USHER SYNDROME

WHAT IS USHER SYNDROME?

Usher syndrome is a genetic condition characterised by hearing loss, progressive vision loss, and sometimes 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:

	Type 1	Type 2	Type 3
Hearing Loss	From birth Severe	From birth Moderate to severe	Progressive Moderate to severe
Retinitis Pigmentosa	First or second decade	Second or third decade	Variable
Vestibular dysfunction	Altered	Usually normal	Variable

WHAT IS A GENETIC CONDITION?

Usher syndrome is a genetic condition, meaning it is caused by mistakes in **genes**. Genes are made up of **DNA**. They contain instructions we need to function. Genes are stored in **chromosomes** which can be found in almost every cell in our bodies.

Currently, Usher syndrome is linked to 11 genes, each related to a different sub-type:

Type 1:

Subtype	1B	1C	1D	1F	1G	1J
Gene	*MYO7A*	*USH1C*	*CDH23*	*PCDH15*	*USH1G*	*CIB2*

Type 2:

Subtype	2A	2C	2D
Gene	*USH2A*	*ADGRV1*	*WHRN*

Type 3:

3A	3B
CLRN1	*USH3B*

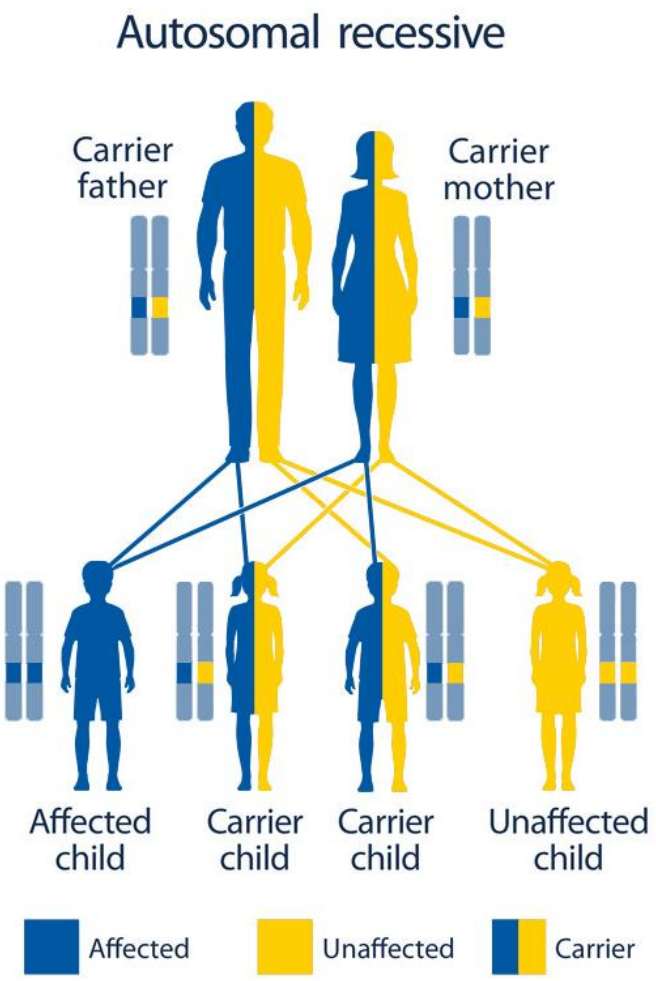
HOW IS USHER SYNDROME INHERITED?

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 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.**



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 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

AUSSIE DEAF KIDS
www.aussiedeafkids.org.au

DEAF CHILDREN AUSTRALIA
www.deafchildrenaustralia.org.au

GUIDE DOGS AUSTRALIA
www.guidedogsaustralia.com

VISION AUSTRALIA
www.visionaustralia.org.au

ROYAL INSTITUTE FOR DEAF AND BLIND CHILDREN
www.ridbc.org.au

RESEARCH

CENTRE FOR EYE RESEARCH
www.cera.org.au

LIONS EYE INSTITUTE
www.lei.org.au/

RETINA AUSTRALIA
www.retinaaustralia.org.au

STEM CELLS AUSTRALIA
www.stemcellsaustralia.edu.au

SAVE SIGHT
www.savesightinstitute.org.au