Pediatric Clinical Support: Usher Syndrome

Usher Syndrome is an autosomal recessive genetic disorder that is responsible for the majority of deaf-blindness, occurring in 1 out of 23,000 people in the United States. Males and females are equally likely to inherit this disorder. Prevalence of this condition is commonly reported at 3% to 6% (Boughman, Vernon, & Shaver, 1983).

Characterized by: deafness (varying degrees dependent on Type of Usher Syndrome); gradual vision loss (Retinitis Pigmentosa, RP*)

3 Types of Usher Syndrome:
Types I and II are the most common types of Usher Syndrome, accounting for approximately 90 to 95% of all cases.

Type I	Type II	Type III
Individual is born profoundly deaf	Born with SNHL but are not deaf; hearing loss can be progressive	Not born with hearing loss or vision loss
Vision loss appears in the 1st decade of life	Vision loss begins at 2nd decade of life; some vision is preserved into middle age	Hearing and vision loss is gradual over lifespan
Problem with the vestibular system are apparent; delays in learning how to walk due to balance difficulties	Questionable whether balance difficulties are present	May or may not experience balance problems
Gene mutation in one of **six** identified genes	Gene mutation in one of **three** identified genes	Gene mutation in only **one** identified gene

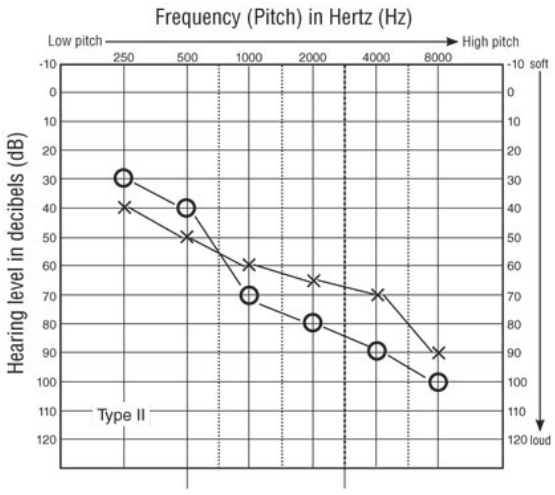

Figure 1. Typical hearing loss configuration for Type II Usher Syndrome.
(www.asha.org/aud/articles/UsherSyndrome.htm)

Audiological Treatment & Rehabilitation:

Hearing evaluations to determine severity, which can determine appropriate amplification or cochlear implantation for profound losses. Assistive listening devices can be used in addition to or in conjunction with amplification and/or cochlear implants. American Sign Language can be used as a communication method. Orientation and mobility training will aid in balance issues.

There is no cure for RP, but Braille instruction and low-vision services should be made available. As vision and hearing status decline, tactile signing and communication devices (e.g., Tellatouch, TeleBraille) may be of benefit.

*Retinitis Pigmentosa: gradual vision loss that is caused by degeneration of retinal cells in the eyes. Starts with degeneration of rod cells, which can affect night vision and leads to early onset of night blindness. Will eventually affect the peripheral vision system, leaving only central ("tunnel") vision.

