

Usher Syndrome

What Is Usher Syndrome?

Usher syndrome (US) is the most common condition that involves both hearing and vision problems. A syndrome is a disease or disorder that has more than one feature or symptom. The major symptoms of US are hearing impairment and an eye disorder called retinitis pigmentosa, in which vision worsens over time. Some people with US also have balance problems. There are three different types of US. Although it was first described by Albrecht Von Graefe in 1858, US was named for Charles Usher, a British eye doctor, who believed that this condition was inherited or passed from parents to their children.

Who Is Affected by Usher Syndrome?

More than half of the estimated 16,000 deaf-blind people in the United States are believed to have US.

What Causes Usher Syndrome?

US is inherited or passed from parents to their children through genes. Genes are located in every cell of the body and contain the instructions that tell cells what to do. Some genes specify traits such as hair color. Other genes are involved in the development of body parts, such as the ear. Still others determine how parts of the body work. Each person inherits two copies of each gene; one gene comes from each parent. Sometimes genes are altered or mutated. Mutated genes may cause cells to act differently than expected.

US is passed along in families by autosomal recessive inheritance, which requires two copies of the US gene before the disorder is seen. Each parent of a child with US usually has one standard and one mutated US gene. A child with US receives two mutated genes, one from each parent. Usually parents are unaware that they have or carry a US gene. This is because they would need two of the mutated genes in order to have signs of US. Presently, at least eight different genes are thought to cause the various types of US.

What Are the Types of Usher Syndrome?

The three types of US are called US type 1 (US1), US type 2 (US2), and US type 3 (US3). US1 and US2 are the most common types of US. Together, US1 and US2 account for approximately 10 percent of all cases of children who are born deaf.

What Are the Characteristics of the Three Types of Usher Syndrome?

People with US1 are profoundly deaf from birth and have severe balance problems. Many of these individuals obtain little or no benefit from hearing aids. Most use sign language as their primary means of communication. Because of the balance problems, children with US1 are slow to sit without support

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and rarely learn to walk before they are 18 months old. These children usually begin to develop vision problems by the time they are 10. Visual problems most often begin with difficulty seeing at night, but tend to progress rapidly until the individual is completely blind.

Those with US2 are born with moderate to severe hearing impairment and normal balance. Although the severity of hearing impairment varies, most of these children perform well in regular classrooms and can benefit from hearing aids. These children most commonly use speech to communicate. Retinitis pigmentosa, which is a degeneration of the retina or the part of the eye that receives images of objects, is characterized by blind spots that begin to appear shortly after the teenage years. The visual problems in US2 tend to progress more slowly than the visual problems in US1. When an individual's vision deteriorates to blindness, his or her ability to read speech from the lips is lost.

Children born with USH3 have normal hearing and normal to near-normal balance. Hearing worsens over time. Children develop noticeable hearing problems by their teenage years and usually become deaf by mid- to late adulthood. Retinitis pigmentosa in the form of night blindness usually begins sometime during puberty. Blind spots appear by the late teenage years to early adulthood. By mid-adulthood, the individual is usually blind.

How Is Usher Syndrome Diagnosed?

Hearing loss and retinitis pigmentosa are rarely found in combination. Therefore, most people who have retinitis pigmentosa and hearing loss probably have US1 or US2. Special tests such as electronystagmography (ENG) to detect balance problems and electroretinography (ERG) to detect retinitis pigmentosa help doctors to detect US early. Early diagnosis is important in order to begin special educational training programs to help the individual deal with the combined hearing and vision difficulties.

How Is Usher Syndrome Treated?

Presently, there is no cure for US. The best treatment involves early identification in order to begin educational programs. The exact nature of these educational programs will depend on the severity of the hearing and vision impairments as well as the age and abilities of the individual. Typically individuals will benefit from adjustment and career counseling; access to technology such as hearing aids, assistive listening devices, or cochlear implants; orientation and mobility training; and communication services and independent living training that may include Braille instruction, low vision services, or auditory training.

What Research Is Being Conducted on Usher Syndrome?

The current emphasis of US research is locating the genes that cause the syndrome and identifying the function of those genes. This research will lead to improved genetic counseling and early diagnosis, and may eventually expand treatment options. Scientists are also developing mice that have the same characteristics as humans who have the various types of US. Mouse models will make it easier to determine the function of the various genes involved in US. Research is also being conducted to improve the early identification of children with the syndrome. Treatment strategies such as the use of cochlear implants for hearing impairment and intervention strategies to alleviate retinitis pigmentosa are also being examined.

Where Can I Get Additional Information?

American Association of the Deaf-Blind
814 Thayer Avenue, Suite 302
Silver Spring, MD 20910
(301) 588-6545 (TTY)
(301) 588-8705 (Fax)
www.tr.wosc.osshe.edu/dblink/aadb.htm (Internet)

Better Hearing Institute
515 King Street, Suite 420
Alexandria, VA 22314
(703) 684-3391 (Voice/TTY)
(800) EAR-WELL (Toll-free Voice/TTY)
(703) 684-3394 (Fax)
www.betterhearing.org (Internet)

**Boys Town National Research Hospital
Genetics Department, Usher Syndrome Project**
555 North 30th Street
Omaha, NE 68131
(800) 835-1468 (Toll-free Voice/TTY)
(402) 498-6331 (Fax)
www.boystown.org/genetics/usher.htm (Internet)

The Foundation Fighting Blindness
Executive Plaza 1, Suite 800
11350 McCormick Road
Hunt Valley, MD 21031-1014
(410) 785-1414 (Voice)
(888) 394-3937 (Toll-free Voice)
(800) 683-5551 (Toll-free TTY)
(410) 771-9470 (Fax)
www.blindness.org (Internet)

**Helen Keller National Center for
Deaf-Blind Youths & Adults**
111 Middle Neck Road
Sands Point, NY 11050
(516) 944-8900 (Voice)
(516) 944-8637 (TTY)
(516) 944-7302 (Fax)
www.helenkeller.org/national (Internet)

**National Organization for Rare Disorders
(NORD)**
P.O. Box 8923
New Fairfield, CT 06811
(203) 746-6518 (Voice)
(800) 999-6673 (Toll-free Voice)
(203) 746-6481 (Fax)
www.rarediseases.org (Internet)

Self Help for Hard of Hearing People
7910 Woodmont Avenue, Suite 1200
Bethesda, MD 20814
(301) 657-2248 (Voice)
(301) 657-2249 (TTY)
(301) 913-9413 (Fax)
www.shhh.org (Internet)

**For further scientific information
about the molecular biology of
Usher syndrome:**

Hereditary Hearing Loss Homepage
dnalab-www.uia.ac.be/dnalab/hhh (Internet)