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

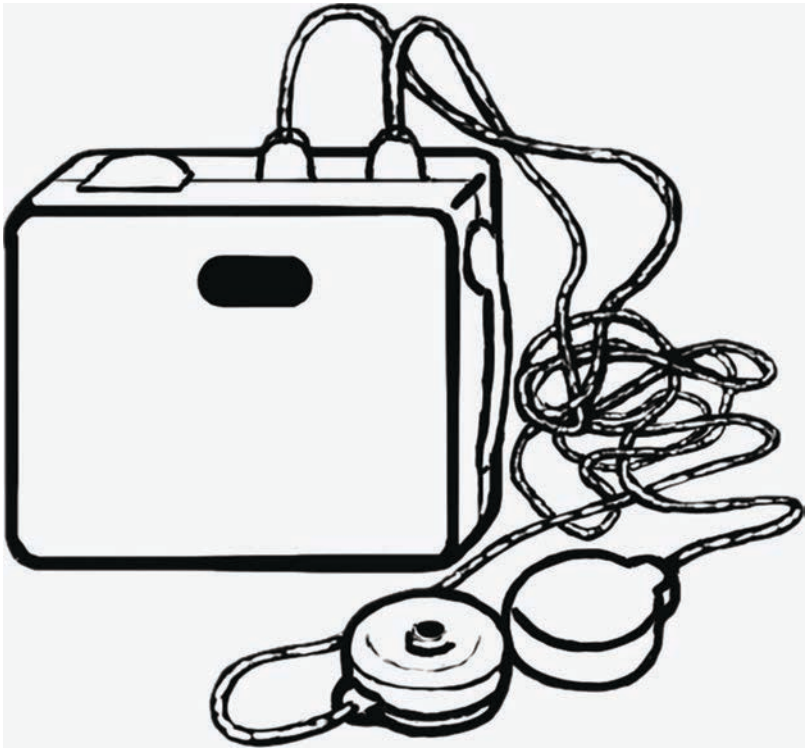


Figure 1: Calaid hearing aid

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.

Deafblindness-dual sensory impairment

The lived experience of Usher syndrome is one of complexities. It would be better described as living experience, as Usher syndrome is an ongoing series of changes and adjustments throughout the life stages. It is not a static, same or stable condition. Usher syndrome is, in fact, a spectrum of residuals, languages, resources, capabilities and diverse impacts. For many, living with Usher syndrome is a slow journey to deafblindness where both senses can no longer compensate for the losses. For others, it's more rapid.

In order to begin to understand what it is like living with Usher syndrome, we need to examine deafblindness or dual sensory impairment to give two common terminologies. For this book, we use the Nordic definition as revised in 2024. It's a detailed, clear and useful definition:

> Deafblindness is a combined vision and hearing impairment of such severity that it is hard for the impaired senses to compensate for each other. Thus, deafblindness is a distinct disability. (Nordic Welfare Centre, 2024)

But the definition itself doesn't capture the complexities, and the principal reason this definition is a good one is that it comes with a set of explanatory notes:

Main implications:

- To varying degrees, deafblindness limits activities and restricts full participation in society. It affects communication, access to information, orientation and the ability to move around freely and safely.

- To help compensate for the combined vision and hearing impairment, the tactile sense becomes especially important.
- There is a high risk that the physical and psychological health as well as social life will be affected. (Nordic Welfare Centre, 2024)
- See Chapter 2 for the full set of comments.

Identity

There are contentions around definitions and terminologies in the group of people with combined hearing and vision loss such that neither sense can compensate for the other. These are closely linked with the identity that some choose. They complicate research as academics and scientists value consistent terminologies. However, the way we use language can be an act of power and of both inclusion and exclusion. We have chosen to use a hyphenated entity: deafblindness-dual sensory impairment to describe the combined hearing and vision loss of Usher syndrome (see Wittich et al., 2013, for a longer discussion on these tensions and a viewpoint that a universal single term 'deafblindness' is preferred in research).

The development of identity is a very personal one. Some people will identify as having Usher syndrome, some as deafblind, some as neither and many as something else. What is important is that you ASK (Acquire Specific Knowledge) each person how they would like to be known. Are they a disabled person, or a person with disability or a person who lives with disability? Recent work by Sharif et al. suggests that people with disability themselves are divided in the language they use for themselves – 49% prefer identity-first language: a disabled person; but 33% prefer person-first language: person with disability; and 18% are happy with either (Sharif

et al., 2022). In this book, we use person with disability, for example, person with deafblindness or person with Usher syndrome. We use identity-first language wherever we know a person prefers this.

For example, Annmaree prefers identity-first, as her deafblindness (as a result of Usher syndrome) is both integral and entwined in everything she does. Annmaree says, 'I once believed the only time I wasn't a deafblind person was when I was asleep. But one night, the fire alarm went off, and I didn't hear it. That made me realise I AM deafblind even when I am asleep. The idea of an identity where I am a person living with deafblindness or living with Usher syndrome also doesn't sit comfortably with me. It makes the deafblind part sound like a flatmate that I can shut the door on and be quite separate from, even while in the same home. Because the deafness and blindness are ever present and interacting in quite difficult ways, sometimes with activities of daily living, social life, and my work (and sleep as it turns out, too).'

There are also exceptions to seeing identity as impairment or loss related. Many people identify as Deaf or Deafblind but not as hearing impaired or socially disadvantaged. Being a part of the Deaf and/or Deafblind culture is a positive and affirming identity for some with Usher syndrome. There is a distinct difference between lowercase and capitalised 'deafblindness' (d/Deafblindness), in that Deafblindness refers explicitly to people who identify as part an emergent culturally and linguistically diverse (CALD) group (Roy, 2019). Sometimes, this has been referred to as a 'minority within a minority', denoting how Deaf communities may have historically marginalised people living with Deafblindness.

A brief note on Deafblind culture

Deafblind communities are an emerging CALD group locally and internationally. Features of Deafblind culture include:

- A central affirming identity as Deafblind
- Tactile language, norms and conventions
- Vibrotactile conventions such as banging on a table to attract attention or emphasise a point and stamping the feet on the floor to applaud or agree with an important point
- Unique social structure, including combinations of assistance animals, support workers, communication guides and interpreters

See Clark (2014), Shariff (2015), Roy (2019) for more. Shariff (2015) explores the idea of a critical theory of Deafblindness that runs counter to other theories of disability, including DeafCrit.

Model of disability

Underpinning this book is a social relational model of disability. This takes the social model of disability in which we are disabled by society and its barriers to inclusion and in which society is held responsible for providing supports, policies and barrier removal. A more relational approach says there are impairment effects to consider beyond the social structures (Thomas 2004, Shakespeare & Watson, 2011). Tom Shakespeare put it in strong, helpful terms when he noted that we are disabled not only by society but by our minds and bodies as well. (2014, 2018). There are even 'insurmountable realities' (Vehmas & Watson, 2014), which means that sometimes, for some people with some disabilities, no amount of social support and barrier removal will

eliminate the consequences of impairments. We like to illustrate that by talking about movie-going: no amount of transcription, interpretation, or audio description can give people with Usher syndrome a similar experience to someone with hearing and sight. And that's okay, mostly. What is important to acknowledge is that the social model on its own is not enough to explain the Usher syndrome experience. Usher syndrome, for most, is a march of worsening impairments; sometimes, you haven't quite adjusted to the last decline in vision before the next one is upon you. Everyone's journey is different but there are some constants, some commonalities and some conundrums.

Taking a social relational view means we see disability as a 'complex interaction between biological, psychological, cultural and sociopolitical interactions' (Shakespeare, 2014, pp. 25–26).

How to use this book

We know many will dip in and out, so each chapter can stand alone. It helps to read Chapter 2 on the data, the types and subtypes, the genetics, the current research landscape and more as a foundation for understanding important facts and figures about Usher syndrome. From there, the chapters will deal with key living realities throughout the life span as well as showcasing doable things to better support those with Usher syndrome. Better accessibility is a big one. Better supports and environmental modifications so that moving around and getting oriented is safer and easier. And, above all, better communication. This book is about better understanding the lives and complexities, so you won't be the umpteenth person who comments, 'Usher syndrome? Never heard of it.'

2
The Usher syndrome basics

Introduction

Usher syndrome is a degenerative genetic condition characterised by hearing loss and progressive loss of vision. The hearing loss may, in some types, be degenerative as well. The syndrome may be accompanied by a balance disorder (vestibular dysfunction).

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).

In a typical case, the hearing loss or deafness is sensorineural in nature and present usually from birth. The vision loss is acquired later and is progressive as a result of an inherited condition called retinitis pigmentosa. Retinitis pigmentosa usually begins in the first or second decade of life. There are several types and subtypes, and 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, types, diagnosis and its eventual consequence of deafblindness

now. In the next chapter, we look at more details on the genetics of Usher syndrome and the complexities inherent in the search for treatments and cures. It also explores early diagnosis and the changing knowledge and early intervention base of the condition. What was once a clinical diagnosis after the fact, when vision loss had been established, is now an early childhood or even *in utero* genetic diagnosis. For now, we focus on the basic data, types and important considerations. We also look at understanding what deafblindness or dual sensory impairment looks like as a consequence of Usher syndrome.

