Annmaree Watharow,
Emily Shepard, and
Emma Boswell

USHER SYNDROME

About Us, by Us

Disability Studies

Collection Editor
DAMIAN MELLIFONT

LIVED PLACES
PUBLISHING

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Abstract

Few books for health and social care students, professionals and practitioners are written by the very people impacted by a condition. Usher syndrome is a diverse collection of types, subtypes and complexities. It has been called 'a life of adjustments', and this book unpacks these and explains how you can help support and communicate with us and others who live with Usher syndrome. The three authors all have lived experience, expertise and knowledge of life with Usher syndrome for individuals, parents, families, partners, siblings and supporters. Lived experiences inform and imbue each chapter. The lived expertise that comes from being clinicians, colleagues, researchers, peer supports, advocates, parents of and parents with Usher syndrome directs content to reflect what the community wants you, the reader, to know about Usher: the basics, the genetics, diagnoses, diversity, extended family impacts, life-long adjustments, downsides and upsides. We give the reader blueprints for better communication, better health and well-being and solutions for every day. Usher syndrome is complex and challenging, but we are capable contributors when well supported. This book is strong evidence of this.

Keywords: Usher syndrome, deafblindness, dual sensory impairment, communication, disability, life adjustment, Charles Bonnet syndrome

Contents

Learning objectives

What we three authors want all people, including health and social care students and professionals particularly, to do for us is:

Ask:

- Your patients, clients, constituents and customers what they need
- Your patients, clients, constituents and customers what their goals are
- Your patients, clients, constituents and customers how can you help
- Your patients, clients, customers and constituents what they need for good communication, and then provide this
- The carers of your patients, clients, constituents and customers what they need
- How can I work collaboratively with other health and social care providers to provide the best care for those living with Usher and their families/carers?

Identify:

- Patients, clients, constituents and customers who are experiencing the complexities of living with Usher syndrome and ask about their needs
- People with the combination of hearing loss and low vision who may be seen in the course of your work and will have diverse communication needs
- Some communication solutions for an individual
- When to pivot to another communication solution

- The residual sense/s an individual has at any stage of their Usher syndrome journey and creative ways of working with these
- The needs of the families and carers who may need your support

Provide:

- Kindness
- Support
- Understanding
- Care and communication
- Advice on minimising health and well-being risks
- Peer support
- Accessible formats
- Devices
- Aids
- Specific methods of communication for an individual at different stages
- The right interpreter every time one is needed or wanted, and work effectively with them
- Care, support and education for families and carers too

Practice:

- Being part of a person with Usher syndrome's super team of supports: collaborative care wins
- Being inclusive
- 'Walking the talk' with accessibility
- Upskilling: learning more
- Learning touch messaging and incorporating into your practice

- Asking about your patients, clients, constituents or customers' goals and providing support to achieve those goals
- Including carers and families in care and communication
- Building an awareness of what it is like to be in the shoes of a person with a dual sensory impairment and then put that awareness into practice
- Exhibiting kindness, patience and when appropriate, humour!

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About us, by us

Introduction

Annmaree

In the late 1960s, Myra and Patrick Watharow, my sighted-hearing parents, had five children, two with severe hearing loss, myself and my youngest brother. My hearing loss was long suspected. There's a family story that, while sitting up at 6 months old, someone dropped a pile of plates they were carrying on a concrete floor, and I didn't respond. It would be four more years of poor speech development, 'disobedience' and 'daydreaming' before an ear, nose and throat surgeon casually says to my parents, 'By the way, your kid is really deaf.' Shortly after, my 18-month-old brother was diagnosed with deafness too.

Our diagnoses were followed by ugly hearing aids (large boxes worn in pockets on the chest, cords that snaked from them to hard, painful moulds in the ear).

Figure 1 shows a drawing of a Calaid hearing aid from the 1960s, showing hard earpieces connected by long loops of plastic-coated wires to a metal box that is the audio processing component.

