

Factsheet – 2

ABOUT USHER SYNDROME

Usher syndrome is one of the most challenging disabilities that anyone can face. People with Usher are born deaf or hard of hearing, then gradually start to lose their sight in late childhood.

But despite the enormous challenges this brings, people with Usher can achieve a great deal. With the right support, many people can go to college, get jobs, find a partner, and enjoy their leisure, just like anyone else.

What is Usher syndrome?

Usher syndrome is a genetic condition which causes profound deafness or partial hearing from birth, and sight loss over a number of years. This sight loss often begins in late childhood and is caused by an eye condition known as Retinitis Pigmentosa (sometimes called RP). Usher is also a significant cause of deafblindness in adults.

Deafness

People who have Usher syndrome experience moderate to profound deafness.

Children born profoundly deaf have average losses of 95 db and over, which means that they cannot hear continuous speech sounds, even with a powerful hearing aid. Profoundly deaf children tend to prefer sign language as their natural language, although some do develop speech. Many sign language users feel very much part of the deaf community and culture.

Children who have partial hearing are much more likely to acquire speech and language if they use hearing aids from an early age. Partially hearing children tend to go to mainstream schools, use speech as their main means of communication, and rely heavily on hearing aids to hear conversation.

What is Retinitis Pigmentosa?

Retinitis Pigmentosa is a disease of the retina. The retina is the light sensitive tissue at the back of the eye where the first stages of 'seeing' take place. With RP the retina slowly degenerates and loses its ability to transmit pictures to the brain. The early

symptoms of RP include difficulty seeing in the dark, often coupled with problems in adapting to bright light and changing light conditions. Loss of visual fields which may give a patchy effect to side vision is also common.

Tunnel vision

In one type of retinal degeneration the cells at the edge of the retina gradually stop sending information about changes in light levels and the shape of objects on the edge of someone's vision. This loss of visual field, or 'tunnel vision' means that someone cannot see objects unless they are directly in front of him. This may mean for example, that they can still read the bus timetable, but will need help to find the bus station.

Loss of central vision

Another type of retinal degeneration causes the cone cells in the retina to be affected first so that the individual can no longer see the details of the object in front of him and will find it increasingly difficult to read print.

In cone dystrophy, low vision aids such as hand magnifiers, and magnification of print using a closed circuit television as well as large print texts may be helpful.

Low vision aids

In both these cases, it is also important to think about lighting conditions that will suit the individual, together with the use of contrasting tones to make things easier to see. Ask for an assessment at the Low Vision Aid Clinic at your local hospital.

There are many people who have retinitis pigmentosa only. This is the second major cause of blindness in people under 40.

Does Usher syndrome lead to blindness?

With both types of retinal degeneration, how much and how quickly someone's sight deteriorates will vary a great deal.

Very few people with Usher will become totally blind - that is, have no light awareness. Many will become night blind in their teens, but in daylight will keep their normal vision for reading and seeing fine detail. Loss of visual field is also common, which means someone's ability to see around them (rather than straight ahead). This causes problems for people in moving around safely, especially in unfamiliar places. Many people will also find it difficult to judge depth, or to see monotonous (such as grey, white and black), and find that they are easily dazzled by bright light.

What are the main types of Usher?

Our current understanding is that people who have Usher fall into three distinct groups known as Usher Type 1, Usher Type 2, and Usher Type 3 which has only been recognised recently. The three types have different genetic make ups and effects:

- **Usher Type 1** causes profound deafness from birth, and poor balance, which in turn can cause late development in development. Retinitis Pigmentosa may be

noticed before the age of 10. People with this type usually prefer to communicate by using sign language, and see themselves as part of the deaf community;

- **Usher Type 2** causes partial to severe hearing loss; but the organ of balance seems to be unaffected with no effect on walking development. RP may not become apparent until adolescence. People born partially hearing with Usher invariably use speech to communicate; but this will become increasingly difficult as their sight worsens.
- **Usher Type 3** is rarer and has been specifically recognised in Finland. People with Type 3 seem to have normal hearing from birth, but develop RP in adolescence or later when hearing loss also occurs. Hearing can deteriorate steadily over ten or fifteen years, and some people also experience balance problems. This group may experience great difficulty in adapting to major losses in hearing and sight.

What causes Usher syndrome?

Usher syndrome is an inherited condition which is passed through the family. It is possible to be a carrier of Usher syndrome but to be unaffected by the symptoms.

For a child to be born with Usher syndrome both parents must carry an autosomal recessive gene. Autosomal means that the gene is not sex linked and so males and females are equally affected by the condition. In recessive inheritance both parents carry the gene, but are not aware that they are carriers until the condition is diagnosed in their children. The child with Usher syndrome has two recessive genes for the trait and will transmit these genes to their offspring.

What challenges does Usher bring?

When someone finds out that they have Usher syndrome the news can be devastating. Many people do not realise they have Usher until they are teenagers and begin to have problems with their sight. They will have grown up as deaf or partially hearing, and coming to terms with losing their sight as well can be very hard.

