

Documented Etiologies of Deafness

Cardiac

CHARGE (C and/or SN)
Jervell and Lange-Nielsen syndrome (AR) (SN)
Kartagener Syndrome (C) (AR)
Kawasaki disease (SN)
Klippel-Feil sequence (C and/or SN)
Mitral insufficiency, joint fusion, and hearing loss (AD)
Noonan syndrome (SN)
Trisomy 13 syndrome (SN)
Turner syndrome (SN)

Degenerative

Friedreich ataxia
Huntington disease
Myoclonic epilepsy

External ear malformations

Ear malformation and conductive hearing loss (AD)
Malformed low-set ears and conductive hearing loss (AR)
Otofaciocervical abnormalities (AD)
Preauricular pits and neural hearing loss (AD)
Thickened ears and incudostapedial abnormality (AD)

Hereditary with no associated anomalies

Congenital moderate hearing loss (AR)
Congenital neural deafness (SL)
Congenital severe deafness (AD)
Congenital severe deafness (AR)
Early-onset neural deafness (AR)
Early-onset neural deafness (SL)
Low-frequency hearing loss (AD)
Midfrequency hearing loss (AD)
Moderate hearing loss (SL)
Otosclerosis (AD)
Progressive nerve deafness (AD)
Unilateral nerve deafness (AD)

Infectious

Bacterial
Acute meningitis (SN)
Otitis media (SN and/or C)
Pertussis (SN)
Streptococcal (scarlet fever) (SN)
Tuberculosis (SN)
Typhoid (SN)
Prenatal
Cytomegalovirus (SN)

Herpes simplex (SN)
Rubella (SN and/or C)
Syphilis (SN)
Toxoplasmosis (SN)
Viral
Adenovirus 3 (SN)
Herpes zoster (SN)
Infectious mononucleosis (SN)
Mumps (SN)
Rubella (SN)
Rubeola (SN)
Varicella (SN)
Viral encephalitis (SN)

Integumentary system disease

Albinism and congenital deafness (AD)
Anhidrosis and progressive hearing loss (AD)
Atopic dermatitis and neural hearing loss (AR)
Hereditary piebaldness and congenital deafness (AR?)
Keratopachydermia, digital constrictions, and deafness (AD)
Knuckle pads, leukonychia, and hearing loss (AD)
Leopard syndrome (lentiginos and congenital deafness) (SN) (AD)
Onchodystrophy and congenital deafness (AD)
Onychodystrophy and deafness (AR)
Onychoclystrophy, coniform teeth, and hearing loss (AD)
Onchodystrophy, digital abnormalities, and deafness (AR)
Pigmentary abnormalities and congenital deafness (SL)
Pili torti and hearing loss (AR)
Pili torti and hearing loss (AR?)
Senter syndrome (SN)
Vitiligo, congenital deafness, muscle wasting, and achalasia (AR)
Waardenburg syndrome (SN) (AD)

Metabolic and endocrinologic diseases

Alport syndrome (progressive renal impairment and deafness)
Diabetes mellitus, optic atrophy, and perceptual hearing loss
Goiter, increased protein-bound iodine, stippled epiphyses, and deafness (AR)
Hunter syndrome (SN and/or C) (SL)
Hurler syndrome (SN and/or C) (AR)
Hypoglycemia
Hypoproteinemia
Hypothyroidism (SN)
Kernicterus (SN)
Maroteaux-Lamy mucopolysaccharidosis (SN) (AR)
Morquio syndrome (SN) (AR)
Pendred disease (goiter and deafness) (AR)
Pyridoxine dependency
Refsum disease (AR)
Tay-Sachs disease (AR)
Thiamine dependency
Wilson disease (AR)

Neoplastic

Leukemia

Neurofibromatosis

Neurologic

Acoustic neuromas (AD)

Photomyoclonus, hearing loss, diabetes₁ and nephropathy (AD)

Richards-Rundel disease (deafness, mental deficiency, ataxia, and hypogonadism) (AR)

Sensory radicular neuropathy (AD)

Other malformation syndromes

Camurati-Engelmann syndrome (SN) (AD)

Cleidocranial dysostosis syndrome (SN) (AD)

Diastrophic dysplasia syndrome (C) (AR)

Ectodermal dysplasia (SL)

EEC syndrome (AD)

18q syndrome (C)

Facioauriculovertebral spectrum (Goldenhar syndrome) (C)

Fanconi pancytopenia (SN) (AR)

Frontometaphyseal I dysplasia syndrome (C and/or SN) (SL)

Frontonasal dysplasia sequence

Hag-Wells syndrome

Johanson-Blizzard syndrome (SN) (AR)

Killian/Teschler-Nicola syndrome (SN)

Langer-Giedion syndrome (SN)

Laurence-Moon-Biedl syndrome (AR)

Levy-Hollister syndrome (C and/or SN) (AD)

Melnick-Fraser syndrome (AD)

MURCS association (SN)

Nager syndrome (C)

Oculodentodigital syndrome (AD)

Progeria (SN)

Saethre-Chotzen syndrome (SN) (AD)

Scheje syndrome (SN) (AR)

Shprintzen syndrome (velocardiofacial syndrome)(C) (AD)

Townes (SN) (AD)

Trisomy 8S

Prenatal toxic syndromes

Fetal alcohol syndrome (SN)

Fetal iodine deficiency effect (SN)

Fetal methyl mercury effects (SN)

Fetal trimethadione effects

Renal

Alport disease (nephritis and hearing loss) (AD)

Branchio-otorenal (BOR) syndrome (C and/or SN) (AD)

Renal, genital, and middle ear anomalies

Urticaria amyloidosis, nephritis, and hearing loss (AD)

Skeletal disease

Absence of tibia and deafness (AR)
Albers-Schonberg disease (osteopetrosis) (SN) (AR)
Cranialmetaphyseal dysplasia (SN) (AD and AR)
Crouzon syndrome (SN) (AD)
Engelmann disease (progressive diaphyseal dysplasia) (AR)
Multiple synostoses syndrome (C) (AD)
Oralfaciodigital (OFO) syndrome II (Mohr syndrome) (C) (AR)
Otopalatodigital syndrome (C) (X-linked, semidominant)
Paget disease of bone (osteitis deformans) (AD)
Proximal symphalangism and hearing loss (AD)
Pyles disease (craniometaphyseal dysplasia) (AD)
Sclerosteosis (SN) (AR)
Split hand and foot syndrome (AR)
Treacher-Collins syndrome (C) (AD)
Van Buchen disease (hyperostosis corticis glomerata) (AR)

Trauma

Acoustic trauma
Hypoxia
Skull fracture

Visual system

Alstrom disease (SN) (AR)
Cockayne syndrome (SN) (AR)
Familial hearing loss, polyneuropathy, optic atrophy (AR and SL)
Marshall syndrome (SN) (AD)
Myopia and hearing loss (AR)
Myopia, hearing loss, peripheral neuropathy, and skeletal abnormalities (AD)
Optic atrophy, hearing loss, and juvenile diabetes (AR)
Retinal changes, deafness, muscular wasting, and mental retardation (AR)
Saddle nose, myopia, cataracts, and hearing loss (AD)
Stickler syndrome (C and/or SN) (AD)
Usher syndrome (SN) (AR)

Key AD-Autosomal Dominant
AR-Autosomal Recessive
SL-Sex Linked
C -Conductive
SN-Sensorineural