Usher syndrome

Summary

- Usher syndrome is a genetic condition.
- People with Usher syndrome often have hearing and vision impairment.
- Some people with Usher syndrome also experience problems with their balance.
- For a child to have Usher syndrome, they must inherit the gene change from both their mother and their father.
- There are information and support services available for people and families living with Usher syndrome.

Usher syndrome is a genetic condition. It is the most common genetic cause of combined vision and hearing impairment and deafblindness. People with Usher syndrome often have partial or total hearing loss and a gradual vision loss caused by retinitis pigmentosa. 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 – Types 1, 2 and 3. Type 1 and Type 2 account for approximately five per cent of all children born with a hearing impairment.

There is currently no cure for Usher syndrome. Services aim to help the person and their family prepare for and cope with their vision and hearing loss.

Symptoms of Usher syndrome

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

- **hearing impairment and deafness**
- **vision impairment leading to blindness** – in Usher syndrome this is caused by retinitis pigmentosa (RP). RP is a form of eye disease that results in the gradual loss of vision over time, due to deterioration of the retina. At first, a person with RP becomes ‘night blind’, meaning that they can no longer see in dim light. Over time, their field of vision becomes smaller until they only have a ‘tunnel’ of central vision
- **balance problems** – can occur due to vision and inner ear impairments. This is because our ability to balance relies on information being sent to our brain from our:
  - **eyes**
  - **vestibular system** – a series of fluid-filled canals within the ear, set at different angles. When the head moves, the movement of the 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
  - **proprioception system** – special receptors in our muscles, joints and skin that sense the movement and position of the joints
- **speech difficulties**.

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)**

There are six subtypes of US1. The characteristics include:

- The person is profoundly deaf from birth.
- Speech difficulties are evident.
• 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 US2. The characteristics 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 US3 has been discovered to date. The characteristics 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.
• Most people with US3 are of Finnish descent.

**What causes Usher syndrome**

Usher syndrome is a genetic condition.

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 changes in different genes. Researchers believe there are about 15 genes that can cause Usher syndrome. Usher syndrome follows an autosomal recessive inheritance pattern. This means that for a child to develop Usher syndrome, they must inherit the same Usher gene change from both their mother and their father.

If just one changed 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 gene changes, their child won’t develop either syndrome, but will be a carrier for both.

**Diagnosis of Usher syndrome**

Tests to confirm a diagnosis of Usher syndrome can include:

• eye tests to check for retinitis pigmentosa
• hearing tests
• tests to determine the person’s degree of balance, such as electronystagmography (ENG)
• genetic testing.

**Living with Usher syndrome**

There is currently no treatment or cure for Usher syndrome. ‘Treatment’ involves managing the vision, hearing and balance problems involved with the condition.

Managing hearing impairment associated with Usher syndrome may involve the use of:

• hearing aids

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• cochlear implants
• adaptive technology
• Auslan sign language training
• other communication skills training
• services for people with a hearing impairment such as those offered by:
  ○ Hearing Australia
  ○ Better Hearing Australia
  ○ Expression Australia (previously Vicdeaf)
  ○ the Cochlear Implant Clinic at the Royal Eye and Ear Hospital
  ○ Aussie Deaf Kids.

Managing balance problems associated with Usher syndrome may involve vestibular rehabilitation therapy. This is an exercise-based program run by a vestibular physiotherapist. In children with Usher syndrome it aims to improve balance and coordination.

Managing vision problems associated with Usher syndrome may involve:
• learning to use Braille
• magnifying aids
• adaptive technology
• orientation and mobility training
• access to low-vision services, such as those offered by:
  ○ Vision Australia
  ○ Guide Dogs Victoria.

Support for people living with Usher syndrome

Support services available for people with Usher syndrome, include:
• support and advocacy groups
• genetic counselling
• community support.

The Genetic Support Network of Victoria (GSNV) is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by Usher syndrome.

UsherKids Australia is a support and advocacy group for children and families affected by Usher syndrome. Learn more about support available for families living with Usher syndrome.

Genetic testing and counselling for Usher syndrome

If you or your child have been diagnosed with Usher syndrome, or if it runs in your family, it can be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand Usher syndrome, what causes it, how it is inherited, and what a diagnosis means for your or your child’s health and development and plans for the future. Genetic counsellors are trained provide information and support that is sensitive to your family circumstances, culture and beliefs.

If Usher syndrome runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family, to find out your risk of passing that condition on to your child, or to arrange for prenatal tests.

Where to get help
• Your GP (doctor)
• Able Australia Tel. 1300 225 369

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