Usher syndrome

What you need to know



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New diagnosis of Usher syndrome?

We are here to help...

We know that a diagnosis of Usher syndrome can lead to lots of questions. This pack is designed to give you access to information and support, to help you look ahead with confidence.

What is Usher syndrome?

There are roughly 2,000 children and young people aged 0–25yrs living with Usher syndrome in the UK. So whilst it may be rare, you are definitely not alone.

It is a rare genetic condition which causes:

- Hearing loss
- Progressive vision loss*
- In some cases, vestibular dysfunction (difficulties with balance)

Vision loss
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 difficulty seeing in the dark, followed by progressive loss of peripheral vision (reduced visual field).

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There are four clinical types of Usher syndrome:

Type 1
Hearing Loss
From birth – severe to profound
Vision Loss
Usually affects individual by age 10 to 20
Balance difficulties
From birth

Type 2
Hearing Loss
From birth – moderate to profound
Vision Loss
Usually affects individual by age 20 to 30
Balance difficulties
Usually not affected

Type 3
Hearing Loss
Progressive - moderate to severe
Vision Loss
Variable
Balance difficulties
Variable

Type 4
Hearing Loss
Progressive - beginning in adulthood
Vision Loss
Usually affects individual by age 50 to 60
Balance difficulties
No issues

The age and rate at which changes to vision take place varies between individuals, even with the same type of Usher syndrome.

There are other forms of combined hearing loss and retinitis pigmentosa. A genetic counsellor could provide more information about these.

Search **'What is Usher syndrome'** at **usherkidsuk.org** for more information.

How is it diagnosed?

These are the tests and services which are involved in diagnosis and management of Usher syndrome.

Stage 1: Diagnosis

Hearing

There are a variety of different hearing assessments to identify and monitor a child's hearing loss. The tests used will depend on your child's age and stage of development, ranging from Auditory Brainstem Response test for babies, to more well known hearing tests using headphones and a push button for older children. Where permanent hearing loss is found, the audiologists may suggest use of hearing aids or cochlear implants.

Genetics

If hearing loss has been identified in both ears, a genetic test should be offered by the paediatrician, Audiology or Genetics team. This test will use a blood sample from your child to look at genes involved in hearing pathways. You may be seen by a Genetics service to discuss the results, including what was found and whether there is any uncertainty or further tests needed in your child or in you to understand what the genetic results mean for your family.

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The **gene.vision** website is an excellent source of information on Usher syndrome.

Vision

A variety of tests will be performed to assess the retina of the child, including an electroretinogram (ERG) which provides flashes of light or patterns to look at with wires placed on the head to measure how light travels from the eye to the brain.

Balance

A variety of tests can be used to assess the balance system in the inner ear and brain, and identify what services or activities could be helpful. These may involve the child watching a small light move on a screen, wearing goggles whilst their head is gently moved, or sitting in a rotating chair to assess how the eyes move in response to motion.

Stage 2: After diagnosis

Eye clinic:

A number of retinal scans and vision tests will be repeated over the years depending on age and stage of the condition. These may be more frequent at first, and then become annual appointments.

Audiology or Hearing aid / Implant clinic:

Hearing loss will be monitored in regular clinic appointments. If changes are noticed between appointments, parents can request for their child to be seen for extra testing.

Specialist physiotherapy (if balance is affected, and if available in your area):

Children are encouraged to undertake fun activities to strengthen the balance function they have – or learn how to use other systems to assist and optimise balance, such as their sense of touch and sensation of movement in the joints (proprioception).

Sensory support team involvement (Teacher of Deaf / Vision Impairment / Multi-Sensory Impairment)

Co-ordinates support to address educational, social and emotional needs of child in school, home and social settings.

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Professionals you may meet

There can be many different professionals involved in the care of your child. It is helpful to know about their roles, so that you know what to expect and who to turn to if you have a question.

Ophthalmologist

An ophthalmologist is a medically trained doctor who examines and diagnoses eye conditions. They will review tests carried out by orthoptists and provide useful information about changes to the eye.

Genetic Counsellor

A genetic counsellor can explain the results of genetic tests, and how these are likely to impact your child, as well as what this means for any future children you might have.

Eye Clinic Liaison Officer (ECLO)

An ECLO is a great point of contact in your eye clinic. They can answer questions about what to expect in the eye clinic, give you information about relevant benefits and local services, and connect you with emotional and practical support.

