

Usher Syndrome

Clinical Characteristics

There are three types of Usher syndrome and the clinical characteristics vary depending upon the type. In general, Usher syndrome is characterized by hearing loss and vision loss due to retinitis pigmentosum (RP). RP is an eye disorder that results in progressive and bilateral degeneration of the rods and cones of the retina. The onset of RP usually begins with night blindness, but can steadily worsen until an individual is completely blind. Those with Usher syndrome type I (the “classic” form) tend to have adolescent onset RP; those with type II have adolescent-to-adult onset; those with type III have late onset. Type IV is extremely rare.

Usher Syndrome and Hearing Loss

Again, the type and degree of hearing loss depends on the type of Usher syndrome. Individuals with type I tend to have congenital, bilateral, profound sensorineural hearing loss, as well as vestibular dysfunction. Those diagnosed with type II are more likely to have mild to severe loss and normal vestibular function. Those with type III usually have postlingual, progressive hearing loss and variable vestibular function.

Natural History

Usher syndrome affects both males and females, as well as all ethnicities. The prevalence is estimated to be 4-5 per 100,000, with a carrier frequency as high as 1 in 70. Usher syndrome type I is much more common than types II or III. Usher syndrome is the most common condition to involve vision and hearing, and affects about 3-6% of all deaf children.

Type I: Hearing loss is present at birth. Due to vestibular dysfunction, walking is delayed (typically begins between 18 and 24 months). “Clumsiness” or lack of coordination persists through childhood and adolescence. It is rare that affected persons ever develop speech. Individuals are often misdiagnosed with nonsyndromic deafness until RP begins to manifest, usually in adolescence. The rate and degree of vision loss is variable, even among affected family members.

Type II: Hearing loss is present at birth. Since vestibular function is normal, children walk at the typical age. Most individuals develop speech, but some do not. RP typically develops in late adolescence or early adulthood and can be variable among family members. The progression of RP is typically slower in type II than in type I.

Type III: Hearing is normal at birth. Hearing loss typically begins in adolescence and progresses to complete deafness in mid- to late-adulthood. Vision loss usually begins around the same time as hearing loss and the individual is typically blind by mid-adulthood. The rate and degree of both hearing and vision loss is variable, even among affected family members.

For all types of Usher syndrome, the overall prognosis is good, as is general health. Intelligence is usually normal, though some educational intervention may be necessary due to issues with hearing and vision loss. Life expectancy is normal.

Genetics

Eight genes on six chromosomes have been identified as being associated with the three types of Usher syndrome: *MYO7A*, *USH1C*, *CDH23*, *PCDH15*, *USH1G*, *USH2A*, *VLGR1*, and *USH3A*. Genetic testing for Usher syndrome is available on a research basis. Usher syndrome is **autosomal recessive**. This means that both parents of an affected individual must at least be carriers for the condition. Whenever two asymptomatic carriers for Usher syndrome have a child, there is a 1 in 4 (25%) chance the child will be affected, a 2 in 4 (50%) chance the child will be an unaffected carrier just like his or her parents, and a 1 in 4 (25%) chance the child will be neither affected nor a carrier. Overall, there is a 75% chance with each pregnancy the child will **not** have Usher syndrome. If one parent has Usher syndrome, the chance of having an affected child depends upon the carrier status of the other parent. If both parents have Usher syndrome, every single child will also have the syndrome.

Management

The diagnosis of Usher syndrome is made clinically. An initial evaluation for an individual suspected of having Usher syndrome should include assessments by audiology and ophthalmology. The audiologic evaluation should include otoscopy, puretone audiometry, speech perception assessment, and possibly ABR and DPOAE. Vestibular function should be tested as well. Hearing aids will generally be ineffectual for those with type I, though may be of some benefit to those with type II. Cochlear implants may be considered. Routine audiologic evaluation is recommended. Any interventions for hearing loss should be designed keeping in mind that vision impairment and loss will eventually occur.

Routine ophthalmologic evaluation is recommended. As vision worsens, appropriate accommodations will need to be made (e.g. adapting communication styles, discontinue driving, etc.).

A scheduled appointment with a geneticist and/or genetic counselor is recommended.

An individual with type I has impaired balance and should therefore be cautious about entering competitive sports. Disorientation in water has been known to be particularly troublesome. With proper supervision, though, athletic activity could help to improve a person's balance and coordination.

Supplementation with vitamin A has been postulated as a means to slow the progression of RP in individuals with Usher syndrome. The effectiveness of this therapy has not been thoroughly investigated. High doses of vitamin A should be administered only under the supervision of a physician who can monitor for harmful side effects.

Resources for Families

Statewide Genetics Program

Phone: 608-267-7148

Fax: 608-267-3824

Email: meyeram@dhfs.state.wi.us

Wisconsin First Step Hotline

Phone: 1-800-642-7837 voice/TTY

Website: www.mch-hotlines.org

Wisconsin Bureau for Deaf and Hard of Hearing

Phone: 1-608-266-3118 voice/TTY

Website: www.dhfs.state.wi.us/sensory

Regional Children and Youth with Special Health Care Needs Centers

Centers in Green Bay, Wausau, Milwaukee, Madison, and Chippewa Falls

Website: http://dfhs.wisconsin.gov/DPH_BFCH/cshcn/index.HTM

Parent-to-Parent of Wisconsin

Phone: 1-888-266-0028

Email: mathea@shsmh.org

Family Village online resource

Library Card Catalog of Disorders

www.familyvillage.wisc.edu

Usher Family Support

4918 42nd Avenue, S

Minneapolis, MN 55417

Phone: 612-724-6982

Email: kadbmn@aol.com

National Organization for Rare Disorders (NORD)

www.rarediseases.org