Data

In Australia, it is thought that Usher syndrome occurs in 1 in 6,000 people. In the UK, there are old estimates of 400,000 people living with deafblindness. Of these, 10,000 have a diagnosis of Usher syndrome. This data is 15 years old and reflects one of the key challenges in policies and services for people living with combined sight and hearing loss. Global estimates suggest over 400,000 people live with Usher syndrome.

Types of Usher

Usher syndrome falls broadly into three different clinical types: type 1, type 2 and type 3. Some authors also include an 'unclassified' or atypical category for those cases that don't fit into an existing type/subtype. The advent of genetic testing has meant, first, confirmation of clinical types for 1, 2 and 3 and subtypes for 1 and 2. Second, some of these held to be unclassifiable are found to be other types of Usher syndrome or of other forms of retinitis pigmentosa, both syndromic and non-syndromic.

There are ten genetic variations of Usher syndrome that have been found:

Type 1 (and six different subtypes 1B, 1C, 1D, 1F, 1G and 1J)

The clinical features are generally:

- Profound hearing loss at birth
- Progressive vision loss, usually beginning in the first decade
- Associated with vestibular (balance) dysfunction

Type 2 (and three different subtypes 2A, 2C and 2D)

The clinical features are generally:

- Severe hearing loss at birth, which may be progressive
- Progressive vision loss, usually beginning in the second decade of life
- No associated vestibular (balance) dysfunction

Type 3 (rarest form)

The clinical features are generally:

- Present in the decade after birth
- Normal hearing at birth with onset of progressive hearing loss at the end of the first decade, often around puberty
- Progressive vision loss can begin early in the first and sometimes second decade
- Around half will have associated vestibular dysfunction
- This rare type is most often found in people of Finnish heritage

Emma

I attended the 2025 Usher syndrome conference in the Netherlands. There was research presented there that shows the

types of Usher syndrome are not absolute: some Usher type 1 individuals do not have balance issues, and then some Usher type 2 individuals do have balance issues, sometimes associated with vertigo. A study of Usher type 2A showed that the pineal gland in the brain produces low melatonin, which contributes to the fatigue syndrome it is thought (Hendricks et al., 2023). So, the knowledge keeps improving.

Diagnosis

Historically, Usher syndrome has been a clinical diagnosis, but the advent of cheaper, more widely available and earlier genetic testing has changed the clinical, family and individual landscapes:

1. The age of diagnosis has shifted in the past two decades in many developed nations to infancy and earlier; for Emma and Annmaree, for example, diagnosis came with the advent of the vision loss and its symptoms.
2. Genetic testing has demonstrated that not every case that fits the clinical diagnosis does, in fact, have 'Usher syndrome'. There are over 200 different types/causations of retinitis pigmentosa, and hearing loss being reasonably common means that some are not affirmed on genetic testing.
3. In nations where newborn hearing testing is the norm and where clinical pathways include genetic testing of babies with hearing loss, a diagnosis of Usher syndrome now commonly occurs during infancy.
4. The advance of *in utero*/prenatal diagnosis means that this information may be available before the birth of the child.

Hearing loss: More about sensorineural hearing loss

In Usher syndrome, hearing loss is caused by damage to the inner ear structures, principally the hair cells of the cochlear, and/or the auditory nerve and its pathways into the brain.

Diagnosis is made as a newborn via the universal newborn hearing screening test, which in Australia began in 2002 and currently reaches 97% of newborns. Other countries with universal newborn hearing screening include Canada, China, the United Kingdom and the United States of America.

Clinical pathways exist in Australia where babies with hearing loss detected on screening are referred for a follow-up repeat test by a specialist audiologist with a diagnostic audiological assessment to confirm the hearing loss. This is followed by access to early intervention services (amplification, e.g. hearing aids, before the baby is three months old), referrals to support organisations such as Hearing Australia and, depending on the case, specialists such as ear, nose and throat specialists, paediatricians and geneticists.

Hearing loss will not always be detected at birth as in Usher type 3, where the loss typically starts in childhood.

Annmaree

My hearing loss (severe) was not diagnosed until I was 5 years old. My parents had concerns about my lack of awareness of what was going on around me and my very delayed speech. My kindergarten teacher expressed concern about my disobedience but wondered at how I taught myself to read. My brother was

diagnosed much earlier at 18 months of age. Younger extended family members were diagnosed at birth with the newborn hearing screening test.

Vision loss: More about retinitis pigmentosa

Retinitis pigmentosa is the leading cause of vision loss in people under 60, with an incidence of 1 in 4,000 people in the United States and Australia. It is the most common form of inherited retinal disease (IRD), but it is not a singular condition as such. There are over 200 different disorders of which Usher syndrome is one.

Retinitis pigmentosa is a condition that affects the light sensitive layers of the retina, which is the layer of tissue at the back of the eye.

The light sensitive layer of the retina is made up of two types of cells: rods that are concerned with night vision and peripheral fields; and cones which are involved in what we see in the centre of our visual fields and for colour. Central vision is important for seeing people's faces (and so for lip-reading, which means for some of us we get 'deafer' as we get 'blinder' when our central vision is affected).

Usually, in the retinitis pigmentosa of Usher syndrome, the rods are affected first and so the person experiences difficulty seeing at night and avoiding obstacles. Over time, this worsens until the central vision is impacted.

National Geographic has made a documentary about retinitis pigmentosa (not specifically Usher syndrome), which is compelling viewing, called 'Blink' (National Geographic, 2024).

Annmaree

In my case, I was hearing-aided and speech-therapied as a child and then diagnosed with retinitis pigmentosa in my early 20s. This vision loss had begun 10 years earlier in my teens, but I struggled to get an ophthalmologist to take my concerns seriously.

At first, the loss of peripheral vision manifested as night blindness and difficulty seeing in panorama. I fell into fountains and off jetties and bumped into people and objects, especially at night. This made me anxious and I took various steps to maintain my safety:

> Not drinking alcohol beyond a single glass (fully functioning mental capacity is critical to navigating the world, especially at night.
> Not going to unfamiliar environments alone or unprepared at night.
> Using my hands in front of me to feel for obstacles.

Diagnosis then was simultaneously a relief and a profound shock.

Balance disorder: More about vestibular dysfunction

This is caused by damage to the vestibular system, which is part of the inner ear. The vestibular system controls balance and spatial awareness. For those with the type of Usher syndrome that includes vestibular dysfunction, the combination of hearing and vision loss impacts child development, walking, dexterity and co-ordination, for example.

Genetics of Usher syndrome

This is explained in Chapter 3.

Treatments and cures

Currently, there is no definitive treatment or cure. There is much research and this is explored in the next chapter.

Consequences of Usher

The lived experience of Usher syndrome is one of complexities. It would be better called living experience, as Usher syndrome is an ongoing experience of change and adjustment throughout the life stages. It is not a static, same or stable condition.

The principal consequence is of degeneration of vision and/or hearing loss towards deafblindness. In types 1 and 3, this may be accompanied by vestibular dysfunction. These create complex consequences in the explanatory notes of the Nordic definition. The definition doesn't encompass vestibular dysfunction, however. We look at some of these complexities in the next few chapters, but they include:

- Reduced access to information
- Communication challenges
- Safe orientation and mobile difficulties

These may be accompanied in a social and physical world that is not supportive or adaptive by:

- Increased falls and accidents
- Increased rate of experiencing visual hallucinations
- Increased psycho-emotional adjustments
- Increased health and social care support needs
- Increased risk of social isolation and the accompanying health and well-being reduction

- Increased risk of cognitive decline in older age (potentially remediable/preventable with good health and social care)

This may seem an insurmountable list of challenges and complexities, but we repeat here that living with Usher syndrome is better in a world that provides good health and social care and support. We are enormously capable, and part of the reason for this book is to increase understanding among health and social care students, professionals, practitioners and the institutions they work in.

Important consideration

One of the challenges in supporting people with Usher syndrome is the tendency to approach and manage each impairment separately without considering the impact of the sensory losses upon each other. This is particularly true where there is no recognition and dedicated specialist support of the hearing loss and low vision (and balance disorder when it co-occurs). Much of the single impairment support and assistive technology uses the other sense, for example, screen readers for low vision rely on hearing voices, and sign language for hearing loss requires sight unless a tactile form is learned.

Furthermore, each individual's Usher journey and experience is different, and what is needed to support one phase may not be enough for the next. There is lifelong adjustment for most. Usher syndrome is a psycho-emotional journey as well, which can be experienced as anticipatory grief, bereavement reactions to each loss and as Gullacksen et al. suggest, adjustment to each stage may take years and may be superimposed by the next phase of loss/es (2011). Partners, parents, family members and carers may also experience grief and life adjustment stresses as well.

Deafblindness-dual sensory impairment

To begin to understand what it is like living with Usher syndrome, we need to examine deafblindness or dual sensory impairment, to give two common terminologies. Repeating the point made earlier, the impact of each sensory loss needs to be considered not only separately but also together. An individual with Usher syndrome may not consider themselves deafblind for years or decades, but the condition over time moves towards increasing vision loss that compromises the ability of residual hearing to cope. For many with Usher, there may be further or ongoing hearing loss that accompanies the vision loss alongside balance for those with some types.