Teachers of the Deaf and Visually Impaired

Qualified Teachers of children with Multi-Sensory Impairment (QTMSIs) are specialist teachers who have additional qualifications in working with children and young people who have hearing and vision loss. They work alongside Teachers of the Deaf and Visual Impairment to help ensure the combined impact of dual sensory loss is taken into account.

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What can I do to help?



Give yourself time to absorb the diagnosis and take in new information. It can be overwhelming, and you're likely to be feeling a range of emotions, **this is normal**.

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We know many families are unsure about how and when to talk to their child about their diagnosis.

Search **'Talk to your child about their diagnosis'** at **usherkidsuk.org** for advice from parents and professionals.

Ask

Ask the geneticist to confirm the diagnosis in writing and provide you with a copy of the genetic lab results for your records. They can also share a copy with the GP, Audiologist, Sensory Support team, and any other healthcare professionals involved.

Learn

Learn more about Usher syndrome so that you can make informed decisions. Usher Kids UK has useful resources on our website, and holds regular virtual and face to face events designed to connect parents with experts.

Talk

Talk to other families of children with Usher syndrome. They will be such a valuable resource to you, as you will become to them.

Tell

Tell your teachers or key workers about the diagnosis when you feel ready. If you think it will be useful, organise a team meeting with all the professionals working with your child to discuss any changes which might be needed to current support, and any options to consider further down the line. If these professionals want more information about Usher syndrome they can access this from Usher Kids UK.

Organise

Start a folder with a section for each service you access. You can add test results and letters in date order to each section, with the most recent on top for easy access.

Question

Keep a list of questions ready for your appointments. Audiology appointments take place frequently in the early years giving lots of opportunities to discuss any questions, but ophthalmology appointments only take place once or twice a year, so it is helpful to have your questions ready. Take a notebook so that you can remember and share what you learn, and don't be afraid to ask questions – it is important you understand what is being said.

Live

Lastly, and most importantly, keep enjoying time with your child, and don't change your hopes and dreams for them.

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Support

What support is available for the family?

Parents and family members often respond to the diagnosis differently. Some are ready to take on board new information straight away, and others need more time and support. Some want to share the news with family and friends, others prefer to keep it to the immediate family until they find their feet. Go at a pace that suits you, and encourage each family member to think about what support or outlets work best for them.

Many people find that talking with another parent or family member who has experienced a diagnosis of Usher syndrome for a loved one can be really reassuring. Usher Kids UK can put you in touch with other families of children at similar or different ages, stages and types of Usher syndrome, so that you can learn from the experiences of others and build your own network of support.

Usher Kids UK works closely with other charities which serve the Usher syndrome community. CUREUsher is a UK-based charity which focuses on raising awareness and funds for research. Molly Watt Trust raises awareness of the challenges of the condition.



Your child will take their cues from you, so it is really important to look after yourself and find outlets which help as you adjust to the diagnosis. If you need support please reach out for help.

Search **'Find support'** at **usherkidsuk.org** for more information.

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What I wish I'd known at diagnosis

Often the best experts you'll ever meet are other parents who are on the same journey. Here our community shares some of the things they've learnt since diagnosis.





The most useful advice often comes from other families and those living with Usher syndrome.

Search **'Usher stories'** at **usherkidsuk.org** for more information.

As soon as I connected with another parent, it felt so different - it felt better, less scary.

It is hardest when they are little and you feel so protective. It gets easier as they grow up, and you can follow their lead about how they want to live with it.

Our daughter is in high school, and we now have to remind people that she has Usher syndrome – everyone forgets because she is just living her life.

It isn't the end of the world, it's just turned out to be a different journey than we were expecting.

It helps to stay one step ahead, finding out from other parents and experts what the options are for the next stage.

Don't focus on the negative.
Focus on what they can do.

We wish we'd known that everyone is affected differently.

My mind raced ahead, but I now wish I'd worried less about the future. By the time whatever I'd been worrying about came around, we were in a totally different place. I could have saved myself a lot of effort if I'd kept focused on the here and now!

You don't have to change your hopes for your child.

Get in touch

As a charity created by families for families, we understand the journey you are on. We exist to empower our community of children, young people and families, and the professionals working with them, by giving access to tailored information, support and regular opportunities to connect.

We'd love to hear from you – get in touch if you have questions, or want to connect.

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