COLORADO
Assistance for those
SERVICES TO
with both vision
CHILDREN WITH
and hearing loss
DEAFBLINDNESS

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

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -	
Aicardi Syndrome • 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)	Website About Aicardi Syndrome
Alport Syndrome • X-linked disorder • Hereditary kidney damage • Nerve deafness • Congenital eye abnormalities • Ankle, feet, and leg swelling	Website About Alport Syndrome
Alstrom Syndrome • 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 failure • Liver disease • Pulmonary fibrosis • Renal failure • Progressive disease • Normal intelligence	Website About Alstrom Syndrome

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -	
Apert Syndrome • Prematurely fused cranial structures • A reruded midface • Fused fingers and toes • Various heart defects • Pulmonary atresia • Tracheoesophageal Fistula • Sleep apnea • Ear infections • Severe acne • Increased incidence of eye injuries	Website About Apert Syndrome
Bardet-Biedl Syndrome (BBS) • Obesity • Pigmentary retinopathy • Plydactyly • Hypogonadism • Renal failure • Mental retardation	Website About Bardet-Biedl Syndrome (BBS)
Batten Disease • Mental retardation • Seizures • Progressive loss of sight • Progressive loss of motor skills • Fatal	Website About Batten Disease
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 About CHARGE Syndrome
Ring 18 Syndrome • Mental retardation • Microcephaly (abnormally small head) • Hypertelorism • Speech deficit • Deafness • Heart anomalies • Poor muscle tone	Website About Ring 18 Syndrome
Cockayne Syndrome • Dwarfism • Microcephaly (abnormally small head) • Progressive neurodevelopmental delay • Unsteady gait • Sunburns easily • Retinopathy and/or cataracts • Progressive hearing loss • Premature aging	Website About Cockayne Syndrome

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -	
Cogan's Syndrome • Inflammation of the eye • Hearing problems • Dizziness • Progressive disease	Website About Cogan's Syndrome
Cornelia de Lange Syndrome (CdLS) • Small stature • Microcephaly (abnormally small head) • Excessive body hair • Small hands and feet • GERD • Seizures • Heart defects • Cleft palate • Developmental delays • Missing limbs or portions of limbs	Website About Cornelia de Lange Syndrome (CdLS)
Cri-du-Chat Syndrome • High pitched cry at birth • Low birth weight • Poor muscle tone • Microcephaly (abnormally small head) • Potential medical complications	Website About Cri-du-Chat Syndrome
Crigler-Najjar Syndrome • Very rare disorder • Hyperbilirubinemia (must have daily 12 hour exposure to special blue lights) • Jaundiced	Website About Crigler-Najjar Syndrome
Crouzon Syndrome • Craniaosynostosis • Hypertelorism • Exophthalmos • Strabismus • Beaked nose • Short upper lip • Hypoplastic maxilla • Upper airway obstruction develops secondary to septal deviation	Website About Crouzon Syndrome
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 About Cytomegalovirus (CMV)
Dandy-Walker Syndrome • Slow motor development • Progressive enlargement of the skull • Convulsions • Unsteadiness • Lack of muscle coordination • Jerky movements of the eyes	Website About Dandy-Walker Syndrome

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -	
Down Syndrome (Trisomy 21) • Smaller stature along with slower development physically and mentally • Mental retardation • Congenital heart disease • Intestinal abnormalities • Thyroid dysfunctions • Skeletal problems • Obesity in adolescence • Small ear canals	Website About Down Syndrome (Trisomy 21)
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 About Encephalitis
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, smooth and/or long ridge 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 About Fetal Alcohol Syndrome
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/use • Small or misshapen ears, sometimes no outer ear structure • Skin tags or pits usually in front of 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 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 About Goldenhar Syndrome
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 About Hand-Schuller-Christian Disease (Histiocytosis)

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -	
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 About Herpes Zoster (Ramsey Hunt Syndrome)
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 About Hunter Syndrome (Mucopolysaccharidos Type II or MPS II)
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 About Hydrocephaly
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 About Kearns-Sayre Syndrome
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 1 About Klippel-Feil Sequence Website 2 About Klippel-Feil Sequence
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 About Kniest Dysplasia

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) • Developmental delay • Epilepsy • Motor skill impairment • Sensorineural hearing loss	Website About Leber's Congenital Amaurosis
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 About Leigh Disease
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 About Marfan Syndrome
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 About Marshall Syndrome
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 About Maroteaux Lamy Syndrome
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 About Meningitis

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -	
Chromosome 10, Monosomy 10p • Severe mental retardation • Growth delays • Malformations of the skull and facial region • Short neck • Congenital heart defects	Website About Chromosome 10, Monosomy 10p
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 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 about Moebius Syndrome
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 about Morquio Syndrome (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 about Neurofibromatosis
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 1 about Norrie Disease Website 2 about Norrie Disease
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 about Pfeiffer Syndrome

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -	
Prader-Willi Syndrome • Profound poor muscle tone • Underdeveloped sex organs • Short stature • Retarded bone age • Developmental delays • 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 about Prader-Willi Syndrome
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 1 about Pierre Robin Sequence Website 2 about Pierre Robin Sequence
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) • Severe mental retardation	Website about Infantile Refsum Syndrome
Scheie Syndrome • Corneal clouding • Deafness • Joint stiffness • Coarse facial features • Potential glaucoma • Claw Hands • Carpal tunnel syndrome • Deformed feet	Website about Scheie Syndrome
Smith-Lemli-Opitz Syndrome • Psychomotor and growth retardation • Cleft palate • Hypospadias • Microcephaly (abnormally small head) • Ptosis • Mental retardation	Website about Smith-Lemli-Opitz Syndrome

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -	
Stickler Syndrome • Myopia, cataracts, glaucoma, detached retinas, astigmitism • 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 • 30-40% also have Pierre Robin sequence	Website about Stickler Syndrome
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 • Developmental delay • Glaucoma • Growth hormone deficiency • Severe headaches	Website about Sturge-Weber Syndrome
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 about Treacher Collins Syndrome
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 about Patau Syndrome (Trisomy 13)
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 about Edward Syndrome (Trisomy 18)

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -	
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 about Turner Syndrome
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 about Usher Syndrome (Type I)
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 about Usher Syndrome (Type II)
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 about Usher Syndrome (Type III)
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 about Vogt-Koyanagi-Harada Syndrome
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 about Waardenburg Syndrome

SYNDROMES and DISEASES - PRIMARY CHARACTERISTICS -	
Wildervanck Syndrome • Primarily affects females • Hearing impairment • Nystagmus • Fusion of two or more bones in the spinal column within the neck	Website about Wilderyanck 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 about Wolf-Hirschhorn Syndrome

Resources:

National Consortium on Deaf-Blindness (NCDB)
Sense
Texas School for the Blind and Visually Impaired (TSBVI)
Washington State Services for Children with Deaf-Blindness - Family Leadership Training Series materials

For more information about the CO Services for Children and Youth with Combined Vision and Hearing Loss Project:

Colorado Department of Education
Phone Number: 303-866-6694 – Ask to speak with a Deafblind Specialist on staff with ESSU
Fax: 303-866-6918
CDE Deafblind Webpage

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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. The contents of this Fact Sheet were developed under a grant from the United States Department of Education (US DOE), #H326C080044. However, these contents do not necessarily represent the policy of the US DOE and you should not assume endorsement by the Federal Government. 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. Reviewed: 3/17