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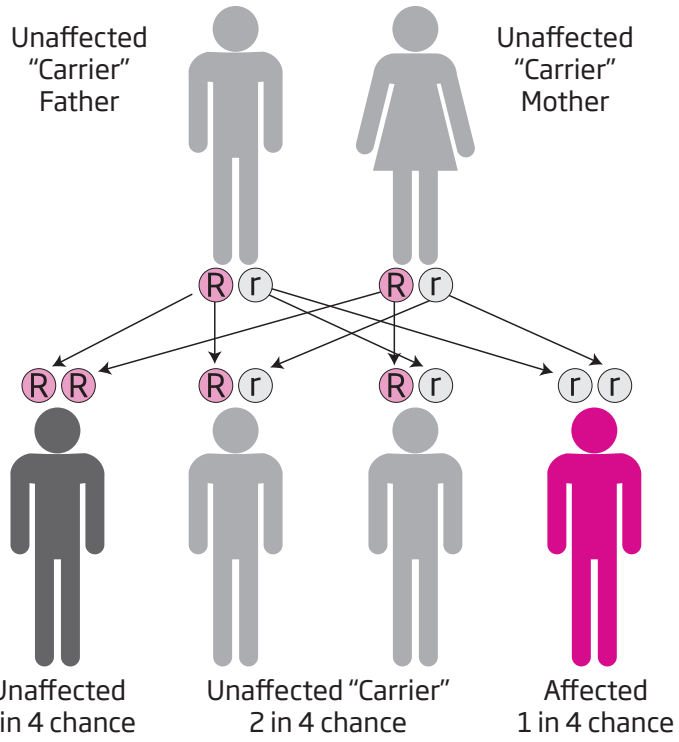
Educational Considerations: The presence of Usher Syndrome should be indicated and addressed in the child's IEP. This includes allowing access to information, including strategies to systematically develop the use of sensory information with remaining functional vision and hearing, and effective strategies and approaches that teach the use of environmental cues that the child may be missing. Integration in social environments could include methods to orient the child to other students and locations, and improve the number and quality of interactions and relationships that the child has with others.

Professional Considerations: Due to the progression of vision loss, quality of life considerations should be addressed, including the implications of dual sensory loss, with all professionals who interact with the child. This includes, but may not be limited to, the audiologist, speech-language pathologist, occupational therapist, low vision or mobility instructor, and child social worker or psychologist.

Gene Expression



Source: www.nidcd.nih.gov/health/hearing/pages/usher.aspx

Prognosis: Incurable, although gene therapies in mice have shown promise in reversing one type (1B) of the disorder. Research has suggested that high doses of vitamin A palmitate may slow the progression of vision loss, but will not cease the progression of vision loss.

Differential Diagnosis: Completed by testing for chromosomal mutations and using electroretinography (ERG), although this is not recommended in children as it can cause unreliable results if the child moves due to discomfort during testing. Type I may be considered with signs of profound deafness from birth and slow advancement in walking. Another factor in diagnosis is parental consanguinity. Other syndromes with similar characteristics to Usher syndrome: Alport syndrome, Alstrom syndrome, Bardet-Biedl syndrome, Cockayne syndrome, spondyloepiphyseal dysplasia congenita, Flynn-Aird syndrome, Friedreich ataxia, Hurler syndrome, Kearns-Sayre syndrome, Norrie syndrome, osteopetrosis, Refsum's disease, and Zellweger syndrome.

Online Support Sources:
http://www.deafblind.com/nidcd.html
http://www.kumc.edu/gec/support/usher.html
http://resources.pepnet.org/files/414_2011_1_26_10_10_AM.pdf
http://nationaldb.org/
http://deafblindnetworks.blogspot.com/
http://www.deafblind.co.uk/

Online and other References:
http://www.asha.org/aud/articles/UsherSyndrome.htm
http://www.blindness.org/index.php?option=com_content&id=50&Itemid=67 http://www.deafblind.com/whatushe.html
http://www.ushersyndrome.nih.gov/
http://www.nidcd.nih.gov/health/hearing/pages/usher.aspx

Berson EL, Rosner B, Sandberg MA, et al. (1993). A randomized trial of vitamin A and vitamin E supplementation for retinitis pigmentosa. Archives of Ophthalmology, 111(6): 761-72.

Boughman, J., Vernon, M., & Shaver, K. (1983). Usher syndrome: Definition and estimate of prevalence from two high-risk populations. Journal of Chronic Diseases, 36, 595-603.

Hashimoto T, Gibbs D, Lillo C, Azarian SM, Legacki E, Zhang XM, Yang XJ, Williams DS (2007). "Lentiviral gene replacement therapy of retinas in a mouse model for Usher syndrome type 1B". Gene Therapy, 14(7): 584-594.

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