These were the first 'in-the-ear' hearing aids developed in Australia in the 1960s. They were so awful (not only hurting my

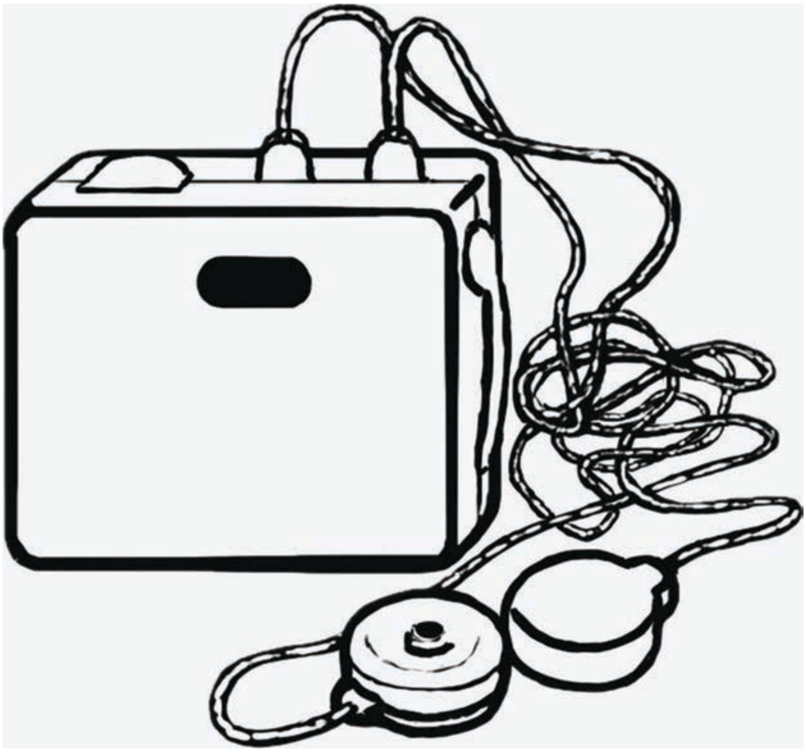

Figure 1: Calaid hearing aid

ears with pressure sores from rigid moulds, but the kids at school also viciously made fun of me, calling me 'an alien'). I used to take them off in the girls' bathroom at school before class and then put them back on after getting off the school bus in the afternoons. There began months, years, decades of speech therapy. My brother also began the hearing aid and speech therapy odyssey. No one ever told my parents that hearing loss could be associated with 'other things', and no one ever looked for them. No one made the association between the visual changes that I had begun noticing at 15 years of age (and present well before then, but assumed to be clumsiness and carelessness) with the hearing loss I already had. Not one of the professionals consulted, including ophthalmologists, general practitioners, ear, nose and throat surgeons, audiologists and speech therapists. I saw three ophthalmologists in 1985, trying to get a name for and help for distressing visual problems: falling into fountains, from jetties and off boats; difficulties playing tennis and softball where the objective was to actually SEE the ball and then hit the ball with racquet or bat; and the extreme difficulties at night-time seeing and avoiding obstacles and keeping safe. I was told to eat more carrots and 'see a physiotherapist for clumsy people'.

As a final year medical student (the first deaf student at the University of Sydney and, as it would turn out, the first deafblind medical student there – the second in Australia), I finally found someone (my friend's new boyfriend, an ophthalmology trainee) who listened to the story of deafness, and the 'weird not seeing things' symptoms. He looked at the eyes and put it all together. The diagnosis now became Usher syndrome, and it explained an awful lot.

I was sent to a Professor at the Sydney Eye Hospital who confirmed the diagnosis. He was quite cheerful when he asked me if I would come to the registrar examinations the next week just after he had said, 'You will go blind soon.' And I realised that would make me 'deafblind' too. I didn't get any more information, any referrals, any empathy.

Being a medical student, I went to the medical librarian and asked for a good article on Usher syndrome. She ordered a copy of Vernon's 1969 article 'Usher's syndrome—deafness and progressive blindness: Clinical cases, prevention, theory and literature survey.' Reading this paper was visceral. It made me run to the bathroom and throw up. It principally described the syndrome as one of incapacity, psychosis, institutionalisation and feeble-mindedness. I thought this was my future. This book (and most more recent research) tells a very different truth of capabilities and capacity.

Emma

I was born in the 1970s to hearing parents along with an older brother, all of whom had normal vision. My sister Claire and I were diagnosed with Usher syndrome, but our mother kept this information from us until we were older, following medical advice at the time. We attended Mill Hall School for the Deaf as weekly boarders. Growing up, I identified as Deaf, unaware of my vision issues until around age 10, when I started experiencing night blindness and balance difficulties.

