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Usher's Syndrome is a genetic disease that affects hearing, vision, and sometimes balance.

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5018 NE 15th Ave.
Portland, OR 97211
1-800-837-8428
info@vestibular.org
vestibular.org

Usher's Syndrome

ByRobert Allen, AuD - Topple Diagnostics
Reviewed by Eric Slattery, MD - House of Hearing

WHAT IS USHER'S SYNDROME?

Usher's syndrome is an inherited genetic disease that affects both hearing and vision, as well as sometimes affecting balance function. It is the most common dual sensory impairment syndrome that affects both hearing and vision.1 It is an inherited genetic disease that is present at birth, but most people with Usher's syndrome are diagnosed during childhood or adolescence.[1,3] The main symptoms are hearing loss/deafness and progressive vision loss through a condition known as retinitis pigmentosa (RP).[2] Researchers have identified at least 9 different genes that can cause Usher's syndrome.[1,4,6] Usher's syndrome is grouped into three categories depending on the characteristics that are present.[2,3]

WHAT ARE THE SYMPTOMS OF USHER'S SYNDROME?

The main symptoms are hearing loss/deafness and vision loss. Most people with Usher's syndrome are born with moderate/severe hearing loss; however, in some people, the hearing loss can develop in adolescence. Balance function can also be compromised in patients with Usher's syndrome, but it is not present in all patients. The hearing loss is caused by abnormal development of the sensory hair cells in the inner ear.[1]

Vision loss in Usher's syndrome is due to a condition called retinitis pigmentosa (RP).[2,3] RP is a condition that causes progressive vision loss due breakdown of the cells in the retina, the part of the eye that is light sensitive.[2,3] Initially, RP causes night blindness and peripheral vision loss. As the condition worsens, the vision continues to narrow until only central vision remains. This is also sometimes referred to as tunnel vision.[2,3]

Balance function can be affected in patients with Usher's syndrome either through abnormal development of the sensory hair cells in the balance organs in the ears or through the difficulty with low light vision and progressive vision loss caused by RP.[1,2,7]

WHAT ARE THE TYPES OF USHER'S SYNDROME?

Usher's syndrome is categorized into three types based on the characteristics of the impairment in three areas: hearing, vision, and balance. Types 1 and 2 are the most common presentations.[2]

Type 1:

- Profound hearing loss present at birth (may only respond to very loud sounds and receive little to no benefit from traditional hearing aids)
- Night blindness by age 10 and severe vision loss by middle age
- Severe balance problems, including sitting up and walking

Type 2:

- Moderate to severe hearing loss in early childhood
- Night blindness by teenage years, severe vision loss by middle age
- No balance impairment

Type 3:

- Born with normal hearing, but hearing loss begins as a child
- Night blindness by teenage years, severe vision loss by middle age
- Normal balance[1,2,7]

HOW IS USHER'S SYNDROME DIAGNOSED?

Diagnosis involves a multidisciplinary care team of specialists to evaluate the eye and ear issues associated with Usher's syndrome as well as a detailed medical history focusing on impairments associated with the condition.[1,2,7] Optometry/ophthalmology can evaluate for signs of structural changes to the retina as well as evaluate for signs of peripheral vision loss indicating RP.[2,4,5] Audiology/ENT can evaluate for balance problems using specialized diagnostic tools to track abnormal eye movements associated with balance dysfunction as well as evaluating the nature and severity of hearing loss.[1,4,6,7] Genetic testing can also aid in the diagnosis, and different genes have been implicated in the different types of Usher's syndrome.[3,5,6]

- Type 1 has been associated with CDH23, MY07A, PCHD15, USH1C, USH1G
- Type 2 has been associated with DFNB31, ADGRV1, USH2A
- Type 3 has been associated with CLRN[13,5,7]

HOW IS USHER'S SYNDROME TREATED?

There is no current cure for Usher's syndrome. Management consists of addressing the problems of the patient

including hearing loss, vision issues, and balance problems. Early diagnosis is paramount in providing better outcomes as educational programs can be better tailored for the individual.[1] Recent research has suggested that vitamin A supplementation may be beneficial in the management of retinitis pigmentosa. Research is ongoing to identify other genes associated with Usher's syndrome as well as other methods for early detection and identification.[1]

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