USHER SYNDROME

WHAT IS USHER SYNDROME?

Usher syndrome is a genetic condition characterised by hearing loss, progressive vision loss, and in some cases, vestibular dysfunction.

Vision loss in Usher syndrome is caused by retinitis pigmentosa. This condition affects the area at the back of the eye which usually senses light (the retina). The first symptom is usually night blindness, followed by tunnel vision – also known as peripheral vision loss.

Vestibular dysfunction means the ears, eyes, muscles, brain, & bones cannot work together in the usual way. This can lead to delayed motor skills & balance issues.

There are three clinical types of Usher syndrome, with type 1 being most severe:

<table>
<thead>
<tr>
<th></th>
<th>Type 1</th>
<th>Type 2</th>
<th>Type 3</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Hearing Loss</strong></td>
<td>From birth</td>
<td>From birth</td>
<td>Progressive</td>
</tr>
<tr>
<td></td>
<td>Severe</td>
<td>Moderate to severe</td>
<td>Moderate to severe</td>
</tr>
<tr>
<td><strong>Retinitis Pigmentosa</strong></td>
<td>First or second decade</td>
<td>Second or third decade</td>
<td>Variable</td>
</tr>
<tr>
<td><strong>Vestibular dysfunction</strong></td>
<td>Altered</td>
<td>Usually normal</td>
<td>Variable</td>
</tr>
</tbody>
</table>

HOW IS USHER SYNDROME INHERITED?

Every individual has 2 copies of each gene, one inherited from each parent. These genes may contain a mistake associated with Usher syndrome. Usher syndrome is caused by a mistake in both copies of an Usher gene.

People with a mistake in only one copy of an Usher gene are called carriers. Carriers will not show symptoms of Usher syndrome but may pass the mistake on if they have children. This means that two unaffected parents may have affected children.

2 carrier parents have a 25% chance of having a child with Usher syndrome, a 50% chance of a carrier child, and a 25% chance of an unaffected child.

This type of inheritance pattern is called autosomal recessive inheritance.

WHAT IS A GENETIC CONDITION?

Usher syndrome is a genetic condition, meaning it is caused by mistakes in DNA. DNA is present in almost every cell in our bodies and contains the instructions needed for our bodies to function.

Instructions are stored in genes. Mistakes in genes can cause genetic conditions. There are currently 11 genes known to cause different subtypes of Usher syndrome:

**Type 1:**

<table>
<thead>
<tr>
<th>Subtype</th>
<th>1B</th>
<th>1C</th>
<th>1D</th>
<th>1F</th>
<th>1G</th>
<th>1J</th>
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</thead>
<tbody>
<tr>
<td>Gene</td>
<td>MYO7A</td>
<td>USH1C</td>
<td>CDH23</td>
<td>PCDH15</td>
<td>USH1G</td>
<td>CIB2</td>
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</table>

**Type 2:**

<table>
<thead>
<tr>
<th>Subtype</th>
<th>2A</th>
<th>2C</th>
<th>2D</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gene</td>
<td>USH2A</td>
<td>ADGRV1</td>
<td>WHRN</td>
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</tbody>
</table>

**Type 3:**

<table>
<thead>
<tr>
<th>Subtype</th>
<th>3A</th>
<th>3B</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gene</td>
<td>CLRN1</td>
<td>USH3B</td>
</tr>
</tbody>
</table>
HOW IS USHER SYNDROME DIAGNOSED?

Genetic testing is necessary for a definitive diagnosis of Usher syndrome. When an individual is diagnosed, there is a chance that other family members are carriers of the same gene mistake. This can be determined through genetic testing.

Genetic testing in other family members can:

• Assist in early diagnosis of others
• Assess the probability of having a child with Usher syndrome

There are many considerations before genetic testing, which can be discussed with a genetic counsellor, who can also provide ongoing support for the family.

WHAT CAN BE DONE ABOUT USHER SYNDROME?

There is ongoing research into stem cell therapies, drug therapies, and genetic interventions for Usher syndrome. At the moment there is no cure, but there are interventions that can help manage symptoms.

These interventions are most effective early in life, so early diagnosis is very important.

HEARING

Hearing aids and cochlear implants can be considered to help live with hearing impairment. An audiologist can work with families dealing with Usher syndrome to better explain these options. Speech pathologists can support children with Usher syndrome with their communication. Auslan can also help in supplementing communication skills.

VISION

Mobility aids can help manage the orientation and mobility issues associated with Usher syndrome. Orientation & mobility specialists can work with families dealing with Usher syndrome to better explain these options, while ophthalmologists can manage and monitor eye health.

Since children with Usher syndrome are born with sight, it is important for families to plan for the future by giving these children environmental and practical knowledge that will help them as their vision deteriorates. Some examples of this include:

• Touch typing skills
• Learning public transport
• Training in the use of a mobility aid
• Environmental familiarisation (school, parks, etc.)

VESTIBULAR DYSFUNCTION

Occupational therapists and physiotherapists can help children with Type 1 Usher syndrome to meet their developmental milestones and compensate for vestibular dysfunction.

WHAT SUPPORT IS AVAILABLE?

People with Usher syndrome may experience many different negative emotions as they battle with their condition. Individuals and families dealing with Usher syndrome may find it beneficial to see a psychologist to manage their emotions and develop strategies to cope.

Across Australia, many different organisations can give support and information to people dealing with Usher syndrome or the symptoms associated with it.

**SUPPORT**

ABLE AUSTRALIA  
[www.ableaustralia.org.au](http://www.ableaustralia.org.au)

AUSSIE DEAF KIDS  
[www.aussiedeafkids.org.au](http://www.aussiedeafkids.org.au)

DEAF CHILDREN AUSTRALIA  
[www.deafchildrenaustralia.org.au](http://www.deafchildrenaustralia.org.au)

GUIDE DOGS AUSTRALIA  
[www.guidedogsaustralia.com](http://www.guidedogsaustralia.com)

VISION AUSTRALIA  
[www.visionaustralia.org.au](http://www.visionaustralia.org.au)

ROYAL INSTITUTE FOR DEAF AND BLIND CHILDREN  
[www.ridbc.org.au](http://www.ridbc.org.au)

**RESEARCH**

CENTRE FOR EYE RESEARCH  
[www.cera.org.au](http://www.cera.org.au)

LIONS EYE INSTITUTE  
[www.lei.org.au](http://www.lei.org.au)

RETTINA AUSTRALIA  
[www.retinaaustralia.org.au](http://www.retinaaustralia.org.au)

STEM CELLS AUSTRALIA  
[www.stemcellsaustralia.edu.au](http://www.stemcellsaustralia.edu.au)

SAVE SIGHT  
[www.savesightinstitute.org.au](http://www.savesightinstitute.org.au)

ROYAL INSTITUTE FOR DEAF AND BLIND CHILDREN  
[www.ridbc.org.au](http://www.ridbc.org.au)

**ADDITIONAL INFORMATION IS AVAILABLE AT:**

[www.usherkidsaustralia.com](http://www.usherkidsaustralia.com)