Definition

For this book, we use the Nordic definition as revised in 2024. It's a detailed, clear and useful definition:

> Deafblindness is a combined vision and hearing impairment of such severity that it is hard for the impaired senses to compensate for each other. Thus, deafblindness is a distinct disability.

But the definition itself doesn't capture the complexities, and the principal reason this definition is a good one is that it comes with a set of explanatory notes.

Main implications:

- To varying degrees, deafblindness limits activities and restricts full participation in society. It affects communication, access

to information, orientation and the ability to move around freely and safely.

- To help compensate for the combined vision and hearing impairment, the tactile sense becomes especially important.
- There is a high risk that the physical and psychological health as well as social life will be affected.

Comments:

On the combined vision and hearing impairment, the severity of the combined vision and hearing impairment depends on:

- The time of onset, that is, whether it is congenital, acquired or age-related, especially in relation to communication development and language acquisition.
- The degree and nature of the vision and hearing impairments.
- Whether it is combined with other impairments.
- Whether it is stable or progressive.

On the distinct disability, the fact that it is hard for the impaired senses to compensate for each other means that:

- Attempting to use one impaired sense to compensate for the other one is time consuming and energy draining; thus, sensory perception is often fragmented.
- A decrease in the function of vision and hearing increases the need to utilise other sensory stimuli (i.e. tactile, kinaesthetic, proprioceptive, haptic, smell and taste).
- It limits the access to information from a distance.
- It creates a need to rely on information within the near surroundings.
- To create meaning, it becomes necessary to rely on memory and draw conclusions from fragmented information.

On activities and participation, deafblindness limits activities and restricts full participation in society.

According to the United Nations Convention on the Rights of Persons with Disabilities (UNCRPD), participation is a given right for all human beings. Thus, to enable the individual to use their potential capacity and resources, society is required to facilitate specialised services.

The individual and their environment should be equally involved, but the responsibility for granting access to activities lies with society. An accessible society should at least include:

- Available competent communication partners.
- Available specialised deafblind interpreting, including interpreting of speech, environmental description and guiding.
- Available information for everyone.
- Human support to ease everyday life.
- An adapted physical environment.
- Accessible technology and technological aids.

> A person with deafblindness may be more disabled in one activity and less disabled in another. Variation in functioning might be the consequence of both environmental and personal factors.
> Specialised competence related to deafblindness, including an interdisciplinary approach, is vital for proper service provision (Nordic Welfare Centre, 2024).

Again, for some with Usher syndrome, the deafblindness-dual sensory impairment will be accompanied by vestibular dysfunction that will challenge attaining physical motor milestones, safe orientation and mobilising, for example, on top of the complexities of hearing and vision loss.

We examine some of the complexities in chapters 4–9, from the individual with Usher syndrome perspective, with diagnosis, communication, health and well-being. From the family, carer, partner, child of a parent with Usher syndrome or a sibling, we present some perspectives here. Usher syndrome is not a solo journey.

3
Usher syndrome, genetics and more

Introduction

Usher syndrome is a complex, diverse and sometimes challenging condition for people and families. It can also be challenging to untangle the types, the genetics and the emotional fallout. This is why we need health and social care professionals and practitioners as well as wider society to take the time to understand these complexities and support us all to have the best lives possible. In this chapter we look at:

Usher Syndrome Types
Usher Syndrome Type 1
Usher Syndrome Type 2
Usher Syndrome Type 3
Diagnostic Pathways and Considerations
Newborn Hearing Screening
Genetic Diagnosis
Early Intervention
Audiological Intervention
Vestibular Dysfunction

Usher syndrome types

The genetics of Usher syndrome are complex and variable. To assist in understanding the genetics of Usher syndrome and how Usher subtypes are classified, it may be useful to explain a few of the basics.

Our 'genotype' refers to the unique genetic makeup contained in our DNA (deoxyribonucleic acid), which consists of approximately 20,000 genes (National Human Genome Research Institute, 2025). These genes provide instructions for making proteins that are required for our bodies to function properly. Sometimes, changes in our genes occur that affect our ability to function properly. A gene change that is known to cause disease is called a 'pathogenic variant'. A 'benign variant' is a gene change that does not lead to disease. Some gene changes may also be classified as a 'variant of unknown significance', meaning that they have not yet been linked to disease, but further research is necessary to confirm that they do not cause health problems.

Our 'phenotype', however, represents how our characteristics, physical traits, behaviours and health conditions manifest as

a result of our specific genotype (National Human Genome Research Institute, 2025). In the case of Usher syndrome, there are currently 10 genes known to be associated with the condition, each with multiple pathogenic changes contributing to the characteristics, that is, the phenotype, of Usher syndrome. These are categorised into three types of Usher syndrome: type 1, type 2 and type 3, which are further categorised into subtypes based on the specific gene involved.

Usher syndrome type 1

There are six Usher syndrome type 1 subtypes – 1B, 1C, 1D, 1F, 1G, and 1J – that are caused by different changes in genes MYO7A, USH1C, CDH23, PCDH15, USH1G and CIB2, respectively. All affect the development and function of stereocilia in the inner ear and photoreceptors in the retina. Stereocilia are tiny hairs within the inner ear that help detect sound and movement. When they are absent or not functioning properly, as is the case with Usher type 1, it typically causes a congenitally profound deafness and vestibular dysfunction. Deafness is typically managed with cochlear implants. Absent or non-functioning photoreceptor cells in the retina cause an eye condition called retinitis pigmentosa. In Usher type 1, retinitis pigmentosa typically presents as night blindness in the first decade of life, with progressive loss of peripheral vision into adolescence.

Usher syndrome type 2

Usher syndrome type 2 has three subtypes caused by three different gene changes. Changes in the USH2A are known as Usher syndrome type 2A and changes in ADGRV1 and WHRN (aka

DFNB31) are commonly known as Usher syndrome type 2C and 2D. These gene changes affect the functioning of the stereocilia in the inner ear and photoreceptors in the retina. Unlike type 1, the stereocilia remain partially functioning, leading to moderate to severe hearing loss that can typically be well managed with hearing aids. Stereocilia in the vestibular system are not affected, meaning individuals with Usher syndrome type 2 have normal balance function. The signs of retinitis pigmentosa are present in late adolescence or early adulthood.

Usher syndrome type 3

Usher syndrome type 3 is relatively rare compared to types 1 and 2, but is more prevalent in certain populations such as Finnish and Ashkenazi Jews. Type 3 is caused by changes in the CLRN1 gene and causes the progressive degeneration of stereocilia in the inner ear and photoreceptors in the retina. As such, those with type 3 experience both progressive hearing and vision loss. Individuals are typically born with normal hearing, meaning that their pathway to diagnosis will be different from those with types 1 and 2. Vestibular dysfunction can also develop and degenerate over time. Due to the variability in symptoms, diagnosis can be difficult and often late, leading to a lack of support.

Diagnostic pathways and considerations

As healthcare professionals, it is imperative to understand how advances in newborn hearing screening, genetic testing, early intervention and audiological interventions have changed the

trajectory of those born with Usher syndrome today compared to those who are now in their 30s and older.

Newborn hearing screening

Today, babies born with Usher syndrome in highly developed countries often have their deafness identified during routine newborn hearing screening and subsequent diagnostic audiology. Newborn hearing screening programmes have only become widespread over the past two decades, allowing deafness to be identified within weeks of birth rather than taking years, as it did in the past (Butcher et al., 2019).

Genetic testing

Over the past few decades, advancements in genetic testing have transformed the diagnosis of those born with Usher syndrome. Previously, Usher syndrome was diagnosed after the onset of vision symptoms, which typically occurs in early adolescence to early adulthood, years after the first symptoms presented. Today, parents of children who have been identified through newborn hearing screening programmes can elect to have genetic testing for a range of genetic conditions known to cause deafness. This testing has gone from costing families thousands of dollars and taking many months to receive results to being done for a few hundred dollars in a matter of weeks. Advancements in genetic testing have led to Usher syndrome being a condition that can now be diagnosed and proactively managed from infancy.

For parents of children with Usher syndrome who *do not* have early genetic testing to identify the cause of their child's hearing loss (either by choice or by lack of access), the Usher syndrome

diagnosis can be made on a functional and/or clinical basis (Maxwell et al., 2025). But this can be years later, with missed opportunities for timely interventions and supports.

Early intervention

Soon after the diagnosis of deafness, parents are often swept up in a pathway of early intervention that supports their child's language and communication development, social-emotional well-being, gross motor development and access to learning. This comprehensive support is provided by a multidisciplinary team that may include speech therapy, sign language support, family counselling and early childhood education. If an Usher syndrome diagnosis is made during these years, early intervention may also include targeted physiotherapy and occupational therapy for compensatory skill development caused by the vestibular dysfunction as well as additional counselling for families to cope with the grief and loss caused by the Usher syndrome diagnosis.

