
Usher syndrome

Summary

- Usher syndrome is a genetic disorder characterised by the progressive loss of both hearing and vision.
 - Some people with Usher syndrome also experience problems with their balance.
 - For a child to develop Usher syndrome, they must inherit the particular faulty gene from both their mother and their father.
 - There are information and support services available.
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Usher syndrome is a genetic disorder characterised by partial or total hearing loss and a gradual vision loss caused by retinitis pigmentosa (RP).

The name retinitis pigmentosa refers to the deposits of black pigment found in the retina of people with this condition. This is a form of eye disease that results in the gradual loss of vision over time, due to deterioration of the retina. At first, the person becomes night blind, as the eyes can no longer see in dim light. Over time, the field of vision diminishes until the person has only a 'tunnel' of central vision. Some people with Usher syndrome also experience problems with their balance.

Usher syndrome is categorised into three broad groups according to the type and severity of symptoms. Type 1 and Type 2 account for approximately five per cent of all children born deaf and hard of hearing. There is no cure. Services aim to help the person prepare for and cope with this dual loss.

Symptoms of Usher syndrome

The symptoms and signs of Usher syndrome depend on the type, but generally include:

- hearing loss leading to deafness
- loss of vision leading to blindness
- balance problems
- speech difficulties.

Balance explained

The human body has three systems that contribute to balance. They are:

- **The eyes** – simply looking around tells our brain where we are.
- **Proprioception system** – this is the information the brain receives from special receptors in muscle, joints and skin, which sense the movement and position of the joints.
- **Vestibular system** – this is within the ear and consists of a series of fluid-filled canals set at different angles. When the head moves, the rolling fluid tells the brain exactly how far, how fast and in what direction the head is moving, by relaying the information down the vestibular nerve. If the brain knows the position of the head, it can work out the position of the rest of the body.

Types of Usher syndrome

Usher syndrome is categorised into three broad groups according to the type and severity of symptoms. These groups are called Type 1, Type 2 and Type 3. Each type is further divided into subtypes.

Usher syndrome Type 1 (US1)

Although there are six subtypes, the characteristics of Usher syndrome Type 1 include:

- The person is profoundly deaf from birth.
 - Speech difficulties are evident.
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- Other means of communication, like sign language (Auslan), are needed.
- Balance may be affected and children are often late to sit, stand and walk.
- Eye problems vary with each person, but may start to develop at around ten years of age.
- Vision loss varies with each person, but often develops in the first ten years of life.

Usher syndrome Type 2 (US2)

There are three subtypes of Type 2. The characteristics of Usher syndrome Type 2 include:

- Moderate loss of hearing is experienced in the lower sound frequency range.
- Severe loss of hearing is experienced in the higher sound frequency range.
- Balance may be affected in later years.
- Vision loss, such as blind spots, starts to develop during the teenage years and may progress during life.

Usher syndrome Type 3 (US3)

Only one subtype of Type 3 has been discovered to date. The characteristics of Usher syndrome Type 3 include:

- The person is born with normal hearing and close to normal balance.
- The loss of hearing becomes more pronounced as the person gets older.
- Measurable hearing loss occurs by puberty.
- Vision loss starts to develop during the teenage years and may progress during life.
- Balance deteriorates as the person gets older.
- The majority of people with US3 live in Finland.

The cause of Usher syndrome

Humans have 46 paired chromosomes, with two sex chromosomes that decide gender and 44 chromosomes that dictate other factors. The estimated 30,000 to 40,000 genes are beaded along these tightly bundled strands. These genes also come in pairs – one from each parent.

The different types of Usher syndrome are caused by mutations of different genes. Researchers believe there are about 15 genes that can cause Usher syndrome. For a child to develop Usher syndrome, they must inherit the particular Usher gene from both their mother and their father. If just one Usher gene is inherited, the child won't develop the condition, but will be a carrier. If the parents are carriers of two different Usher genes, their child won't develop either syndrome, but will be a carrier for both.

Diagnosis of Usher syndrome

Usher syndrome is the most common genetic cause of combined vision and hearing impairment and deafblindness. Tests to confirm the diagnosis can include:

- eye tests to check for retinitis pigmentosa
- hearing tests
- tests to determine the person's degree of balance, such as electronystagmography (ENG).

Support for Usher syndrome

There is no cure for Usher syndrome. The amount of support required depends on the severity, but may include:

- hearing aids
- cochlear implants
- communication skills training
- orientation and mobility training
- adaptive technology
- braille
- counselling
- access to low vision services

- access to community support.

Where to get help

- Your doctor
- Able Australia Tel. 1300 225 369, TTY (03) 9882 6786
- Vicdeaf Tel. (03) 9473 1111, TTY (03) 9473 1199, toll free for country callers Tel. 1300 780 225, TTY 1300 780 235
- Royal Children's Hospital Audiology Department Tel. (03) 9345 5550
- Royal Children's Hospital Ophthalmology Department Tel. (03) 9345 5630

Things to remember

- Usher syndrome is a genetic disorder characterised by the progressive loss of both hearing and vision.
- Some people with Usher syndrome also experience problems with their balance.
- For a child to develop Usher syndrome, they must inherit the particular faulty gene from both their mother and their father.
- There are information and support services available.

This page has been produced in consultation with and approved by:

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