As their field of vision shrinks, people with Usher often feel very isolated, not just from the hearing sighted world, but also from other deaf people who may have made up many of their friends and acquaintances. They will also often experience communication problems as it becomes increasingly difficult for them to see sign language, or to lipread. Some may have poor balance which can severely affect their freedom to move around safely.

For most people their teenage years can be a very exciting but also very challenging time. So for those young people who have learned to live with deafness, but then discover that they are losing their sight as well this can be a very tough time. Young adulthood is the time to be out and about, enjoying leisure activities, socialising with friends, and experimenting with relationships. The young person who also has RP will find that nightblindness places severe limitations on their social life, on opportunities for meeting people and on developing friendships

Emotional reactions

Most people will be very shocked and upset when they are diagnosed as having Usher syndrome. They will often feel very frightened and may become extremely depressed about the future. They may also feel very angry and ask, why me? Most will benefit from ongoing psychological counselling as they attempt to adjust to their decreasing vision and changed life circumstances. Centres where Usher syndrome is understood, and where they can meet with others similarly affected to talk through their difficulties are also important. See the end of this factsheet for the support that Sense offers.

Reactions of parents and friends

Intense emotional shock is the inevitable and understandable reaction of parents to the discovery that their deaf child also has Retinitis Pigmentosa, and it may take a long time for them to really accept what is happening. Many parents experience strong feelings, including grief, depression, guilt, fear, helplessness and anger

A prolonged period of mourning often follows before family and friends are able to adjust to the reality of the situation and start to come to terms with it. Many parents find it very helpful to meet up with parents in a similar position - see the back of this factsheet for information about support from Sense.

Learning new skills

With the right training and support, people with Usher Syndrome can be taught to minimise the effects of their new disability. And they can learn new skills which will help them to cope with life when their sight gets worse. As someone's vision deteriorates, they will need help to develop their mobility and daily living skills such as cooking. They may learn braille, even if it is only for labelling and learn finger spelling.

Many people born deaf with Usher will have learned British Sign Language to communicate. As their sight narrows they will still be able to use and understand sign, provided people sign to them within their remaining visual field. When this remaining vision fails people can use tactile or 'hands on' signing by placing their hands over the hands of the signer so that they feel the signs being used. People born partially hearing invariably use speech to communicate, although some may learn sign.

Education and work

By their twenties, most deaf people will be in training for employment, or already in work. Those who have RP may well be doing work which will eventually prove difficult or even dangerous to carry out.

The rate at which someone's vision declines in their twenties is hard to predict, and so it may not be easy to decide when to change course. Should the keen young motor mechanic and student nurse carry on, and have perhaps 10 to 15 years of work, or should they change to something that they can work at even when their vision has become severely restricted?

It is important that the young person with Usher receives career guidance that is realistic but not too restrictive. To an employer, labels such as 'deafblind' will mean 'unable to hear or see anything' although this is rarely the case, particularly with young adults. What needs to be answered is 'can the employee, given the restricted vision and hearing he has, do the job? The employer should know that certain aids such as closed circuit televisions can be loaned through the Placement Advisory Teams to help the employee.

Obviously certain jobs involving the use of dangerous machinery should be avoided, and early diagnosis will help prevent these choices.

Successful Usher people

Despite the enormous challenges that Usher syndrome brings, it is important to emphasise that many people with Usher have come to terms with their condition and go on to lead fulfilling independent lives. People with Usher can and do have relationships, get married, have children, go to university, travel, go to work and enjoy numerous leisure pursuits.

How Sense helps

Sense Usher Services helps people with Usher syndrome to live as full a life as possible. It also supports their families, carers and the professionals who work with them. Our services include:

Providing advice and support to people with Usher and their families - particularly when the condition is first diagnosed.

Training education, health and social services professionals to understand Usher syndrome and its effects.

Offering information to people with Usher syndrome, family and professionals about practical ways to get the most out of life.

Running Ashley House. This is a small family house with low cost adaptations for people with a dual sensory impairment, and is available for, Insight Days, short courses, group meetings and assessments.

Other sources of help:

Usher Services

Sense South East Regional Office
Newplan House, 41, East Street
Epsom, KT17 1BL
Tel: 0845 127 0076
Text: 0845 127 0078

Deafblind UK

National Centre for Deafblindness
John and Lucille van Geest Place
Cygnet Road, Hampton
Peterborough, Cambridgeshire, PE7 8FD

Telephone: 01733 358 100

Textphone: 01733 358 100

Fax: 01733 358 356

24 Hour helpline: 0800 132 320

British Retinitis Pigmentosa Society

PO Box 350
Buckingham, MK18 1GZ
Tel: 01280 821334
Fax: 01280 815900
Helpline: 01280 860363

Where can I go for help?

If you:

- would like to find out more about deafblindness or the services for deafblind people in your area
- require information in alternative formats including braille, large print, audio or disk - or would like this factsheet to be translated into your first language - please contact Sense's Information Team.

Telephone: 0845 127 0060

Textphone: 0845 127 0062

Fax: 0845 127 0061

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