Audiological intervention

The ongoing management of the child's hearing needs can include the fitting of hearing aids and assessment of the child's eligibility for cochlear implantation. This typically includes an MRI (Magnetic Resonance Imaging), which is a precise medical image to confirm the formation of the cochlea. A cochlear implant is a medical device that helps people with profound deafness hear, including those with Usher syndrome type 1 (see Figure 1). There is an internal component called the electrode array that is implanted during surgery

as well as an external component consisting of a sound processor that is worn behind the ear, similar to a hearing aid, and a magnetic coil that connects the internal and external components.

Where a hearing aid amplifies sound, the cochlear implant bypasses the absent or damaged stereocilia in the inner ear to convert sounds into electronic signals through the auditory nerve, which are then interpreted by the brain. For infants, this surgery is ideally done bilaterally (both ears at the same time) from 6 to 12 months of age (Leigh et al., 2024). This intervention was pioneered by Professor Graeme Clark in Melbourne, Australia,

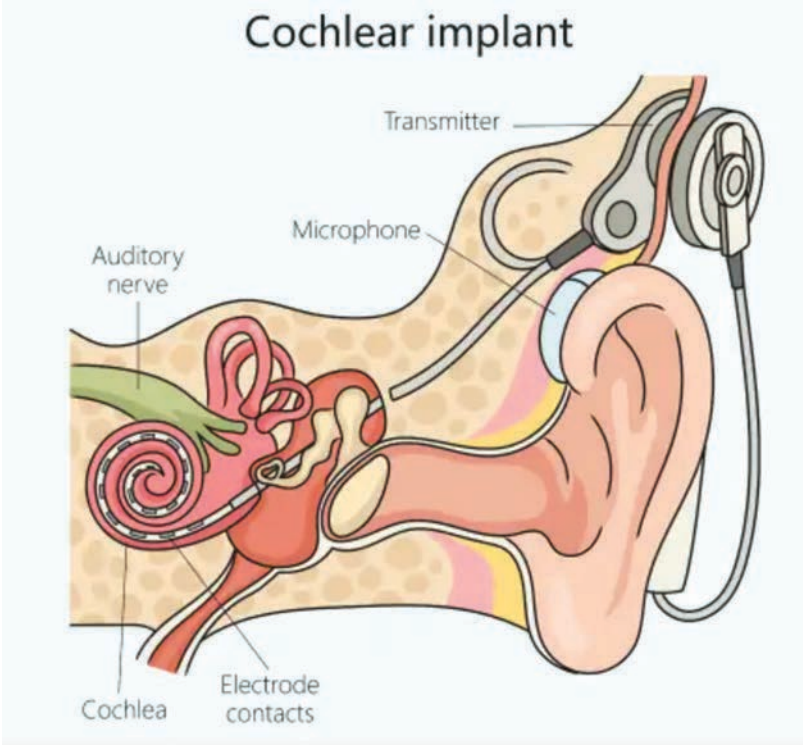


Figure 1: Cochlear implant

and became routine for adults in the 1980s and for children and babies from about the 1990s (Montgomery & Mankekar, 2024).

Before routine cochlear implantation, individuals with Usher syndrome type 1 relied on sign language as their primary mode of communication. This is a really important distinction to make, as the support needs of individuals with Usher syndrome type 1 who use sign language to communicate are very different from those of those who have had access to prelingual cochlear implantation and go on to develop age-appropriate speech and language.

Vestibular dysfunction

Another distinctive characteristic of Usher syndrome type 1, and at times, type 3, is vestibular dysfunction. This is present at birth in those with type 1 and disrupts the development of gross motor skills due to missing or damaged stereocilia within the inner ear. Individuals with vestibular dysfunction will have difficulty with balance, orientation and movement, which can result in infants appearing floppy and struggling to meet typical gross motor milestones such as sitting, crawling and walking. The Childhood Hearing Australasian Medical Professionals (CHAMP) network recommends further investigation of any deaf children who are not walking by 18 months of age to rule out Usher syndrome type 1 (Sung et al., 2019).

As the child grows, healthcare providers and educators need to understand the impact of vestibular dysfunction on safety. This includes issues related to swimming, such as disorientation in the water, poor balance and co-ordination, as well as a higher

risk of falls due to poor balance and spatial awareness combined with a reduced visual field of vision. When the child begins to experience peripheral vision loss, this exacerbates the vestibular dysfunction, putting the child at further risk of falls and accidents.

Fatigue

There is also the issue of fatigue. Regardless of age, we would expect anyone with Usher syndrome to fatigue more than their peers due to having reduced compensatory mechanisms for their hearing, vision and vestibular deficits (Hendricks et al., 2023). This is important to know as healthcare professionals, as they may require more breaks between appointments, assessments and therapy as well as accommodations and modifications within educational and work settings.

More on genetic diagnoses

The only way to definitively diagnose Usher syndrome and determine the gene involved is through genetic testing. This is important for a few reasons:

It gives individuals and their families information on the prognosis of symptoms. Each type of Usher syndrome has a variable onset and severity of symptoms, so knowing the exact gene involved helps to understand the natural history of the disease. This can help prepare individuals and their families to ensure that the right supports are in place.

It helps clinical researchers understand the natural disease progression of the various types of Usher syndrome. By tracking disease progression over time, researchers can adjust their

techniques to cater for the various gene changes associated with Usher syndrome. It will also become particularly important as clinical trials and gene therapies emerge, as knowing the specific gene change ensures the eligibility of individuals for treatments.

It gives families information about inheritance risks so they can make informed decisions about family planning. Typically, Usher syndrome is caused by an autosomal recessive inheritance; autosomal – meaning that the gene change is located on one of the first 22 pairs of chromosomes, called the autosomes, as opposed to a gene change on the 23rd pair known as the sex chromosomes, and making Usher syndrome equally affect males and females; and recessive – meaning that a child must inherit the same gene change from both parents to be affected. Most often, the parents are carriers of one copy of the gene change without showing any symptoms themselves. For each pregnancy, two carriers of the same gene change have a 25% chance of having a child *with* Usher syndrome; a 50% chance of a child being a *carrier*; and a 25% chance that the child will be neither a carrier nor affected with Usher syndrome.

An important emerging genetic advance is the increasing acceptability and availability of carrier screening. This is where prospective parents investigate the risk of potential pregnancies being born with certain recessive conditions, including Usher syndrome. One parent provides a blood or saliva sample to test for genetic changes known to cause disease. If genetic changes are found, the second prospective parent undergoes targeted testing to determine if they also carry the same gene change. This would result in knowing if both parents are carriers of the same condition *before* a pregnancy and can then explore

reproductive options, including IVF (In Vitro Fertilisation) combined with PGD (preimplantation genetic diagnosis), a process where the embryos are tested for the genetic condition *before* implantation into the uterus.

IVF and PGD are also options for parents who already have a child with Usher syndrome and would like to reduce their risk of any subsequent pregnancies having the condition. Another option is a prenatal genetic test to determine if the foetus has Usher syndrome during pregnancy in a couple known to be at high risk, either due to positive carrier screening results or having previously had a child with Usher syndrome. This information allows prospective parents to make informed reproductive choices, including continuing with the pregnancy, planning early supports or exploring other reproductive options.

In rare cases, a spontaneous gene change (called a *de novo* change or 'new' change) can occur during the development of a foetus *in utero*. This causes the same genotype and phenotype as recessive inheritance without the parents being carriers of a gene change. Therefore, it is important to determine if both parents are carriers to allow accurate and informed decisions about family planning.

Suffice it to say, genes are complex, and so are the implications of genetic changes. Individuals and families must receive adequate information and support through genetic counselling before undergoing any genetic testing, carrier screening or when receiving genetic results.

The need for collaboration and research balance

There is a need for cross-disciplinary collaboration across health and social care to reduce the burden on parent (Johansen et al., 2024). The current system is fragmented and focusses on single impairments of hearing and vision without considering the uniqueness and distinctiveness of a combined hearing and vision loss (and balance disorders for some). We look at this in more detail in Chapter 13 on health and well-being. We look there at the idea of super teams where everyone works together, to learn from each other and share experiences to help those with Usher syndrome and their families to reach their goals and flourish.

It's not just the professionals and practitioners working with Usher syndrome children, young people, adults and their families that must be collaborative. There is a clear need to balance the scientific and clinical research with psychosocial research for the benefit of all.

The role of the geneticist and genetic counsellor

Suffice it to say, genes are complex, and so are the implications of genetic changes. Individuals and families must receive adequate information and support from a geneticist and a genetic counsellor before undergoing any genetic testing or carrier screening, and when receiving genetic results.

While the geneticist focuses on the medical and scientific aspects of diagnosis, the genetic counsellor complements this

by providing psychosocial support, education and guidance, helping families process complex medical information and make informed decisions.

Geneticist

A geneticist is a medical doctor who specialises in the study of genes, genetic variations and heredity in living organisms. Geneticists focus on understanding how genes influence health and disease, playing a key role in diagnosing and treating genetic conditions. The geneticist is responsible for ordering appropriate genetic tests based on the patient's clinical symptoms and family history.

Once the genetic tests are complete, the geneticist interprets the results to confirm a diagnosis. They determine the specific genetic mutation responsible for the condition and how it aligns with the clinical symptoms.

