

WEBSITE: <http://www.crouzon.org/>

Cytomegalovirus (CMV)

- Most common congenital infection
- Low birth weight
- Microcephaly (abnormally small head)
- Seizures
- Rash – little red spots under the skin
- Enlarged liver and spleen (with jaundice)
- Abnormal muscle tone

WEBSITE: <http://www.bcm.edu/pedi/infect/cmv/index.htm>

Dandy-Walker Syndrome

- Slow motor development
- Progressive enlargement of the skull
- Convulsions
- Unsteadiness
- Lack of muscle coordination
- Jerky movements of the eyes

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -

WEBSITE: <http://www.dandy-walker.org/>

Down Syndrome (Trisomy 21)

- Smaller stature along with slower development physically and mentally
- Cognitive Disability
- Congenital heart disease
- Intestinal abnormalities
- Thyroid dysfunctions
- Skeletal problems
- Obesity in adolescence
- Small ear canals

WEBSITE: <http://www.ndss.org/>

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
- Limp
- 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)

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -

Herpes Zoster (Ramsay 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>

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
- Cognitive Disability

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>

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -

Leber's Congenital Amaurosis

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

- Cognitive Disability

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -

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

WEBSITE: <http://www.moebius syndrome.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
- Hypermobility 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
- Cognitive Disability
- 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
- Cognitive Disability
- 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>

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -

Prader-Willi Syndrome

- Profound poor muscle tone
- Underdeveloped sex organs
- Short stature
- Retarded bone age
- Cognitive Disability
- Rapid weight gain between ages 1 and 6 leading to obesity
- Obsession with food
- Distinctive facial features: narrow face, almond-shaped eyes, small-appearing mouth with thin upper lip and down-turned corners of mouth

WEBSITE: <http://www.pwsausa.org/>

Pierre Robin Sequence

- Lower jaw is abnormally small, but usually grows out as individual ages
- Tongue is displaced downwards
- Cleft Palate
- Many ear infections, leading to hearing impairment
- Often present with another genetic disorder
- Breathing and feeding issues

WEBSITE: <http://www.pierrerobin.org/> AND <http://www.faces-cranio.org/Disord/PierreRobin.htm>

Infantile Refsum Syndrome (Peroxisomal Biogenesis Disorder: Zellweger and Neonatal Adrenoleukodystrophy)

- Progressive loss of vision from retinitis pigmentosa
- Loss of smell
- Hearing loss from nerve damage
- Heart abnormalities
- Nerve disorder causing loss of sensation
- Ataxia (balance disorder)
- Ichthyosis (dry, scaly skin)
- Cognitive Disability

WEBSITE: <http://home.pacifier.com/~mstephe/>

Scheie Syndrome

- Corneal clouding
- Deafness
- Joint stiffness
- Coarse facial features
- Potential glaucoma
- Claw Hands
- Carpal tunnel syndrome
- Deformed feet

WEBSITE: http://www.mpssociety.org/content/4021/MPS_I/

Smith-Lemli-Opitz syndrome

- Psychomotor and growth retardation
- Cleft palate
- Hypospadias
- Microcephaly (abnormally small head)
- Ptosis
- Cognitive Disability

WEBSITE: <http://www.smithleliopitz.org/>

Stickler Syndrome

- Myopia, cataracts, glaucoma, detached retinas, astigmatism
- Stiff joints and over-flexible joints, arthritis
- Cleft palate
- Flat face with a small nose and little or no nasal bridge
- Middle or inner ear hearing loss
- Scoliosis

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -

- 30-40% also have Pierre Robin sequence

WEBSITE: <http://www.sticklers.org/sip2/>

Sturge-Weber syndrome

- Facial birthmark "Port Wine Stain," usually over the eye and forehead region
- Seizures, often starting by one year of age
- Weakening or loss of use of one side of the body (hemiparesis), usually on the opposite side of the port wine stain
- Cognitive Disability
- 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
- GERD
- Seizures
- Cognitive Disability
- 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
- Cognitive Disability
- 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

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -

- Thyroid problems
- Scoliosis
- Hearing disturbances from ear infections (otitis media)

WEBSITE: <http://www.turnersyndrome.org/>

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

Voqt-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

**SYNDROMES and DISEASES
- PRIMARY CHARACTERISTICS -**

- Coloboma of the eye
- Heart defects

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

Resources:

National Consortium on Deaf-Blindness (NCDB) – <http://www.nationaldb.org/ISLibrary.php>

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 Deaf-Blindness - Family Leadership Training Series materials

For more information contact:

Tanni Anthony

Phone: 303-866-6681

Email: anthony_t@cde.state.co.us

Gina Quintana

Phone: 303-866-6605

Email: quintana_g@cde.state.co.us

Colorado Department of Education
Exceptional Student Leadership Unit
1560 Broadway, Suite 1175
Denver, CO 80202

Fax: 303-866-6767

Web Page Address:

<http://www.cde.state.co.us/cdesped/Deafblind.asp>

Fact Sheets from the Colorado Services to Children and Youth with Combined Vision and Hearing Loss Project are to be used by both families and professionals serving individuals with vision and hearing loss. The information applies to children, birth through 21 years of age. The purpose of the Fact Sheet is to give general information on a specific topic. More specific information for an individual student can be provided through personalized technical assistance available from the project. For more information call (303) 866-6681 or (303) 866-6605. Updated: 07/10