



Usher Syndrome ~ Overview

Usher syndrome is a family of genetic disorders characterized by both early hearing loss, and gradual vision loss. There are three distinct types described below.

Medical Description

The genetic changes responsible for Usher syndrome damage the sensory receptor cells (hair cells) in the inner ear, causing sensorineural hearing loss. This means that a structure in the inner ear, called the cochlea, is not able to detect sound and send information about it to the brain. In Usher Syndrome type 1, hair cells in the vestibular portion of the inner ear are also damaged, resulting in balance problems. In many people with Usher syndrome, these problems are present at birth and detected in the first years of life.

Retinitis pigmentosa is the medical term for the type of gradual vision loss experienced by people with Usher syndrome. This vision

loss is due to the slow death of rod and/or cone photoreceptor cells in the retina.

Early Symptoms

Parents of children with Usher syndrome are usually the first to notice that their child has limited hearing, so hearing problems are usually diagnosed in the first few years of life. At this point, vision loss will likely not be apparent, although subtle changes might be detected with vision testing.

When vision loss begins to be apparent, the first signs are usually a loss of night vision, followed by the gradual narrowing of a person's vision field.

Diagnosis

Early hearing loss can be detected and diagnosed with standard audiologic testing, which determines what frequency of sounds a child can hear, and how loud the sounds at these frequencies must be before they are audible.

	Type 1	Type 2	Type 3
Hearing	Severe deafness in both ears from birth	Moderate to severe hearing loss from birth usually in higher frequencies	No hearing loss at birth, but slow loss of hearing starting in childhood or teens
Vision	Slow vision loss starts with loss of night vision usually in childhood	Slow vision loss starts with loss of night vision in late childhood or teens	Timing and severity of vision loss vary, but most often night vision loss begins in teens
Balance	Balance problems from birth	No balance problems	Minimal/no balance problems at birth, symptoms many get worse with age

Based on a table created by the US National Institute on Deafness & other Communication Disorders.

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Children deaf at birth or diagnosed in early life should be screened for vision problems to detect Usher syndrome including:

- **Visual field** testing to measure peripheral vision;
- A detailed **retinal examination**;
- **ERG** (electroretinogram) measures the retina's electrical responses to light flashes. A large contact lens is placed on the eye to record them.
- **ENG** (electronystagmogram) may also be useful, revealing abnormal eye movements that sometimes accompany vision loss.

Balance tests are also done to clarify the diagnosis.

Usher syndrome is typically diagnosed with these tests. However, genetic testing – which can identify the mutated gene in about 50% of cases – is becoming more available.

Genetic Causes

Mutations (changes) in nine specific genes have been associated with Usher.

Usher Type 1: *MYO7A, USH1C, CDH23, PCDH15, SANS*;

Usher Type 2: *USH2A, VLGR1, WHRN*;

Usher Type 3: *USH3A*

Clinical blood tests for some of these genes are available; ask your genetic counsellor.

Treatment

Hearing loss in Usher syndrome cannot be reversed (at present); however, young children diagnosed today are often treated with cochlear implants. These devices stimulate the nerves of the inner ear directly, providing a substitute for natural hearing. If

hearing is less severely affected, hearing aids may also be beneficial.

No treatments are currently approved to prevent or slow the vision loss associated with Usher syndrome. However, it is important to have regular eye exams even if your vision is not changing – to avoid serious but treatable complications that might further impair your vision, such as cataracts and macular edema.

Research

Several research groups are working to develop therapies for Usher syndrome including gene therapies in which new genetic material is inserted into the affected retinal cells, to restore function. Gene therapies are specific to only one genetic type of Usher syndrome. A gene therapy, called UshStat®, has been approved for clinical trials in the USA. These trials will likely start in the next few months. UshStat® is designed to treat defects in the for the MYO7A gene (which causes Usher type 1B). A gene therapy for the WHRN gene (type 2D) is also being tested in the laboratory.

The FFB supports scientists working to understand the causes of vision loss and to develop treatments. In addition to gene therapies, FFB-funded scientists are working on many approaches to slow or even reverse retinal disease. To subscribe to our print and/or electronic newsletters on the latest vision research, call 1-800-4611-3331 today or email info@ffb.ca.

Updated Sept. 2011: Reviewed by Dr. Patrick Yang, Ophthalmology Resident, University of Toronto, and Dr. Bill Stell, Director of Research Programs, FFB.



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