Genetic Counsellor

A genetic counsellor is a healthcare professional with specialist knowledge in human genetics, counselling and health communication. They provide critical information and support to individuals and families affected by genetic conditions. Genetic counsellors typically work as part of a multidisciplinary team that includes a geneticist. The genetic counsellor meets with the family to explain the geneticist's findings in a way that is easy to understand. They discuss inheritance patterns, risk factors for other family members, and the long-term implications of the condition. They offer support to help individuals and families navigate the emotional aspects of the diagnosis.

It is important to note that a diagnosis of Usher syndrome is very different from receiving a diagnosis of a chronic illness or viral infection. A diagnosis of a genetic condition has significant implications for other family members and for current and future generations. Genetic counsellors have a duty of care to other family members who may be at risk of being affected or of being carriers of the genetic condition. They can provide information about the likelihood of other family members being affected or being carriers of the same genetic mutation. This is known as cascade testing. The genetic counsellor will provide support with the complexities of who to tell, when to tell, what to tell and how to tell.

It is also important to note that the genetic counsellor is not just useful at the time of a new diagnosis. They can also be a support throughout the family life cycle, including facilitating cascade testing for other family members, allowing parents to make informed decisions about future pregnancies, assisting with the transition of a child with a genetic condition from child to adult health services, and guiding siblings through carrier testing once they reach reproductive age and are capable of making autonomous decisions about their own genetic makeup.

Usher syndrome research

Research into Usher syndrome typically falls under two umbrella terms: *clinical research and psychosocial research*. Clinical research is typically informed by laboratory studies that aim to understand how Usher syndrome affects the body, including genes, cells, eyes and ears. Having a good understanding of how Usher syndrome impacts people at a biological level can lead to developing ways to slow or reduce the impacts of the condition.

Psychosocial research, on the other hand, is undertaken by a range of specialists to understand how Usher syndrome shapes the way people live their lives, their relationships and their participation in the world around them.

Both types of research aim to improve quality of life for those living with the condition and their families, now and in the future. In this section, we cover the basics of clinical research; psychosocial research is discussed in Chapter 4.

Clinical Research

Over the past few decades, technological advances and increased accessibility of the cochlear implant and hearing aid technology has supported the management of the hearing loss associated with Usher syndrome. Therefore, many of the emerging therapies that are showing promise for people with Usher syndrome are focused on ways to treat the vision aspect of Usher syndrome, caused by a degenerative eye condition called retinitis pigmentosa.

Retinitis pigmentosa is characterised by absent or non-functioning photoreceptor cells in the retina. The retina is a thin layer of tissue at the back of the eye and is responsible for detecting light and initiating the visual process. In a healthy eye, photoreceptors in the retina convert this light into electronical signals that are sent to the brain via the optic nerve and interpreted into images. When the photoreceptors are absent or do not function properly, as is the case for those with Usher syndrome, there is a disruption to this process and the brain receives incomplete information.

In Usher syndrome, retinitis pigmentosa affects the periphery first. In Usher Type 1, retinitis pigmentosa typically presents as

night blindness in the first decade of life, with progressive loss of peripheral vision into adolescence. In Usher Type 2, the signs of retinitis pigmentosa typically present in late adolescence or early adulthood.

There are a number of promising clinical trials underway around the world that hope to slow down, halt or reverse the degeneration retinitis pigmentosa causes in individuals with Usher syndrome.

Gene therapy

Gene therapy is a treatment to deliver a healthy copy of the Usher gene into the retina cells. The healthy copy contains information for producing Usher proteins, which are not produced with patients as a result of mutations in Usher genes. The healthy copy is delivered by a viral vector, a transport mechanism that carries the gene to the targeted cells to replace the defective gene. This is done through an injection into the back of the eye by a retinal surgeon. By restoring the protein production, the gene therapy has the potential to slow down or possibly stop the deterioration of the retina.

One of the problems with many of the genes causing Usher syndrome, however, is that they're too big to fit into the normal viral vectors that are typically used to deliver genes to retinal cells. To overcome this, researchers are developing techniques to split the gene into smaller parts, deliver them using viral vectors, and then certain chemical components cause the gene to reassemble after delivery.

Gene therapy is a gene-specific treatment, meaning each therapy needs to be tailored to the specific gene that is affected

for each of the various genes known to cause Usher syndrome. These gene-specific therapies will be most useful for those people in the early stages of their retinal degeneration.

There is currently only one commercial gene therapy that has been regulatory approved for treatment for an inherited retinal disease and that is called Luxturna. It was approved in the US in 2017 and then in Australia in 2020 and is used to treat a condition called Leber's congenital amaurosis specifically caused by mutations in the RPE65 gene.

RNA editing

This is another gene-specific treatment technique that will likely benefit those with more moderate stages of disease progression. Instead of replacing the gene itself, the disease process is modified to correct the errors in the gene messaging before the protein is made, without changing the underlying DNA. The technique is not permanent, meaning patients would need to repeat the treatment over time.

Stem cells

Stem cells are specific cells that have the potential to develop into many different cell types, including heart, nerve, and eye tissue.

There are two main kinds of human stem cells: Embryonic stem cells and non-embryonic (adult) stem cells. Non-embryonic (adult) stem cells can be obtained from tissue such as skin. Unlike embryonic stem cells, skin samples do not have the same associated ethical issues.

Skin samples from people with specific eye problems, such as Usher syndrome, can be grown in the laboratory, turned into

stem cells and reprogrammed to become retinal tissue, i.e. rods and cones which are damaged in retinitis pigmentosa. This allows researchers to study retinal cells without the need to take a sample from your eye. This will help researchers better understand how Usher syndrome develops, explore potential new treatments and test new drugs that may slow or prevent vision loss.

A cautionary message about stem cells. Unfortunately we live in a world where people are taken advantage of and those that prey on our most vulnerable. There have been many stories online where people claim to use stem cells to 'cure' vison loss in people with inherited retinal diseases including Usher syndrome. For a hefty price tag of tens of thousands of dollars, these procedures are experimental and have no medical evidence that they work. They can end up doing more harm than good and also make some ineligible for other treatments that may come in the future.

Gene agnostic treatments

Gene agnostic treatments have the potential to assist a larger number of people with Usher syndrome because they are not limited to a specific gene change. One major area of research at the moment is retinal protection using antioxidant medication. These are tablets typically taken orally every day as opposed to a one-off eye injection and are aimed at reducing the oxidative stress in the retina caused by retinitis pigmentosa. Another gene agnostic approach is optogenetics, where drugs are used to change the function of the other cells in the retina so that they take over for the cells that have been lost.

Natural history studies

Historically, people with inherited eye conditions including Usher syndrome didn't have treatment options, so they tended to fall off the radar of health systems. Researchers in the field are working to overcome this challenge by doing lots of preclinical work to firstly understand the disease progression of Usher syndrome at a cellular level, but also prepare those in the community for effective treatments. This is done through natural history studies or non-interventional studies, where people are monitored over a long period of time to see how the different genes affect vision and retinal health over time. This can lead to a better understanding of how different treatment options will impact the disease at various stages of progression.

There is also a large push in this area of study to ensure those living with the condition all have access to a genetic confirmation of Usher syndrome via genetic testing, as many of the therapies in development will be gene-specific and tailored to the particular gene affected. This also leads to more accurate prevalence data, which is crucial for justifying funding and resources for these conditions.

Registries

Registries are ways of safely storing and sharing the medical information of people with a range of different conditions. The registries are used by researchers across the world to gain a better understanding of the natural history of conditions, to better inform the clinical work they are undertaking. For those with Usher syndrome, there are a number of different registries

available globally, and they are used to find people that may be suitable for clinical trials.

In summary, there is an army of scientists across the world working on treatments for people with Usher syndrome so the message really is for those with the condition to stay connected to the community and remain informed about what potential treatments are in the pipeline. For the healthcare professionals working with people with Usher syndrome, there is reason to remain hopeful about the potential these trials give. In the next chapter, we will explore psychosocial research and how it shapes the way in which people live their lives today.

Usher syndrome is a spectrum

We need to remember that Usher syndrome is a spectrum. Usher syndrome is experienced to different degrees by individuals and at different rates, even between siblings with the same gene changes.

Annmaree

My brother and I have Usher syndrome and we have different rates of deterioration, varying residuals in hearing and vision and different ways of communicating. He has more hearing than I do now (although that probably wasn't the case in childhood: I have lost hearing and he hasn't) and less vision due to other factors imposed upon the retinitis pigmentosa of Usher. This means he will answer your phone call, whereas I won't/can't. I can still read (barely) if electronic, largest font possible and dark moded. I am anticipating a psychosocial storm when I lose reading altogether. My brother uses audiobooks, and I admit it, I am jealous of this superpower.

Ongoing challenges

It is really important we all recognise that no matter what type of Usher syndrome we have, we aren't protected from the usual vicissitudes of modern life: we catch germs, we get cancer, we have breakups and we have accidents. Some things are more common for us, such as falls and accidents, as our sight and hearing loss can increase everyday risk because we don't hear environmental warning signs or see the obstacles in front of us, for example. Some conditions that aren't related to Usher syndrome can negatively impact our sight and/or hearing or our coping: a retinal hole? Bang goes some precious sight. We who live with Usher are still subject to the illness, injuries, life issues and stressors of normal everyday life; we're not immune to these. Chapter 4 looks more intensively at diagnostic challenges and complexities.

