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Fact Sheet

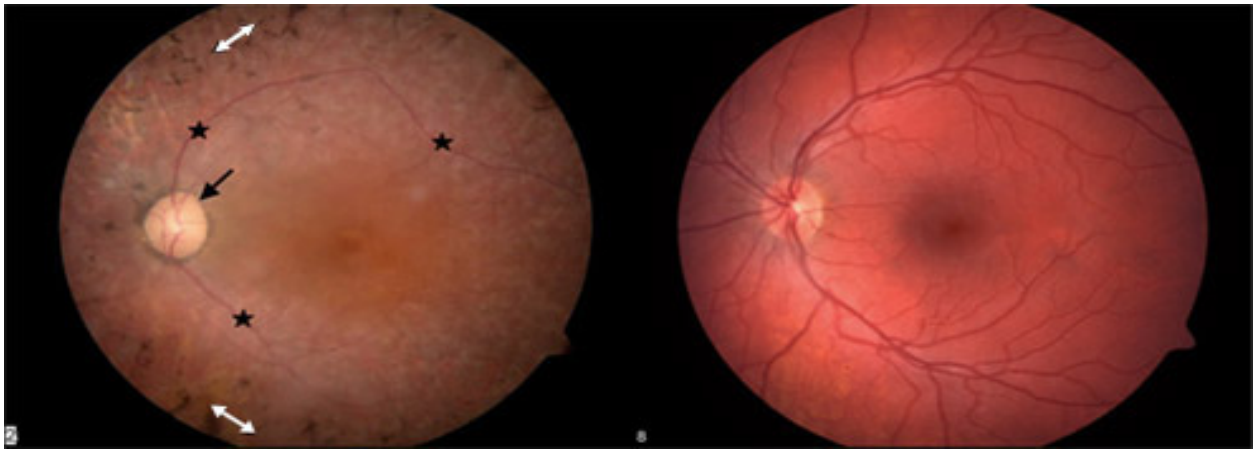
Usher Syndrome

The primary content of this article was retrieved from:
http://www.nidcd.nih.gov/health/hearing/pages/usher.aspx

What is Usher syndrome? Usher syndrome is a leading cause of deafblindness. Approximately 1 in 17,000 or 0.01% or 16,000 people in United States have been diagnosed with Usher syndrome. It is the most common condition that affects both hearing and vision.

A ***syndrome*** is a disease or disorder that has more than one feature or symptom. The major symptoms of Usher syndrome are hearing loss and an eye disorder called retinitis pigmentosa (RP). Retinitis pigmentosa causes night-blindness and a loss of peripheral vision (side vision) through the progressive degeneration of the retina. In other cases, there is early degeneration of the cone cells in the macula, leading to a loss of central acuity. In some cases, the foveal vision is spared, leading to "doughnut vision"; central and peripheral vision are intact, but there is an annulus around the central region in which vision is impaired.

The retina is a light-sensitive tissue in the back of the eye and is crucial for vision. As RP progresses, the field of vision narrows—a condition known as “tunnel vision”—until only central vision (the ability to see straight ahead) remains. Many people with Usher syndrome also have severe balance problems.



Photograph of the retina of a patient with Usher syndrome (left) compared to a normal retina (right). The optic nerve (arrow) looks very pale, the vessels (stars) are very thin and there is characteristic pigment, called bone spicules (double arrows).

Usher syndrome is named after the British ophthalmologist Charles Usher, who examined the pathology and transmission of this illness in 1914 on the basis of 69 cases. However, it was first described in 1858 by Albrecht von Gräfe, a pioneer of modern ophthalmology. He reported the case of a deaf patient with retinitis pigmentosa, who had two brothers with the same symptoms. Three years later, one of his students, Richard Liebreich, examined the population of Berlin for disease pattern of deafness with retinitis pigmentosa. Liebreich noted that Usher syndrome is recessive, since the cases of blind-deafness combinations occurred particularly in the siblings of blood-related marriages or in families with patients in different generations. His observations supplied the first proofs for the coupled transmission of blindness and deafness, since no isolated cases of either could be found in the family tree.

What are the Characteristics of the Three Types of Usher Syndrome? There are three clinical types of Usher syndrome: type 1, type 2, and type 3. In the United States, types 1 and 2 are the most common types. Together they account for approximately 90 to 95 percent of all cases of children who have Usher syndrome.

***Type 1*:** Children with type 1 Usher syndrome are profoundly deaf at birth and have severe balance problems. Many of these children obtain little or no benefit from hearing aids. Intervention should be introduced early, during the first few years of life, so that the child can take advantage of the unique window of time during which the brain is most receptive to learning language, whether spoken or signed. If a child is diagnosed with type 1 Usher syndrome early on, before he or she loses the ability to see, that child is more likely to benefit from the full spectrum of intervention strategies that can help him or her participate more fully in life's activities.

