

Hearing & Amplification

Causes of Hearing Loss

Basic Genetics

What Are the Most Common Forms of Genetic Hearing Loss?

Of the 50% of the genetic forms of hearing loss, an estimated 70% are due to recessive causes, about 15% have a dominant cause; and the remaining 15% include all the other forms of inheritance.

GENETIC	
RECESSIVE 70%	NON-GENETIC AND UNKNOWN
DOMINANT 15%	
OTHER GENETIC 15%	

Genetic scientists subdivide genetic hearing loss into two general categories: "Non-Syndromic" (meaning hearing loss and nothing else) and "Syndromic" (meaning hearing loss with other clinical findings). By far, the more common is Non-syndromic hearing loss which includes 2/3 of all genetic hearing losses.

SYNDROMIC	NON-SYNDROMIC	
RECESSIVE	RECESSIVE	NON-GENETIC AND UNKNOWN
DOMINANT	DOMINANT	
OTHER GENETIC	OTHER GENETIC	

(continued on page 2)

What is the Most Common form of genetic hearing loss?

One gene, known as Connexin 26 (abbreviated CX26), is estimated to be responsible for half of all the recessive cases of hearing loss. There are over 400 known genetic causes involving hearing loss. CX26 alone is responsible for about 1/3 of all the cases of genetic hearing loss!

SYNDROMIC	NON-SYNDROMIC	
	CONNEXIN-26	NON-GENETIC AND UNKNOWN
RECESSIVE	RECESSIVE	
DOMINANT	DOMINANT	
OTHER GENETIC 15%	OTHER GENETIC 15%	

What are the Next Most Common Forms of Genetic Hearing Loss?

Since CX26 accounts for about 1/3 of all cases of genetic hearing loss, that leaves about 1/3 of all cases as non-syndromic (this includes all types of inheritance) with the remaining 1/3 as syndromic. Among the remaining 1/3 of non-syndromic cases of genetic hearing loss, 13 dominant and 8 other recessive genes have been described.

What are the Common Dominant Syndromic Hearing Loss Types?

The following descriptions are only a brief review. Medical professionals can guide you for further information. About 5% of all hearing loss is dominant syndromic in nature. <http://www.familyvillage.wisc.edu/index.htmlx>

Waardenburg Syndrome - The hearing loss in Waardenburg Syndrome may be present in one or both ears and is a sensorineural type of loss. The striking features of Waardenburg syndrome may include: premature graying hair, white forelock, fused eyebrows (synophrys), two different-colored eyes (heterochromia irides, usually bright blue and brown), widely-spaced eyes (hypertelorism), high nasal bridge, under-developed nose tip (hypoplastic alae nasiae) and partial albinism.

<http://www.boystownhospital.org/parents/info/genetics/waardenburg.asp>
<http://www.nidcd.nih.gov/health/hearing/waard.asp>

Branchio-Oto-Renal (BOR) Syndrome - The hearing loss in BOR Syndrome is conductive, sensorineural or mixed. Cysts (or pits) can be found on the neck (branchial cleft) or in front of the outer ear (preauricular). The outer ear (pinna) may be malformed and stapes fixation, inner ear malformations and/or enlarged vestibular aqueducts may be present. A major medical concern with BOR are the associated renal (kidney) problems, which could be life threatening.

<http://www.boystownhospital.org/parents/info/genetics/bor.asp>

Neurofibromatosis Type II (NFII) - The hearing loss in NFII is progressive sensorineural leading to possible deafness. Café-au-lait (coffee with cream-colored) spots may appear on the skin, with freckling and cataracts. Acoustic tumors may grow on the VIIIth cranial nerve, causing hearing loss. Tumors may also grow on other nerves.

<http://www.nfinc.org/>
<http://www.nf.org>
<http://www.nlm.nih.gov/medlineplus/neurofibromatosis.html#nlmnhresources>

(continued on page 3)

Stickler Syndrome - The hearing loss in Stickler Syndrome is usually conductive, although some losses may be mixed or sensorineural. Progressive hearing loss has also been reported. There are three syndrome types. All are related to altered expression of a collagen/connective tissue gene. Associated features may include: cleft palate, downward-placed tongue (glossoptosis), small jaw (micrognathia), under-developed midface, progressive severe near-sightedness (myopia), cataracts, retinal detachment/degeneration, bone/joint disorders, early adult-onset arthritis, and middle ear bone (ossicular) malformations.