4
Complexities and challenges of diagnosis

Introduction

We need to remember that Usher syndrome is a spectrum. Usher syndrome is experienced to different degrees by individuals and at different rates, even between siblings with the same gene changes. It's therefore important that health and social care professionals acknowledge that Usher syndrome is not a static condition, but a dynamic one. A family may confront a newborn diagnosis of hearing loss and then later a genetic diagnosis of Usher syndrome with the future prospect at some unknown time, likely in the first or second decade of life, of onset of progressive vision loss. Some may also have to contend with the impacts of balance disorder on development and daily life. At different life stages, a person with Usher may have different residuals and need different communication methods, additional learning support and various assistive technologies to support access to information and communication.

Thus, a diagnosis of Usher syndrome comes with a raft of complexities and challenges that usually begin from birth. In this chapter, we explore:

Navigating the Impact of Diagnosis
The Pros and Cons of an Early Diagnosis
Challenges of Diagnosis
Diagnosis Disclosure
How and When to Tell: A Guide for Parents
Ongoing Challenges

Navigating the impact of diagnosis

Emily

Now, let me take you back to when my son was 12 months old. It was 2011, and we were attending a routine eye examination with a paediatric ophthalmologist, based on the recommendation that children with hearing loss have annual eye examinations. I was desperate to find out the cause of my son's deafness as I always felt there was more to learn about him. He was floppy, frustrated, and failing to meet his gross motor milestones. Dr Google told me that the combination of deafness and vestibular issues could be caused by Usher syndrome. We knew the condition was rare. However, our early intervention team told us that apart from language and communication milestones, everything should be developing alongside typically hearing children. It wasn't. And I sensed the worst.

I asked the ophthalmologist, 'Could my son have Usher syndrome?' Their reply has stayed with me to this day: 'Why would

you want to know?' As there was no treatment for the vision loss caused by Usher syndrome, they didn't understand the point of wanting to know this early on in his life. 'It could be many years until we can rule it out.' This was based on the diagnosis of Usher syndrome being made on the clinical presentation of retinitis pigmentosa, the eye condition associated with Usher syndrome. It was 2011, and routine genetic testing was not yet widespread. I definitely wanted to know if my son was going to lose his vision. Surely, there were skills I could be teaching my child to prepare him for the deteriorating vision that was ahead of him. Surely there were clinicians, educators, and families who were familiar with Usher syndrome whom I could learn from. And I definitely didn't want to turn up to his annual eye exam with the stress of waiting to be told bad news each time.

We connected with a local genetic service, and because we perhaps had not yet finished our family, were deemed eligible to have a deafness gene panel test done through the University of Iowa in the USA. The gene panel included testing for the approximately 100 genes associated with childhood deafness. Genetic testing at this time was seen more as a family planning decision-making tool than a diagnostic tool to help us give our child the best possible care. We wanted to know what we were up against, and AUD 3,500 and nearly a year later, we received the confirmation we had been dreading: a diagnosis of Usher syndrome for our then 3-year-old. The geneticist went on to talk about recurrence risks for future pregnancies, but all I wanted to do with this information was to work out the best way to care for my son and identify who was going to do that caring.

We left the hospital with no real plan of what to do. We asked about a support group, but due to the rarity of the condition, the geneticist said this was 'highly unlikely' but suggested we Google it. No such organisation existed. We called a national vision organisation, hoping to connect with someone who might be able to guide us. 'How is your son's vision now?' As far as we knew, it was fine, so we weren't eligible for support. 'Call us back when his vision changes' was the message. We were completely alone. And frightened. We made calls to our family to tell them the news. He wasn't just a son, but also a grandson, a nephew, a cousin. I hated not being able to reassure our loved ones nearly as much as they hated not being able to reassure us. I needed to know that everything was going to be okay.

The months that followed were a blur. There was lots of reading and trying to find someone, anyone, who knew about this condition. I was desperate to find another parent who was also walking this path. I wanted someone to understand what I was going through. I now felt out of place in the school ground, with friends and with our mother's group. I now had different challenges than nits and lunchbox fillings. We found an established support organisation, the Usher Coalition, in the USA and they had an International Usher Symposium planned for the following year. Finally, we were going to find some answers.

Several months later, an article in the local paper changed our trajectory and led us to another family living close by. We felt like we had hit the jackpot. This connection led to a joint trip to Boston in 2014, where a new world of clinical research and gene therapy presentations awaited us. It seemed that an army of scientists was working on slowing down or even halting the vision

loss associated with Usher syndrome. There was also a close-knit community of those living with Usher syndrome, their family and clinicians banding together to support the here and now. The Symposium gave us hope for the future as well as the motivation to establish something similar in Australia.

And so UsherKids Australia was born. Co-founded with fellow parent, Hollie Feller, we have become a force in building a pathway of support for families across Australia who face the same devastating diagnosis. We have successfully advocated for improved clinical education and raised much-needed awareness of the condition within the medical, disability and education sectors. Our community has now grown to over 100 families in Australia, with a sister organisation in the UK (www.usherkidsuk.org).

We regularly host community events, including family days, online parent calls and, more recently, an Usher Youth Camp. These opportunities for connection with those with living experience of Usher remain the core of what we do and the reason we were established over 10 years ago. Meeting someone who 'gets it' reduces isolation and fosters a sense of belonging, reminding families and young people they are not alone in their journey. It also helps to have a growing community when it comes to understanding the needs of those with Usher syndrome, and knowing what organisations like ours need to be advocating for.

The pros and cons of an early diagnosis

Over the last 10 years, we have seen a huge shift in the age that our children are diagnosed, well before the onset of visual

symptoms and the opportunity this allows for early supports, including peer support. Today, if a family receives a genetic diagnosis of Usher syndrome for their child, there is a pathway in place that connects the parents to a well-established support organisation. And depending on their location, subtype and needs, we are able to put them in touch with others who understand exactly what they are going through and provide answers to all those questions that we had on those very dark, early days.

But, sometimes, the emotional toll can be too much for some families. Considering many may still be coming to grips with the hearing loss diagnosis, and then within months learn that their child's vision will also be impacted, you can imagine how distraught parents may be.

But are there any cons to early diagnosis? Here are some considerations, keeping in mind the benefits typically outweigh the drawbacks when proper support and counselling are provided.

Underdeveloped pathways for paediatric Usher syndrome

There can still be problems for families accessing support for the vision aspect of their child's Usher syndrome, whether it be through early intervention or education settings, *before* the onset of their functional vision loss. This 'come back when they're vision impaired' message denies the child and family access to supports early on that can build their capacity to cope with the vision loss to come and improve their outcomes in the future (see Maxwell et al., 2025, to explore this further).

Low expectations

Children and/or their parents, family members, healthcare providers and educators might subconsciously limit themselves based on the expectation of future limitations. They may choose 'safe' paths in education or career rather than pursuing their true interests or potential or overlook adaptations or supports that could make participation possible, assuming instead that something is 'not for them'.

No longer living fully in the present

Knowing about future vision loss might cast a shadow over experiences that could otherwise be fully enjoyed, as families may find themselves preoccupied with what is to come rather than living fully in the present. This anticipation can amplify worry, heighten grief for potential future losses, and reduce opportunities to embrace current strengths and abilities. The additional appointments and therapy involved also take children and their families away from typical activities they might be otherwise enjoying, further burdening the experience in the early days after diagnosis.

These potential cons are balanced against the significant benefits of early diagnosis, such as early intervention, better preparation and informed decision-making.

Generational perspectives and the question of independence

Perspectives on early diagnosis also vary across generations within the Usher community. Many adults who grew up without

an early diagnosis describe childhoods where they were free to simply be children, navigating the world independently, learning through experience, and developing problem-solving skills without the presence of mobility aids, specialist support, or constant adult supervision.

For some, seeing children today receive early interventions such as cane training, guide dog preparation, or specialist teacher support while their vision remains relatively strong can feel confronting. These supports may be perceived as unnecessary, prematurely limiting, or as introducing a disability identity before the child themselves experiences functional vision loss.

At the heart of this perspective is a deeply held value: the importance of independence, autonomy, and self-discovery. Many adults with Usher syndrome credit their resilience and confidence to having been allowed to take risks, make mistakes, and develop their own strategies for navigating the world.

Challenges of diagnosis

Despite advances in prenatal, neonatal and childhood diagnostic pathways, there have been, are and still will be young adults and adults who are diagnosed with Usher syndrome at a later age. This is due to:

1. Some subtypes present with hearing loss in later life therefore will not be detected by newborn hearing screening tests.
2. Some younger adults will have missed out on newborn screening as, for example, in Australia, universal screening became national and universal in 2002, 23 years ago, as at the time of this publication.

3. Some low-income developing countries in Southeast Asia and the Middle East don't have universal newborn hearing screening.