Because of the balance problems associated with type 1 Usher syndrome, children with this diagnosis may be slow to sit without support and typically may not walk independently before they are 18 months old. These children usually begin to develop vision problems in early childhood, almost always by the time they reach age 10. Vision problems most often begin with difficulty seeing at night (parents have reported that very young children may not see well dim environments – not reaching for a bottle in a dimly lit nursery, etc.), but tend to progress rapidly until the person is completely blind.

The estimated prevalence worldwide of Usher syndrome type I is three to six per 100,000 people in the general population. Type I has been found to be more common in people of Ashkenazi Jewish ancestry (central and eastern European) and in the French-Acadian populations (Louisiana).

Type 2: Children with type 2 Usher syndrome are born with moderate to severe hearing loss and normal balance. Although the severity of hearing loss varies, most of these children can benefit from hearing aids and can communicate orally. The vision problems in type 2 Usher syndrome tend to progress more slowly than those in type 1, with the onset of RP often not apparent until the teens.

Type 3: Children with type 3 Usher syndrome have normal hearing at birth. Although most children with the disorder have normal to near-normal balance, some may develop balance problems later on. Hearing and sight worsen over time, but the rate at which they decline can vary from person to person, even within the same family. A person with type 3 Usher syndrome may develop hearing loss by the teens, and he or she will usually require hearing aids by mid- to late adulthood. Night blindness usually begins sometime during puberty. Blind spots appear by the late teens to early adulthood, and, by mid-adulthood, the person is usually legally blind.

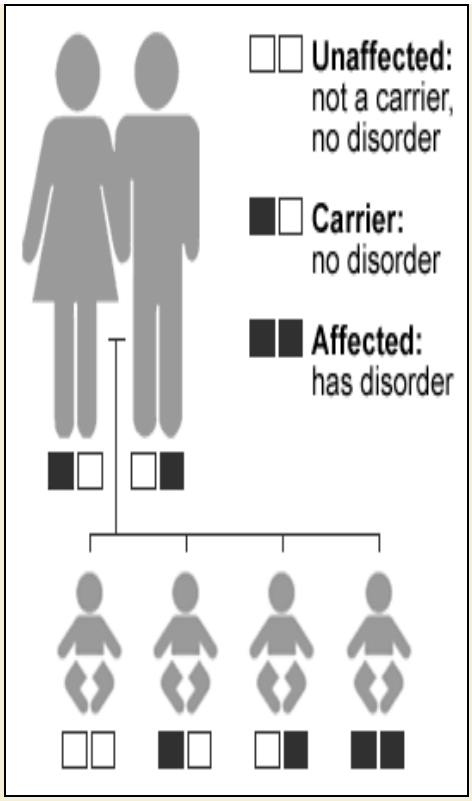
	Type 1	Type 2	Type 3
Hearing	Profound deafness in both ears from birth	Moderate to severe hearing loss from birth	Normal at birth; progressive loss in childhood or early teens
Vision	Decreased night vision before age 10, ongoing loss of vision	Decreased night vision begins in late childhood or teens; ongoing loss of vision	Varies in severity; night vision problems often begin in teens; ongoing loss of vision
Vestibular function (balance)	Balance problems from birth	Normal	Normal to near-normal, chance of later problems

Who is Affected by Usher syndrome? Approximately three to six percent of all children who are deaf and another three to six percent of children who are hard-of-hearing have Usher syndrome. In developed countries such as the United States, about four babies in every 100,000 births have Usher syndrome. It is important that ALL children who have hearing loss have regular vision screenings and appropriate eye care checks / follow-up to ensure that vision is monitored appropriately throughout the individual's life span. Any evidence of poor night vision and/or field loss should be reported to an eye care specialist.

What Causes Usher syndrome? Usher syndrome is inherited, which means that it is passed from parents to their children through genes. Genes are located in almost every cell of the body. Genes contain instructions that tell cells what to do. Every person inherits two copies of each gene, one from each parent. Sometimes genes are altered, or mutated. Mutated genes may cause cells to act differently than expected.

Usher syndrome is inherited as an autosomal recessive trait. The term ***autosomal*** means that the mutated gene is not located on either of the chromosomes that determine a person's sex; in other words, both males and females can have the disorder and can pass it along to a child. The word ***recessive*** means that, to have Usher syndrome, a person must receive a mutated form of the Usher syndrome gene from each parent. If a child has a mutation in one Usher syndrome gene but the other gene is normal, he or she is predicted to have normal vision and hearing. People with a mutation in a gene that can cause an autosomal recessive disorder are called ***carriers,*** because they "carry" the gene with a mutation, but show no symptoms of the disorder. If both parents are carriers of a mutated gene for Usher syndrome, they will have a one-in-four chance of having a child with Usher syndrome with each birth.

Usually, parents who have normal hearing and vision do not know if they are carriers of an Usher syndrome gene mutation. Currently, it is not possible to determine whether a person who does not have a family history of Usher syndrome is a carrier. Scientists at the National Institute on Deafness and Other Communication Disorders are hoping to change this situation, however, as they learn more about the genes responsible for Usher syndrome.