<http://www.sticklers.org/sip/>

Treacher-Collins Syndrome - The hearing loss in Treacher-Collins is conductive. Striking facial features include cleft palate, down-slanting eye-slit openings (palpebral fissures), unusual pupil openings (coloboma), under-developed cheek bones (malar hypoplasia), absent/malformed outer ears, absent (atresia) or narrow (stenosis) ear canals, skin tags in front of the ear (preauricular), teeth alignment problems (malocclusion) and possible balance (vestibular) problems.

<http://www.treachercollinsfnd.org/>

What are the Common Recessive Syndromic Hearing Loss Types?

Recessive Syndromic hearing loss accounts for about 20% of all types of genetic hearing loss.

Usher Syndrome - There are three (3) types of Usher syndrome with different types of hearing loss. Type I has congenital, profound, sensorineural hearing loss. Type II has a downward-sloping, sensorineural hearing loss. Type III has a progressive sensorineural hearing loss. All types have balance (vestibular) problems and progressive vision loss due to retinal degeneration (retinitis pigmentosa).

<http://www.nidcd.nih.gov/health/hearing/usher.asp>

Alport Syndrome - Hearing loss in Alport syndrome may be sensorineural, conductive or mixed and may be progressive. Alport syndrome may also be X-linked (linked to sex-chromosome inheritance). Other features include kidney problems (nephritis), near-sightedness (myopia) or cataracts, and palate abnormalities.

<http://www.cc.utah.edu/~cla6202/ASHP.htm>

Jervell and Lange-Nielson Syndrome - The hearing loss in JL-N syndrome is sensorineural. The other major finding in JL-N is an abnormal heart rhythm (long Q-T), which could lead to fainting spells and possible sudden death. These might be mistaken for seizures. These abnormal heart rhythms are successfully treated with medication (beta blockers).

Pendred Syndrome - The hearing loss in Pendred syndrome may be mixed or sensorineural and may be progressive. Pendred Syndrome may have an associated goiter, and variable inner ear malformations (Mondini malformation with or without enlarged vestibular aqueduct).

<http://www.medicinenet.com/Script/Main/Art.asp?li=MNI&ArticleKey=9460>

What are Some Less-Common Syndromic Hearing Loss Types?

CHARGE Syndrome - The letters in CHARGE stand for Coloboma, Heart, Atresia of the choanae, Retardation of growth and development, Genital and urinary abnormalities, and Ear abnormalities. CHARGE is thought to be multifactorial. The hearing loss may be conductive, sensorineural or mixed and range from mild to profound. In addition to the features in the name, there may be partial facial paralysis, cleft palate, cleft lip, kidney problems, and feeding problems due to an opening between the windpipe and the feeding tube.

<http://www.chargesyndrome.org/>

(continued on page 4)

X-Linked Congenital Stapes Fixation with Perilymph Gusher - For boys with this syndrome, the hearing loss is mixed and may be progressive. Females who carry this gene may have mild, mixed or sensorineural hearing loss. Boys with this gene have a further risk of increased hearing loss if middle ear surgery is performed to correct the stapes fixation because the surgery may result in massive and sudden loss of inner ear fluid (perilymph).

Mitochondrial Conditions

Mitochondria are structures in the cell that produce the energy that cells need to survive. Neither the mitochondria nor the cell can exist without the other. Interestingly, mitochondria have a separate set of genes that are not part of the cell's genes. This is because mitochondria originally came from energy-producing bacteria that merged with the cell. Anything that affects bacteria could also affect the function of the cell's mitochondria, which might eventually affect the cell's energy source. Changes in the mitochondrial genes can also result in syndromes involving hearing loss.

Mitochondrial Encephalopathy, Lactic Acidosis and Stroke-like Episodes (MELAS) - About 30% have sensorineural hearing loss. The findings are highly variable and may include: intermittent vomiting, limb weakness, stroke-like episodes, partial paralysis, partial blindness, seizures, migraine-like headaches, diabetes, short stature, heart problems and kidney problems.

Maternally Inherited Diabetes and Deafness (MIDD) - The hearing loss is sensorineural. The only other finding is diabetes.

Kearns-Sayre Syndrome (KSS) - The hearing loss is sensorineural. Other findings include: unsteady gait (ataxia), short stature, delayed puberty, progressive paralysis of the eye muscles (ophthalmoplegia) and progressive blindness (retinopathy).

Myoclonic Epilepsy and Ragged Red Fibers (MERRF) - The hearing loss is sensorineural. Other findings include unsteady gait (ataxia), epilepsy, and possible blindness (optic atrophy).

1555DELG - The hearing loss is sensorineural and may be progressive. Those who have this condition may have sudden hearing loss when exposed to aminoglycoside antibiotics (e.g., neomycin, gentamycin, streptomycin, kanamycin, tobramycin, or amikacin).