It is also important that health and social care professionals understand later diagnosis in the second or third decade of life, as this reflects the experience of most adults with Usher syndrome today. Over time, this will shift, with later diagnosis becoming less common. For most families and people diagnosed with Usher after childhood, this is a crisis point.

Emma

I was involved in a project looking at the experience of diagnosis of Usher syndrome in adulthood. This arose out of the need I was seeing in the community as I worked with newly diagnosed people with Usher syndrome to navigate this crisis point. I wanted to support them towards acceptance and empowerment. We also wanted to find out what the Usher community wanted from diagnosis in the UK. This was a long term project and involved questionnaires and research phases. We hoped to create an online diagnostic tool and resources. The following data was presented at the 2019 Deafblind International conference in Australia.

Diagnostic clarity

The first step is to get a clear diagnosis, and in an ideal best practice world, this means the person:

1. Gets a clear diagnosis.
2. Is given the right amount of medical information.
3. Is referred to a specialist medical service.
4. Is referred to a specialist support service.

How much information is right?

After a clear diagnosis, we need to know how to get the volume of information right: were people and families overwhelmed by too much information, or were they not given any?: 'I was overwhelmed by far too much information. . .' to: 'I knew nothing compared to now!!!'.

The research results

This project found that nobody gets everything. Some people had parts of best practice. Some had no aspects of best practice.

How respondents found out about their Usher syndrome diagnosis

Eye specialist – 58%
Informed by two sources ie, Eye Specialist and Parent – 30%
I found out for myself – 11%
Parent – 11%
Family member – 2%
I was told little bits about my Usher over a period of time – 6%
Social services – 5%
Cochlear implant assessment – 2%
I do not remember when I found out – 5%

Respondents' Usher type

Type 1 – 21%
Type 2 – 55%
Type 3 – 12%
Don't Know – 12%

Respondents' age at diagnosis (by Usher type)

Type 1: 7–28 years
Type 2: 9–51 years

Type 3: 15–42 years

Type unknown: 14–28 years

What respondents wanted from diagnosis

Confirm what I knew – 8%
Information about Usher syndrome – 23%

Onward referrals – 8%

Meet others with Usher – 15%

Coping strategies – 9%

Practical advice – 10%

Follow up appointments – 12%

Clear communication – 2%

Time with doctor – 13%

Initial reactions

Initial words used to describe first reactions to receiving the diagnosis of Usher syndrome:

Crying
Shocked

Worried

Anxious

Depressed

Frustrated

Angry

Confused

Numb

Guilty

Fear

Lost

Denial

To summarise

The research phase has helped to identify the needs and provision around diagnosis:

- People want a better understanding of their diagnosis
- Clear needs identified for the future

Annmaree

Not much appears to have changed since Emma's work in 2018. In an Australian survey completed in December 2024, people and families living with Usher syndrome told us that 80% did not receive the information they wanted at diagnosis.

Forty per cent felt they still didn't have all the information they needed and/or wanted.

Key themes emerged that were similar to those who had other causes for their dual sensory impairment-deafblindness:

1. There's an information desert in a time of information overload: accessible-to-the-individual information is missing, or people are not being directed to services and resources.
2. Communication, especially with healthcare providers, remains a burning concern.
3. People and families wanted practical advice on how to live a good life, and they wanted to hear from people on the Usher journey, not just from professionals and practitioners.

4. People with Usher syndrome wanted support on the grief journey, which extends lifelong beyond the diagnosis.

So, from my research, the message is clear: health and social care systems and staff need to do better with:

- Accessible information
- Better communication
- Funded peer support programmes
- Stable funding for Usher syndrome (and dual sensory impairment-deafblindness) support organisations
- Credentialing of support workers, communication guides and other service providers so that the complexities and challenges of living with Usher syndrome are understood
- More research funding to continue to grow the knowledge base around Usher syndrome

Diagnosis disclosure

The three authors represent different diagnostic eras and approaches. Early diagnostic pathways mean that younger people and children may receive the knowledge early. Meanwhile, the majority of adults, especially older adults, have had very different journeys to diagnosis.

Annmaree

My diagnosis came after multiple ophthalmologists told me I was just a clumsy person and to eat more carrots. I had spent almost 10 years not being able to label my experiences of poor night vision, not seeing garbage bins in the way when driving and falling off jetties I hadn't seen the end of into bodies of water.

At diagnosis, the conveyance of information was non-existent, no offers of support or referrals were made and it was a traumatic experience. I then entered a long period of denial (not happening, not happening, not happening), which did two things: (1) denial enabled me to get a career happening, but (2) denial meant I didn't prepare for getting worse. I should have started to learn sign language and Braille then.

As a result of my diagnosis at 23 years, my brother who also had hearing loss had his eyes checked before the onset of symptoms. This diagnosis was also most unwelcome for him.

But being diagnosed gave me the name and some linguistic tools to explain my experiences and symptoms. And I am grateful for that. As my sight worsened and then experiencing further loss of hearing, it was a small comfort to have a diagnostic and prognostic perspective to process it all. This doesn't mean it was easy. Usher syndrome, above all, requires a lifelong adjustment to constant changes in competencies and increased social, emotional and physical support needs. So my lived experience says it is important to know about Usher syndrome so that you aren't ever left wondering why you can't see some things clearly or play some sports well. It is important that anyone with Usher syndrome, child or adult, be given the resources and information to process what is happening or about to happen.

Emma

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.

In 1992, prompted by a 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.

Chloe

It all started when I was 13, at what I thought would be a routine appointment at the optician's. I went in expecting the usual eye test, but instead the optician noticed something unusual. She explained gently that it could be retinitis pigmentosa, and advised us to seek further checks at the hospital. I remember sitting there, stunned, didn't fully understand the words, but I could feel the seriousness in her tone.

We were referred from one hospital to another, until finally we arrived at Moorfields. There, the diagnosis was confirmed: I did have retinitis pigmentosa. That news alone was difficult to take in, but during the process the doctors also realised I was deaf, and they recommended genetic testing.

For years, my family and I believed my hearing loss was the result of medication I had been given as a baby. My mum had septicaemia while pregnant with me, and because I was born early, I was treated with strong medication usually used for meningitis. The side effects included hearing loss as well as a loss of taste and smell. It seemed like the obvious explanation, so we never questioned it.

When the genetic test results came back, though, we were completely shocked. I wasn't just dealing with two separate issues, I was diagnosed with Usher syndrome type 1D. In that moment, everything we thought we knew was turned upside down.

The shock of that day has stayed with me ever since. What began as a simple trip to the optician changed the way I understood myself, my health and my future. Now, at 25, I've turned my diagnosis into purpose: I've made incredible connections, become a mentor for Usher Kids UK and now serve as president of the International Federation of Hard of Hearing Young People (IFHOHYP).

Emily

My son was young enough to be unaware of the heartache the Usher syndrome diagnosis took on my husband and I when he was just 3.5 years old. It wasn't until the following year at the International Usher Syndrome Symposium in Boston that another parent asked, 'when do you plan on telling him?' And my answer was, he kind of already knew. He had come to understand that his ears didn't work like the rest of his family, hence the need for the cochlear implants. And we just let it be known from the very beginning that we had also learned that his eyes didn't work the same as everyone else's either. Did he understand what we were telling him? Probably not. But each eye appointment gave us another opportunity to tell him that his eyes are different, and we needed to keep getting them checked by the eye doctor. I was grateful at the time to shield him in some respects to the shock and grief and loss that I was experiencing. I remember thinking how much more difficult it would be to have this diagnosis when he was older and having much more understanding of the significance of the diagnosis.

When he began to struggle with his night vision, again, it was another opportunity for us to have age-appropriate conversations with him about his eyes and how they work differently to

most people's eyes. Today, at 15, he has no recollection of when he learned about his Usher syndrome diagnosis and says, 'I've just always known it was part of me and never really knew any different.'

How and when to tell: A guide for parents

The following information was developed by UsherKids Australia and is used with permission:

Having a conversation with your child about their Usher syndrome diagnosis can feel overwhelming, especially as it involves discussing both hearing and vision loss. However, open and honest communication can provide your child with the understanding and confidence they need to navigate their condition. This guide offers practical tips to help parents and carers approach these conversations with care and empathy.

Give yourself time

Your child's Usher syndrome diagnosis will likely bring on a range of intense emotions in you and questions. Give yourself time to work through these before talking to your child. The more relaxed you are when discussing their diagnosis, the better it will be for them. Seek additional support from a mental health professional if you need it and talking to other parents who have navigated this may also be beneficial.

Tailor it to your child

Every child is unique, and so is the way they process information about their condition. You know your child best – their

personality, strengths and the ways they handle change. Use this understanding as a guide to determine the best approach for discussing Usher syndrome.

Younger children

Focus on simple explanations about how hearing aids or glasses (if they have them) can help them. For example, 'Your eyes and ears work a little differently, and sometimes they might need extra help.'

Older children

Discuss how the condition might impact their future, while emphasising support systems, available resources, strengths and solutions. Be guided by their questions in the information you cover and allow them to ask questions at their own pace. Car rides are good opportunities to chat, but don't be surprised if teens don't want to talk about it.

