Living with Usher syndrome

BY EMMA BOSWELL

"Usher people often still have sharp, clear central vision. It is the corner of their eye that is missing. This is why the Usher person often does not feel or look blind. In the early days they may not even be aware of their vision problems. They just move their eyes around more to compensate [1]. "

Usher syndrome is a combined sensorineural hearing loss with retinitis pigmentosa (RP) which is a visual impairment, so essentially people with Usher are what is known in the field as deaf-blind. Although considered a rare disease, it is a significant cause of deaf-blindness. Usher is genetic and inherited in such a way that geneticists would label it a 'recessive' condition. This means that a defective Usher gene must be inherited from each parent in order for that child to develop Usher. In other words, both parents are carriers and they have a one in four chance of passing two defective copies of the Usher gene to their child (one from each parent). Usher is a progressive condition and falls into three different group types:

Usher type 1

People who have Usher type 1 are predominantly born profoundly to severely deaf and develop problems with their vision (RP) at a young age or during their teenage years. They typically have problems with their balance due to vestibular issues inside the inner ear which affects proprioception ability. In the past people with Usher type 1 would use British sign language as their preferred method of communication however the younger generation of Usher type 1's nowadays have cochlear implants (CIs) as infants so may not use sign language at all, and have clear audible speech thanks to a mixture of speech therapy and CIs.

Usher type 2

People with Usher type 2 are typically born partially deaf / severely deaf, and develop RP later on in life, usually in their 20s-30s. As those with Usher type 2 are born with moderate hearing loss they usually develop speech on a par with their hearing peers. They do not have the same vestibular complications as those with type 1, therefore do not present with the same balance problems as those with type 1 but may experience balance problems as a result of their sight and hearing loss.

Usher type 3

People with Usher type 3 are either born hearing or partially hearing and their RP and hearing loss will develop roughly at the same time as one another. About 50% of those with the third type of Usher will also develop poor balance. The onset of hearing loss and sight loss can happen at any age. Type 3 is the rarest, making up just 2% of all cases of Usher syndrome.

Many people with Usher will also develop additional eye conditions such as cataracts, macular oedema or glaucoma. No two people with Usher are ever the same. Each person will have a varying degree of hearing or sight loss and a mixture of symptoms such as floaters, differences in peripheral and central vision, sensitivity to light, difficulties in adjusting to different light levels and the onset of night blindness, even those from the same family will still have very diverse results. Because of this and because of the combined nature of sight and hearing loss Usher is a very complex condition. It is a challenging and ongoing journey for each person; as they adjust to their diagnosis they also must learn to adjust when their sight and hearing loss deteriorates. This can lead to what *Sense* coin as life crises and they tend to happen at pivotal life stages such as diagnosis, changing or leaving school, going to university, starting employment, marriage, starting a family, losing a driving licence or losing a job.

At *Sense* we concentrate on five main areas of support in terms of helping people through each life crisis they incur.

Communication

Good communication benefits a person's wellbeing helping them psychologically to socialise and 'engage'. Many people with Usher find the communication transition required as their sight deteriorates a daunting challenge. We take a positive stance at Sense and encourage people to think about each different communication method as a new skill. Rehabilitation is an important preparation. We encourage people to try and work with what they have at any given time, for example lip-reading, residual hearing and vision, hearing aids or CIs, sign language, hands-on singing, Deaf-blind Manual, Braille, Haptics, Deaf-blind Block, reading, writing and speaking.

Mobility

We encourage people to think about mobility training, where they live and the access they have to good transport links. Individuals may decide to use a cane (red and white – this symbolises deaf-blindness), a guide dog or dual sensory guide dog, comm-guides or a PA. People with Usher find CIs useful in accessing environmental noise for example busy traffic, sirens or placement.

Access to information

People with Usher, like the general

population, vary in their educational attainment and employment status. We come across many Usher professionals such as directors, managers, psychologists, physiotherapists and business owners to name a few. An important part of education and work will involve reading information. Many Usher people will continue to read for many, many years after diagnosis, however this often depends on their RP and additional eye conditions which may affect their reading vision. They may use large print, different coloured papers, magnifying aids. We are lucky with modern technology, including iPads, apps and screen reading software. It is important that as a person's vision changes their access to information does not stop.

Mental health / wellbeing

Usher syndrome is not a 'death' sentence, however for some people with Usher they may feel their life is 'over' when they discover their diagnosis or their vision deteriorates or their life changes drastically, for example through medical retirement or redundancy. With the right support such as that offered by the Usher service we can help them to start to 'rebuild' their life. This may include a *Sense* mentor or help from the *Sense* legal team or signposting to specialist counselling services.

"Uncertainty is a theme underlying all the participants' experiences of Usher. Predictions have been made, frequently, especially at diagnosis, but in many cases, they have turned out to be inaccurate, or at least inaccurately phrased." [2]

Professional advice

We work with peers (part of our mentoring project), other organisations such as CI services, deaf charities, educational organisations, medical experts, researchers and research teams and those working in the field. *Sense* always celebrates achievements and it is important for people to know that people with Usher can have a good education, go to university, have a good job, own their own property, get married and have children. Usher will not stop them having adventures such as backpacking and travelling around the world, going sky diving, skiing or doing a bungee jump. It is important though for people with Usher to receive factual and good information to help them be better prepared and have different skills ready to control the changes that are to come. In order to adapt to meet their needs and the challenges they face, they have to first accept their Usher. Currently there is no cure despite the medical research happening around the world at present.

"Because of the combined nature of sight and hearing loss Usher is a very complex condition. It is a challenging and ongoing journey for each person."

References

1. *Usher Positive: Watching Myself Go Blind.* 2015; Usherpositive.wordpress.com
2. Ellis L, Hodges L. Life and Change with Usher: The experiences of diagnosis for people with Usher syndrome. University of Birmingham *Research Report for Sense*. Birmingham, UK; University of Birmingham School of Education; 2013.

Emma Boswell,
National Usher Co-ordinator,
Sense,
101 Pentonville Road,
London, N1 9LG, UK.

E: Emma.Boswell@sense.org.uk

Declaration of Competing Interests
None declared

ABOUT THE AUTHOR

Emma Boswell has worked for the charity *Sense* for the past eight years and her current role is National Usher Co-ordinator where she is responsible for the Usher service team. She has delivered many presentations and training sessions. She has a Deafblind Diploma and is Chair of the International Usher Network at Deafblind International. She has Usher syndrome type 1 and is a bilateral cochlear implant (CI) user. She is married with two young children. In her voluntary work, she is Chair of the Deaf Cancer Support Group.