The experiences of diagnosis for people with Usher syndrome

BSL VERSION TO FOLLOW SHORTLY

Usher Project Update

It's been a year and a bit since we started working on the Usher project, trying to gain an insight into the lives of people with Usher syndrome.

In that time we have interviewed 35 people with Usher syndrome. Ten people had type 1 Usher, 20 people had type 2 Usher, and 5 people had type 3 Usher. Twenty one of the people were women and 14 were men. Their ages ranged from 14 to 56 years old, with 11 people in the 14-20 year old age group, 11 people in the 21-35 age group, and 13 people in the 36-56 age group. We have spoken to people from Scotland to Dorset, Northern Ireland to Lincolnshire. They are people who use BSL and people who have used spoken English. People have also used Braille, large print, standard print, and audio formats to read printed things. The names that we have used here are not the people's real names.
Thanks to all those who have talked to us, we are very grateful. It couldn't be done without your willingness to share.

We are pleased that we have now reached almost our target of 40 people for our interviews (some more interviews are arranged). We are looking for one more, probably a young person (14-19). Our contact details are **usher.project@contacts.bham.ac.uk (mailto:usher.project@contacts.bham.ac.uk)** – if you are under 18 years old please get a parent or carer to contact us. While we'd like to interview many more we don't have time or money to do so, as analysing the work we've already recorded takes time.

This is an update on the kind of things that people with Usher have been telling us.

Change

Most people talk to us about CHANGE; the change that Usher brings.
For some change is all in the future. Some young people (14-20) we have interviewed do not feel that their lives have changed very much. Also they don't think that their communication or their social activities will be very different in ten years from where they are now. Other young people understand that things might change in the future but it is difficult for them to imagine what these changes maybe, and anyway they will try to do the same things that they have always done, even if things have to be adapted slightly. James who is a teenager said,

> **Cos if my eyes do get worse I'll still play football. I'd play blind ball instead… So that's alright. It's not that much difference.**

Some others in this age group are taking much bigger changes in their stride already such as learning Braille, becoming long cane users, or getting a guide dog. In reply to the question of whether she is comfortable using her long cane Britney (also a teenager) replied,

> **Well before I wasn't. I didn't like people looking at me. But the teacher said to try. And I tried. And I did I got used to it. The teacher and an interpreter came to my home, and I was a bit fearful at first, I didn't have any confidence….. but as time went I got comfortable with it. (BSL interview; quotation is interpreter)**

As with the younger age group, change was important to the lives of people in the two older age groups. For some people change seems all in the future, but others are living with change on a daily basis. One of the biggest changes was around having to give up driving and the lack of freedom and independence that followed. They may have had to rely more on their partner/family to get out and about. Olivia, who is in the oldest group (36-56) felt that her daughter missed out because she was not able to drive. In addition, Olivia explained,

> **I wish that I could see my friends at any time. Send a text. Pop over there and chat. I can't keep on relying on my husband to take me here, there, and everywhere. I miss that. I used to drive. But the law changed and I decided then to give up. It was thinking about other people as well. The one thing that I would like to do is be able to drive and be independent. (BSL interview; quotation is interpreter)**

Diagnosis

For some people this change began when they were diagnosed as having Usher syndrome. People in the two older age groups could remember being told that they had Usher. Most of the young people we spoke to had been diagnosed at about 10-12 years although some were diagnosed at a very young age.

Often they couldn't remember being told that they had Usher, and having Usher is just something that is part of who they are. A mother of two children with Usher (both teenagers) remarked,

> **I think because they've been diagnosed early, so young, we just took it from that, and carried on as normal.**

The older people with type 1 Usher tended to have been diagnosed with Usher when they were still at school. Most of those with type 2 and 3 Usher were diagnosed in their twenties. One person however was diagnosed in his mid-40s. The parents of one person in her early adulthood had kept her Usher a secret from her, and it was only in her late teens when her friends told her, that she found out that she had Usher.

Often people felt that they were told about Usher very harshly and without the chance to ask any questions. Some people still felt very angry and upset about the way they had been treated by doctors, even twenty years later. Alice (age group 35-56) told us about the shock of being diagnosed with Usher,

> **I was just going for a normal, routine, eye test, check up, and as I said the optician asked if I'd damaged my eye. Referred me on to the clinic. And they tested my eyes, and then next thing they told me was 'sorry you're going blind.' I wasn't expecting that at all. And I think it was the shock. I could see their mouth going but it wasn't sinking in. I came home and I was in bits. I guess that's normal I suppose. I was in bits. I couldn't even tell my mum what it was. She had to then ring the clinic to find out. It was just the way they'd done it, sorry you're going blind. And that was it.**

Understandably being diagnosed with Usher syndrome had a strong impact on the lives of many people with Usher. Shock, anger, denial, and depression were common responses. It was not uncommon for a short course of anti-depressant medication to be prescribed by a GP. Nevertheless most people we spoke to over time have adapted to *being people with Usher*. Vicky, (age group 35-56) who has type 2 Usher, tells us of her experiences just after her diagnosis,

> **That was very difficult, because you know, I kind of got round to accepting I had a hearing problem, then to be told this as well. Oh how can I say it… I suppose this is the reason that precipitated me having a nervous breakdown. There were other reasons as well, but it probably contributed to it... And I think that I'm gradually coming to accept it, but at times I haven't wanted to. Then something might happen that reminds me that I do have a problem. And you know it is hard…**

How far and fast vision loss came does not always match the expectations of the types of Usher (where Usher 2 is expected to affect vision in the mid-teens to early twenties for example). Those young people coping with the most vision difficulties had Usher 2 and Usher 3. Many of those we interviewed were using vision to undertake most tasks, including those in the older adult age group (36-56).

Some people were very worried about the changes that Usher might bring to their life and had decided to actively not think about change, as it worried them too much. Some had taken up yoga and meditation in order to help them to focus on the present. Some people were very keen for new cures and medical developments for Usher, in

particular RP, and were constantly on the look out for new treatments. Dave, (age group 36-56) , said he was always on the net, researching new drugs and gene. Pointing to his friend's 3 year old he said he was sure there would be a cure, in her lifetime at least.

On the other hand people like Rob who is a young adult, saw his Usher, and in particular the changes in his vision, as part of his identity and would not change it,

> **I wouldn't change my sight now. I know that they've brought out the retina chip, and the gene therapy, if they offered me it, I'd say no. It's made me who I am today. It's made me a lot stronger person now, than I was before. I think I do marvellous. And I wouldn't change the way it is.**

Change is a very important feature in the lives of people with Usher. We have seen a full range of responses to Usher. Some people were not thinking about the future. Others were determined that Usher would not affect their life, whatever the future would bring. Some were worrying about the future. Some were coping with feelings which were overwhelming, and causing depression or anxiety. Finally for some people with Usher it's just business as usual.

This is a short introduction to the work we are doing. We will publish more on particular issues in the future. We are continuing to collect information and to analyse the information we have.

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