My vision problems became slightly challenging during secondary school at Mary Hare. I often struggled with low light conditions, had difficulty navigating safely at night and experienced accidents like being hit unexpectedly by a football or injuring myself due to limited peripheral vision. Classroom learning was not easy because the teaching was oral, with no signing allowed. Around age 14, I began noticing visual halos around my teachers, later understanding this was related to Usher.

Despite challenges, I enjoyed sports like volleyball, swimming, skiing, cross country and scuba diving. These were more difficult due to my balance, but determination and patience helped me succeed. During my gap year, my sister and I spontaneously travelled to Paris, and later I backpacked across Europe, Kenya, and joined Operation Raleigh (a youth expedition organisation based in the UK, where volunteers participate in sustainable development, environmental protection and community resilience projects in rural and remote areas of the world) in Malaysia. Travelling required careful planning to avoid difficulties related to night blindness. A challenging but rewarding experience was climbing Mount Kinabalu, supported closely by peers.

In 1992, prompted by a scuba diving instructor’s request, my mother finally told me about my Usher diagnosis. I felt relieved knowing why I’d faced certain struggles. Understanding my condition allowed me to embrace it openly.

In adulthood, I pursued higher education in Art and Interior Design. During college, poor classroom lighting complicated my note-taking process, highlighting ongoing barriers related to Usher. I graduated and worked successfully as an interior designer.

Later, I transitioned into working directly with people living with Usher syndrome, initially volunteering and then becoming manager of an Usher service charity for 20 years. I found fulfillment empowering others with Usher Syndrome through mentoring, training and advocacy. I also married Clive, had two children, Lucybelle and Thomas, and navigated motherhood while managing my sensory impairments. Parenting brought unique challenges, from navigating busy environments to ensuring my children’s needs were met without overly relying on their support.

Over the years, I’ve continued to contribute actively to the Usher community, speaking at international conferences, training globally and serving as a UK ambassador and chair for international networks.

In 2020, cataract surgery temporarily improved my sight before ongoing deterioration significantly impacted my reading vision. I've resumed learning Braille, adapting to new challenges while maintaining independence. In 2023, I lost my reading vision, and in September 2025, my clinician confirmed the loss of my central vision. Despite the changes, I remain passionate about family, travel and advocacy. My involvement in sharing my story aims to educate and foster understanding, empathy and empowerment within and beyond the deafblind and Usher communities.

Emily

My son Louis was born in Melbourne in January 2010 in the middle of a heat wave. We spent the night before sleeping on the floor at my sister's place under her air conditioner as a power outage in our neighbourhood pushed the mercury at our house to an unbearable level. I slept soundly with my husband David and my 3-year-old daughter Frankie nestled in beside me. It would be the last good sleep I would have for many, many years, as his arrival was about to turn our world upside down.

The private hospital where he was born was one of the last hospitals in the country to roll out the infant hearing screening programme (which is ironic now that I am a member of the Australasian Newborn Hearing Screening Committee). We went home without the knowledge that our precious newborn was profoundly deaf, but knew something wasn't quite right. In the weeks and months that followed, we became consumed by appointments, referrals, tiny little hearing aids, early intervention, therapy and investigations. But still, I felt there was more to learn about our boy. I could see his floppy head and his frustrations were a worry to our early intervention specialist. She had seen hundreds of babies with hearing loss and commented 'apart from language and communication, all other milestones should tick along as normal'. But they weren't. Rolling, sitting, crawling, walking. They weren't ticking along at all. We had an unhappy baby who seemed frustrated with his body.

I remember googling 'deafness and balance issues in babies' when my son was less than 1-year-old. I mentioned 'Usher syndrome' to our ophthalmologist (it was recommended that all infants diagnosed with hearing loss undergo regular eye exams to check for associated syndromes) and her response startled me. 'It could well be years before we can rule something like that out.' She then continued with some reassuring comments about the rarity of such a condition and the unlikelihood blah blah. But I wasn't convinced. If my son was going to lose his vision, I wanted to know. I wanted to be prepared. I couldn't live with the anxiety of presenting for an eye exam each year and stressing about what she might find. Surely there was a way of finding out?

We were referred to a genetic clinic at one of the major hospitals in Melbourne, and a blood test and several thousands of dollars later, our son's genetic material was on the way to the University of Iowa for a genetic test for conditions known to be associated with hearing loss.

