



Risk Factors for Late Onset Hearing Loss: Features Associated with Syndromes

Syndromes known to include sensorineural and/or conductive hearing loss include:

CHROMOSOME ABNORMALITIES

● *Down Syndrome*

Mental retardation, an upward slant to the eyes, flattened facial profile, decreased muscle tone, small nose, small mouth, protruding tongue, and abnormally shaped ears, and in some cases congenital hearing loss and heart defects.

Autosomal Dominant Syndromes

Waardenburg Syndrome

- Congenital sensorineural hearing loss, hair hypopigmentation (usually white forelock, eyebrows, and/or eye lashes), hypochromic iridis, and lateral displacement of inner canthi.

● *Branchio-Oto-Renal (BOR) Syndrome (Melnick-Fraser Syndrome)*

Diagnosis without family history based on the presence of any three of the following: mild to profound conductive, sensorineural, or mixed hearing loss (due to abnormalities of the pinnae, external auditory canal, middle ear, or inner ear), pre-auricular pits or tags, "lop-ear" deformity, branchial fistulae or cysts, and renal dysplasia.

● *CHARGE Syndrome*

Coloboma of the eye, Heart anomaly, choanal Atresia, mental Retardation (ranging from mild to profound), microphallus (Genital and urinary difficulties), and Ear anomalies and/or defects.

● *Stickler Syndrome*

High myopia, hypotonia, prominent eyes, variable hearing impairment, clefts of hard and/or soft palate, flat faces with depressed nasal bridge, mitral valve prolapse, and hyperextensible joints.

● *Neurofibromatosis Type II*

Bilateral acoustic neuromas developing usually in the second or third decade, schwannoma, spinal tumors, meningiomas, juvenile posterior subcapsular cataracts, focal weakness, tinnitus, balance dysfunction, and seizure.

● *Treacher Collins Syndrome*

Midface hypoplasia, micrognathia, mandibular hypoplasia, external auditory canal defects, conductive hearing loss, coloboma of the lower eyelid, partial to total absence of lower eyelashes, and preauricular hair displacement.

Autosomal Recessive Syndromes

Usher Syndrome

- Congenital bilateral mild to severe hearing loss or progressive sensorineural hearing loss, vestibular areflexia, and retinitis pigmentosa.

● *Pendred Syndrome*

Hearing loss that is usually congenital (sometimes late onset and progressive), vestibular dysfunction, and temporal bone anomalies, goiter.

● *Jervell & Lange-Nielsen Syndrome (Long QT Syndrome)*

Profound congenital sensorineural deafness, prolonged QTc interval, often presents as a deaf child with syncopal episodes during stress or exercise.

X-Linked Syndromes

Alport Syndrome

- Microhematuria, ocular lesions, anterior lenticonus, maculopathy, hearing loss is never congenital but can be detected in late childhood or early adulthood.

For more information or a list of resources, please visit our website: www.doh.wa.gov/EarlyHearingLoss/Provider

Contact us at:

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