



## What is Usher syndrome?

Usher syndrome is an inherited disease characterized by hearing loss and progressive vision loss. Damaged proteins are unable to maintain healthy cells in the ear, important for both hearing and balance, and healthy cells in the eyes important for sight. Researchers have divided Usher syndrome into three types (and further subtypes) depending on the age of onset and severity of symptoms. The different Usher syndromes are caused by different damaged proteins and may have varying severity. The following information focuses on Usher syndrome types IF and III.<sup>1</sup>

## What are the symptoms of Usher syndrome and what treatment is available?

The symptoms of Usher syndrome may include<sup>1</sup>:

- Hearing loss in both ears
- Retinitis pigmentosa (type of blindness that gets progressively worse over time)
- Problems with balance

### Usher syndrome type IF<sup>1</sup>:

Symptoms begin early in life and include:

- Severe to profound hearing loss in both ears present at birth or within the first year of life
- Balance problems leading to delays in motor development, sitting, and walking
- Onset of retinitis pigmentosa during childhood with rapid progression to blindness

### Usher syndrome type III<sup>2</sup>:

Symptoms appear in late childhood or adolescence and can vary between individuals. They include:

- Moderate to severe hearing loss starting after development of speech, and progressing to profound hearing loss by middle age,
- Retinitis pigmentosa
- Inner ear problems that may cause balance difficulties

There is no cure for Usher syndrome, but a typical lifespan is expected. While normal hearing aids are usually not useful, early use of a special hearing aid called a cochlear implant may be considered.<sup>3</sup>

## How are Usher syndrome types 1F and 3 inherited?

All types of Usher syndrome are autosomal-recessive diseases. Usher syndrome type IF is caused by mutations in the *PCDH15* gene, and Usher syndrome type III is caused by mutations in the *CLRN1* gene.<sup>2,3</sup> An individual who inherits one copy of a *PCDH15* or *CLRN1* gene mutation is a “carrier” and is not expected to have related health problems. An individual who inherits two mutations in the same gene, one from each parent, is expected to be affected.

If both members of a couple are carriers of a mutation in the same gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

## Who is at risk for Usher syndrome?

Usher syndrome types IF and III can occur in individuals of all races and ethnicities, but are known to be more common in individuals of Ashkenazi Jewish ancestry. Worldwide, Usher syndrome is estimated to affect between 1 in 31,000 to 1 in 16,000 people.<sup>1</sup> In the Ashkenazi Jewish population an estimated, 1 in 147 individuals is a carrier for Usher type IF and 1 in 120 individuals is a carrier for Usher type III.<sup>4</sup>





## CARRIER SCREEN

If there is no family history, the risk for an individual to be a carrier depends on an individual's ethnic background as noted above.

### What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

### What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

### Where can I get more information?

- Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/usher-syndrome>
- National Institute on Deafness and Other Communication Disorders: <http://www.nidcd.nih.gov/health/hearing/pages/usher.aspx>
- Helen Keller National Center for Deaf-Blind Youths and Adults: <http://www.hknc.org/WhoWeServeUsher.htm>

### References

1. Millian et al. *Journal of Ophthalmology*, Volume 2011, Article ID #417217, 8 pages
2. Usher Syndrome. <http://ghr.nlm.nih.gov/condition/usher-syndrome> Accessed March 6, 2012.
3. Keats, BJB, et al. Usher Syndrome Type I. <http://www.ncbi.nlm.nih.gov/books/NBK12665> Accessed March 6, 2012.
4. Scott, SA, et al. Experience with Carrier Screening and Prenatal Diagnosis for 16 Ashkenazi Jewish Genetic Diseases. *Hum Mut.* 2010; 31(11): 1240-1250.