When we returned to the clinic 9 months later, I was struck by the geneticist's hands that lay clenched together over a manilla folder. I could sense by her hands that she knew what she was going to tell us was about to derail our family yet again. But even though the diagnosis of Usher syndrome felt like the worst thing imaginable at the time, I still felt a sense of relief knowing what it was. I now knew what we were up against, and I was convinced we would find knowledgeable clinicians and other families who had either been where we now stood, or who were right there in the thick of it. But that didn't exist in Australia.

This was new territory for parents and children – having to navigate hearing loss coupled with the knowledge that vision loss would begin sometime too soon. We didn't yet qualify for vision services ('come back when he loses his vision' was the message we received) and didn't quite know what to do or where to go for support. Faced with the gaps in knowledge and support, together with a fellow parent, Hollie Feller, we founded UsherKids Australia. The purpose of the organisation is to help children and families living with Usher syndrome to 'love their lives'. We had learnt to navigate the world of deafness and now we had to navigate this new world of blindness, and we were just starting from scratch again. Doctors can give you medical advice, but another family going through exactly what you are going through can give you that lived experience. That is just gold. This is why I am writing about Usher syndrome with Annmaree and Emma. To share my lived experiences of being a parent, a CEO (Chief Executive Officer) of a parent-led organisation (UsherKids Australia) in union with current research and professional knowledge so that you, the reader, are enriched by all of these perspectives.

Usher syndrome

Usher syndrome is the most common form of deafblindness (combined hearing and vision loss) in people under 65 years. Usher syndrome accounts for half of the deafblindness in this group (Dammeyer, 2014).

Usher syndrome is usually deafness or hearing loss from birth in combination with a later onset, degenerative vision loss. While this vision loss is the result of an inherited condition called retinitis pigmentosa, it usually begins in the first or second decade of life. There are several types and subtypes. One, Usher type 3 is the exception of the hearing loss at birth, with the deafness beginning in childhood. Sometimes, the syndrome is accompanied by vestibular (balance) dysfunction. Sometimes, hearing further degenerates too. At present, despite much work, there is no treatment and no cure. We look more at Usher syndrome, data, current research and the possibility of treatment or cure in Chapter 2.

Albrecht von Graefe (1828–1870), a German ophthalmologist, considered the founder of modern ophthalmology, first noticed that hearing loss and eye disease sometimes co-existed in one family. He studied three brothers who each had perceptual hearing loss and retinal disease and wrote these familial cases up in the ophthalmology journal he had founded, *Graefe's Archive for Clinical and Experimental Ophthalmology*, a journal that still exists (Grzybowski et al., 2024). But the finding that deafness and retinitis pigmentosa was an inherited condition with an autosomal recessive pattern (a particular type of genetic inheritance) came from a Scottish ophthalmologist, who studied the extensive 'pedigrees' (or genealogies detailing family medical history) of 40 families with retinitis pigmentosa. Charles Howard Usher published an article, 'On the Inheritance of Retinitis Pigmentosa with Notes of Cases' in 1914 (Royal London Ophthalmic Hospital Report, pp. 130–236).

This article noted that hearing loss was present in many (44%) of the cases he studied in family pedigrees of those with retinitis pigmentosa (Royal London Ophthalmic Hospital Report, p. 169). So, Charles Usher was the first to note that the combination of hearing loss from birth and retinitis pigmentosa existed in family groups, meaning that it is an inherited condition. This discovery was an astonishing feat of research before telephones, faxes or the internet were used to facilitate the tracking down of family members of 69 individuals.

Usher syndrome in the past has also been known by other names: in the 1940s as dystrophia retinae-dysacusis syndrome; in the 1950s as Alström-Hallgren syndrome or even more simply as Hallgren syndrome. Allström and Hallgren were two researchers who studied primarily in psychiatric institutions (Hallgren, 1958). From the 1960s onward, Usher's syndrome is the common term, with our present-day usage of Usher syndrome, as used in this book, the norm. The work of Charles Usher cemented the inherited connection between hearing loss and retinitis pigmentosa with later work recognising variations or distinct types and subtypes along with vestibular dysfunction in some.

The knowledge gaps

The knowledge we have about Usher syndrome is not as comprehensive as we would like due to:

1. Our relatively recent understanding of its inherited nature and biogenetics.
2. It being proportionally rare.
3. There being a geographical wide spread of cases.
4. Older academic articles having been based on populations residing in psychiatric or long-term care facilities (see Vernon, 1968, or Hallgren, 1958).
5. An absence or low value on lived experience knowledge in research, although this is changing with the rise in co-design and more participatory research methods.
6. The dependence, until recently, on clinical assessment as the means of diagnosis. This has meant diagnosis largely is not given until noticeable reduction in vision occurs. Genetic testing advances – and affordability – have resulted in early diagnosis of babies with hearing loss (even *in utero*). This has created new complexities for families and children with Usher syndrome.