Chances of Inheriting a Recessive Disorder

Genetic disorders can be caused by a change(s) in a gene. Every individual has two copies of the same gene. Genetic disorders are inherited in different ways. Usher syndrome is a **recessive** disorder.

Recessive means a person must inherit a change in the same gene from each parent in order to have the disorder

A person with one changed gene does not have the disorder, but can pass either the changed or the unchanged gene on to his or her child

An individual with Usher syndrome usually has inherited a change in the same gene from each parent

An individual who has one changed Usher syndrome gene is called a **carrier**. When two carriers of the same Usher syndrome gene have a child together, with each birth there is a: 1-in-4 chance of having a child with Usher syndrome 2-in-4 chance of having a child who is a carrier 1-in-4 chance of having a child who neither has Usher syndrome nor is a carrier

How is Usher syndrome diagnosed? Because Usher syndrome affects hearing, balance, and vision, diagnosis of the disorder usually includes the evaluation of all three senses. Evaluation of the eyes may include a visual field test to measure a person's peripheral vision, an electroretinogram (ERG) to measure the electrical response of the eye's light-sensitive cells, and a retinal examination to observe the retina and other structures in the back of the eye. A hearing (audiologic) evaluation measures how loud sounds at a range of frequencies need to be before a person can hear them. An electronystagmogram (ENG) measures involuntary eye movements that could signify a balance problem.

Early diagnosis of Usher syndrome is very important. The earlier that parents know if their child has Usher syndrome, the sooner that child can begin early intervention (0-3 services) and special educational (3-21 years) programs to manage the loss of hearing and vision. A certified teacher of children who are deaf will be an important team member from the onset of the diagnosis of the hearing loss. As the vision loss becomes apparent, a certified teacher of children with visual impairment (TVI) will be needed, as well as a certified orientation and mobility specialist (COMS) for travel training needs. The child will qualify for services from the state deafblind project.

Families may be interested in connecting with other families of children with Usher syndrome, as well as the child him or herself. Role models can be very helpful.

Is genetic testing for Usher syndrome available? So far, 11 genetic loci (a segment of chromosome on which a certain gene is located) have been found to cause Usher syndrome, and nine genes have been pinpointed that cause the disorder. They are:

- Type 1 Usher syndrome: *MY07A*, *USH1C*, *CDH23*, *PCDH15*, *SANS*
- Type 2 Usher syndrome: *USH2A*, *VLGR1, WHRN*
- Type 3 Usher syndrome: *USH3A*

With so many possible genes involved in Usher syndrome, genetic tests for the disorder are not conducted on a widespread basis. Diagnosis of Usher syndrome is usually performed through hearing, balance, and vision tests. Genetic testing for a few of the identified genes is clinically available. To learn about laboratories that conduct clinical testing, visit the web site www.GeneTests.org and search the laboratory directory by typing in the term "Usher syndrome." Genetic testing for additional Usher syndrome genes may be available through clinical research studies. To learn about clinical trials that include genetic testing for Usher syndrome, visit the Web site www.clinicaltrials.gov and type in the search term "Usher syndrome" or "Usher genetic testing."

How is Usher Syndrome Treated? Currently, there is no cure for Usher syndrome. The best treatment involves early identification so that educational programs can begin as soon as possible. The exact nature of these programs will depend on the severity of the hearing and vision loss as well as the age and abilities of the person. Typically treatment will include hearing aids, assistive listening devices, cochlear implants, or other communication methods such as American Sign Language; orientation and mobility training; and communication services and independent-living training that may include Braille instruction, low-vision services, or auditory training.

Where can I get more information? NIDCD maintains a directory of organizations that can answer questions and provide printed or electronic information on Usher syndrome. Please see the list of organizations at www.nidcd.nih.gov/directory.

NIDCD Information Clearinghouse
1 Communication Avenue
Bethesda, MD 20892-3456
Fax: (301) 770-8977

Toll-free Voice: (800) 241-1044
Toll-free TTY: (800) 241-1055
E-mail: mailto:nidcdinfo@nidcd.nih.gov

For more information about the Colorado Services for Children and Youth with Combined Vision and Hearing Loss Project:

Colorado Department of Education
Phone Number: 303-866-6694 – Ask to speak with a Deaf-Blind Specialist on Staff
Fax: 303-866-6918
Deafblindness Webpage

Fact Sheets from the Colorado Services to Children and Youth with Combined Vision and Hearing Loss Project are to be used by both families and professionals serving individuals with vision and hearing loss. The information applies to children, birth through 21 years of age. The purpose of the Fact Sheet is to give general information on a specific topic. The contents of this Fact Sheet were developed under a grant from the United States Department of Education (US DOE), #H326C080044. However, these contents do not necessarily represent the policy of the US DOE and you should not assume endorsement by the Federal Government. More specific information for an individual student can be provided through personalized technical assistance available from the project. For more information call (303) 866-6681 or (303) 866-6605. Reviewed: 2/17