USHER SYNDROME

WHAT IS USHER SYNDROME?

Usher syndrome is a rare genetic condition characterised by hearing loss, progressive vision loss and, sometimes, vestibular (balance) dysfunction.

Vision loss in Usher syndrome is caused by **retinitis pigmentosa**, which affects the area at the back of the eye which senses light (the retina). The first symptom is usually night blindness, followed by progressive loss of peripheral vision. There are three clinical types of Usher syndrome:

	Type 1	Type 2	Type 3
Hearing loss	From birth Severe	From birth Moderate to severe	Progressive Moderate to severe
Vision loss	Usually affects individual within 1st or 2nd decade	Usually affects individual within 2nd or 3rd decade	Variable
Balance difficulties	From birth	Usually not affected	Variable

The above table gives a guide, but the extent of sensory loss varies between individuals and it is not currently possible to accurately predict the rate of vision loss.

WHAT IS A GENETIC CONDITION?

Genes contain instructions for our body to determine its physical characteristics. Usher syndrome is caused by genetic 'spelling mistakes' (these are sometimes referred to as **mutations** or **variants**).

Currently, Usher syndrome has been linked to mutations in 11 genes, each related to the 3 different sub-types:

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 two copies of each gene, one inherited from each parent. These genes may contain a mistake (or mutation) associated with Usher syndrome. A person is affected by Usher syndrome when there is 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 be affected with symptoms of Usher syndrome but may pass the mistake on if they have children. This means two unaffected parents who are carriers can have affected children. Two carrier parents have a 25% chance of having a child with Usher syndrome.

HOW IS USHER SYNDROME DIAGNOSED?

An eye examination and investigations including an **electroretinogram** (**ERG**), fundus autofluorescence (AF) and optical coherence tomography (OCT), can be carried out within an **ophthalmology** clinic to diagnose retinitis pigmentosa.

Diagnosis and monitoring of hearing loss are undertaken in an **audiology** clinic.

A **clinical diagnosis** of Usher syndrome may be made if someone is affected by both hearing loss and retinitis pigmentosa. A **genetic diagnosis** (via a blood test for genetic testing) can identify which gene and mutation is involved, and will be necessary to identify eligibility for future gene- and mutation-specific therapies.

If Usher syndrome is suspected a GP can request a referral to see a Genetic Eye Disease Specialist. If this service is not available locally the team at Moorfields Eye hospital (London) can be contacted using the address below, and can advise on appropriate next steps:

Dr Mariya Moosajee
Genetic Eye Disease Service
Moorfields Eye Hospital
162 City Road
London
EC1V 2PD

WHAT CAN BE DONE ABOUT USHER SYNDROME?

There is ongoing research into stem cell, gene and drug therapies 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. It is always important to consider the combined (**multi-sensory**) impact of hearing *and* vision loss. Children require qualified support (e.g. Teacher of the Deaf / Visually Impaired / Multi-Sensory Impaired) and this support can be documented and implemented through an **Education and Health Care (EHC) Plan**.

HEARING

Hearing aids and **cochlear implants** can be considered with an **audiologist** to help live with hearing loss. **Speech therapists** can support children with spoken communication. **Sign Language** is another communication option. Smaller adjustments like using subtitles, ensuring good acoustics, reducing background noise and educating family and friends about clear communication techniques can all help reduce the impact of hearing loss.

VISION

Ophthalmologists can manage and monitor eye health, and will decide if/when a child's vision loss makes them eligible to be certified as sight impaired. For everyday life the use of UV-protected tinted glasses/sunglasses and hats can help reduce glare and damage to the retina, whilst good lighting can help compensate for issues with night vision. A healthy balanced diet full of fresh fruit and vegetables is important, especially those rich in antioxidants. **Mobility aids** can help manage the mobility issues associated with Usher syndrome. These options can be discussed with an Orientation and Mobility Officer within the local authority's Sensory Support team.

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
- Training in the use of a mobility aid
- Environmental familiarisation (school, parks, public transport etc.)

VESTIBULAR DYSFUNCTION

Occupational therapists and **physiotherapists** can help children with Type 1 Usher syndrome to learn skills to compensate for vestibular dysfunction. Even with good compensation, living with poor balance can lead to additional tiredness and frustration, and children may need extra breaks or adjustments to help with this.

WHAT SUPPORT IS AVAILABLE?

People with Usher syndrome may experience many different emotions as they adjust to and live with their condition. Individuals and families dealing with Usher syndrome may find it beneficial to see a **counsellor or psychologist** to help talk through their feelings and develop strategies to cope. It can also be really helpful to talk to other people/families that have experience of Usher syndrome to help build a network of support.

Usher Kids UK (www.usherkidsuk.com) exists to support, connect and advocate for children and families in the UK that are living with Usher syndrome.

There are many organisations that can also give **support** and **information** to people living with Usher syndrome:

USHER KIDS UK
www.usherkidsuk.com

RETINA UK
www.retinauk.org.uk

SENSE
www.sense.org.uk

USHER COALITION
www.usher-syndrome.org

RNIB
www.rnib.org.uk

NATIONAL DEAF CHILDREN'S SOCIETY
www.ndcs.org.uk

GUIDE DOGS
www.guidedogs.org.uk

COCHLEAR IMPLANTED CHILDREN'S SUPPORT GROUP
www.cicsgroup.org.uk

MOLLY WATT TRUST
www.molly-watt-trust.org

USH TRUST REGISTRY
www.usher-registry.org

VICTA
www.victa.org.uk

MOORFIELDS EYE CHARITY
www.moorfieldseyecharity.org.uk

CUREUsher
www.cureusher.org

MORE INFORMATION IS AVAILABLE AT
WWW.USHERKIDSUK.COM

Usher Kids UK