Why now?

In late 2024, the Dual Sensory Impairment Project at the University of Sydney conducted a study to determine what people with dual sensory impairment or deafblindness wanted to know themselves and what they wanted professionals to know about them. When we look at the subgroup of people with Usher syndrome, we found many things that reinforced the need for this book.

The Usher syndrome subgroup also had other family members with Usher syndrome (14%). Some worked as professionals providing services to others with Usher syndrome (7%) and some worked for an organisation that provides services to people with disability. As you will have observed from the three of us co-authors – Annmaree, Emily and Emma – is that people with Usher syndrome can occupy multiple social roles and positions in society. We are not simply passive recipients of social care. We do, can and should be supported as needed to contribute to society in every domain and manner. We can be peer supports, knowledge builders, advocates, executives, leaders and more. We can and do and will continue to contribute.

Common themes expressed in the survey by those living with Usher syndrome and their families included:

1. The absence of information at diagnosis from healthcare professionals: When asked, 'Did you have access to good information?' Seventy-five per cent said 'No'. A common related theme was the lack of dual sensory awareness and expertise among healthcare professionals and practitioners, single-sense service professionals and organisations. When we looked at the sections on solutions, all participants living with deafblindness, including those identifying with Usher syndrome, want health and social care professionals and practitioners to have targeted communication skills training and to be aware it is their responsibility to ensure effective information exchanges. This book is then a first step towards this goal.
2. Everyone wanted information that incorporated advice and tips from people with lived experience, and those with lived knowledge of supporting someone with Usher syndrome, especially on how to help ourselves live better (adjusted) lives.

One respondent said:

> Meeting others and realising you are not alone is important; also knowing life is for living and adapting to a constantly changing condition.

Another survey respondent had this to say:

> Usher syndrome is about grief, grief, grief. I have found taking up dancing and dancing lessons has made an enormous difference to my psychological wellbeing. Also, art therapy has been helpful. It's about adjusting what you're interested in and what you can do, to come up with something that keeps you occupied and amused.

Why us?

In the 1970s, Diana Peter had two daughters with hearing loss: Claire the older, and Emma the younger (who is co-author of this book). Diana Peter wrote a children's book about her girls' lives titled *Claire and Emma* (Peter, 1976). This photo-picture book is pitched at younger children to explain the wearing of hearing aids, lip-reading and the need for extra lessons, patience and time while still having a good life – tree housing with cats, for instance. She concludes her book with these sentences: 'Claire and Emma like doing just the same things as other children' and 'They like people to be friendly and talk with them, so that they don't feel left out.' And isn't that what we all want, not to be left out. Our book is a step in that direction.

We are writing this book for you, so you will understand more about Usher syndrome. We are also writing about us, our lives with Usher syndrome, family with Usher syndrome, working with Usher syndrome and finding the best life with Usher syndrome. This book is not about us being subject matter that are being observed by professionals, who then conduct research, make pronouncements and devise practices, although some of us with Usher syndrome do this as part of our work and advocacy. We are all living an Usher syndrome life. We work in different ways to make a difference so that everyone with Usher syndrome or everyone connected to someone who lives with Usher syndrome can live a good life with access to good information, communication and support.

This book is written and informed by those with Usher syndrome, parents of those with Usher syndrome or interpreters, siblings, partners and family members of those living with Usher syndrome. This book recognises the critical importance of lived experience and taps into this wealth of expert knowledge to bring the living realities of Usher syndrome to you within its pages. And not just information, but with practical ways of how you can help support us with better communication, information, strategies and practices.

As stated, all three authors work with adults and/or children with Usher syndrome and write about life with Usher syndrome and its complexities, frustrations and joys. Yes, joys. Rich lives and happiness are not excluded by Usher syndrome, but the social structures around us all can exclude, shame and even harm people with Usher syndrome. Many researchers, policymakers and support organisations deem those with Usher syndrome a 'vulnerable' group. But we are not uniformly and constantly in a state of vulnerability. We are enormously capable, as evidenced by the body of work the three of us have amassed thus far. So, this book is about Usher syndrome, by us. By way of an introduction, we want to explain key concepts that anchor this book.