Discussing the genetic aspects of Usher syndrome with your child should be handled with sensitivity, clarity and reassurance. The goal is to provide information in an age-appropriate way that fosters understanding without causing unnecessary fear or guilt.

WHO should have the conversation?

Parents or carers who are actively involved in the child's care are usually best suited for these conversations. However, audiologists, ophthalmologists or trusted educators may be able to provide additional support and expertise. If you feel unsure about where to start, consult your child's healthcare team for guidance.

WHEN is the right time?

The most appropriate time to discuss an Usher syndrome diagnosis with your child depends on their age, emotional readiness and how they are experiencing symptoms. The goal is to provide honest, age-appropriate information while ensuring they feel supported and empowered. There is no 'perfect' single moment – these discussions need to evolve over time. A great opportunity is when your child begins noticing changes in their hearing or vision, asks questions or faces challenges, such as difficulty seeing in low light conditions or participating in conversations. Early, proactive discussions can prepare them for what to expect and reduce feelings of fear or uncertainty.

If your child is undergoing tests or seeing specialists, they may wonder why. Explain the purpose of these visits in a way that feels empowering rather than scary. 'We're going to see a doctor who knows a lot about how ears/eyes work. They can help us understand more about what you need to make things easier for you.'

Focus on what's relevant now

If your child is experiencing difficulty hearing, start with how their hearing aids or cochlear implants can help them hear better. Gradually introduce discussions about vision if and when it becomes necessary. Be led by their questions and provide simple answers. It doesn't need to be a long-structured conversation. After giving them new information, pause and let them process that, then wait for any questions. Often, what you have said is enough and they'll let you know if they need more information.

Be honest but hopeful

Explain that while Usher syndrome can present challenges, there are tools and strategies that can help them thrive. Share real-life examples of people with Usher syndrome who have accomplished great things.

Use analogies and visuals

For younger children, analogies like 'Your hearing aids are like superpowers for your ears' can make complex concepts more relatable.

Avoid vague reassurances

Terms like 'everything will be fine' might offer unrealistic expectations to your child. Instead, offer realistic hope by focusing on the tools and resources that will help them navigate their condition. For example, explain how regular check-ups and assistive devices can make a big difference in their daily lives. If you don't know the answer to any of their questions, say, 'I'm not sure but we can find out.'

Help your child to develop confidence and independence

It is important to focus on your children's strengths, like their creativity, problem-solving skills or determination. These qualities can help them handle challenges with confidence. Using tools like mobility training, hearing support or technology can also make them more independent. For example, soundfield systems in classrooms or apps with pictures and visual cues can make learning easier and more fun.

Helping children with Usher syndrome meet others with similar experiences can also be extremely valuable. This lets them know they are not alone and that there are others like them.

Let them participate in discussions about their support options and encourage their input on how they want to talk about Usher syndrome with friends, teachers or teammates.

Support children with Usher syndrome every step of the way

Navigating an Usher syndrome diagnosis is a journey that involves both challenges and opportunities for growth. By having regular, open, honest and supportive conversations, you empower your child to embrace their condition with confidence. Remember, you don't have to do this alone – your child's healthcare team and community resources are here to support you.

Ongoing challenges

When disability is mistaken for incapacity

The experiences below highlight how deeply entrenched misconceptions about disability and parenting have been, and in some cases, continue to be, within health and social care systems. While policies and safeguards have improved, these accounts remind us that attitudes do not always change at the same pace as legislation.

Emma

February 2008 I gave birth to my daughter, Lucybelle, and in Autumn of that year I went to Norway to attend the Deafblind International Conference to deliver a workshop for Usher parents.

I collected a lot of data from those becoming parents and indeed grandparents and information was shared about the kinds of support available and attitudes at the time. As my daughter was still very small and I was on maternity leave at the time, my husband accompanied me to look after Lucybelle and I could still feed and comfort her during breaks. I was approached by a social worker who worked in the north of England who questioned whether the child I was holding was my own. When I confirmed that it was my daughter, she said to me that she was supporting a lady with Usher who is grieving after being forcibly sterilised under advice from a doctor that she should not bear children, so that she wouldn't be able to pass on her Usher Syndrome. It was such a harrowing thing to hear and it haunted me for a long time afterwards, that this was given as a medical view and she had undergone such trauma. When I became a mother I found it such a rewarding experience. As a young mum, I had a lot of support around me. My own mother visited often in those early days, giving her support to myself and my husband as we navigated being new parents. My mother told me of a day when she had come to the house to support and had answered the door to a children's social worker. She didn't tell me exactly what she told the social worker, but that she refused entry and shut the door without letting her in. I found out later that on a previous appointment with a midwife when I was heavily pregnant and without my husband or an interpreter with me, the midwife had made assumptions about my ability to be a mother and had raised concerns with social services about whether I was fit to become a parent, and this is what the visit was. It was years later that I found myself supporting a soon-to-be mother with Usher who was dealing

with the same situation, who, on finding out the pregnant lady and her husband both had Usher, had reported to social services, who had plans to remove the baby from them when it was born, deeming them both unfit to be parents because of their Usher Syndrome. It was an extremely emotional time for the family, as well as myself, and very distressing for them throughout. Legal advice had to be sought and the process became a long fight with legality over the concerns coming in to question. At the end of the battle, the parents were able to keep their child and raise them by themselves, with support in place. Going through a pregnancy and experiencing the joy of growing a family is a wonderous thing and the joy and excitement of this time was robbed of this family. Having Usher does not stop you being able to have and care for a child. Families should always be supported and can do all of these things with the right support in place.

The following two chapters canvas the complexities for parents and children of an Usher syndrome diagnosis and especially consider the psychosocial impacts. Chapter 6 considers those with Usher syndrome who are also parents and caregivers.

From chapters 10 to 14, we explore how health and social care can support children, adults and families living with Usher syndrome in the key areas identified by research and lived experience.

5
Parenting 1: Complexities of parenting children with Usher syndrome

Emily

Parenting a child with Usher syndrome is a journey defined by both challenge and resilience. As a leader of a parent-led support group, I have come to understand that the insights from families' experiences are an essential element for health-care professionals and students seeking to provide meaningful, patient-centred care. The narratives shared by parents, siblings and individuals with Usher syndrome highlight not only the medical and functional impacts of the condition, but also the emotional, social and systemic burdens that accompany it. Integrating these perspectives into clinical education fosters empathy, enhances service delivery and highlights opportunities for improving care.

This chapter draws on my experience as a parent, current research and living experience narratives from within the Usher community. We explore the complexities of parenting children with Usher syndrome today compared with decades ago, including the psychosocial impacts, sibling dynamics, advocacy burdens and practical considerations such as schooling and driving.

Historical context: Parenting children with Usher syndrome

Thirty to forty years ago, the experience of parenting a child with Usher syndrome was markedly different. The hearing loss aspect of Usher syndrome, although typically present at birth, was often diagnosed much later, as universal newborn hearing screening programmes were yet to be introduced. Diana, mother to Claire and Emma, both with Usher syndrome now in their 50s, suspected for some time that Claire was not responding to sounds before being diagnosed as severely deaf at 1-year-old. The reliance on astute parents noticing signs such as developmental delays or a lack of responsiveness in their babies placed an enormous burden on them, particularly if it was their first child, as they did not have other children for comparison. If concerns weren't voiced early, or if subtle signs were overlooked, feelings of guilt and blame could often follow. The result was often slower access to intervention, leaving children at a disadvantage in catching up with speech and language development.

Luckily for Claire, and later her younger sister Emma, it was their older brother singing nursery rhymes that prompted their mum to notice the difference in how her new baby was responding. Intervention and therapy has also changed dramatically over the

decades, with Diana recalling a steep learning curve that included daily lessons of teaching Claire to lip-read with constant tuition and help from the family. Juggling the therapy with family life was not easy, a sentiment that is still echoed by parents today.

Diana also recalls 'the body worn hearing aid, and headphones during Claire's lip-reading lessons with amplified sounds'.

The subsequent diagnosis of Usher syndrome was also much later than today, typically occurring in adolescence once the progressive vision loss caused by retinitis pigmentosa became evident. Prior to diagnosis, parents frequently navigated subtle, unexplained signs: tripping over the family cat, difficulty orienting in new environments, or challenges with balance, often labeling their child as clumsy without understanding the underlying cause. The medical literature, as portrayed by Vernon (1969), framed Usher syndrome in a language that was reflective of the times, emphasising psychosis, intellectual impairment, mental retardation and a focus on eradication of the condition through genetic counseling aimed at reducing incidences. Families were positioned as passive recipients of healthcare rather than partners in their care, and children were certainly not recognised for their capacity to contribute to society.

In contrast, today the diagnosis of Usher syndrome occurs much earlier, often in the newborn period through diagnostic audiology following universal newborn hearing screening and genetic testing, enabling families to engage proactively with healthcare providers. The Usher diagnosis is also often well before the functional impact of vision loss, meaning parents now learn and adapt

alongside their young child's developing sensory and communication needs. This shift has transformed the role of the parent from one of delayed understanding to an active advocate and decision-maker. But with this comes additional burdens, as parents now navigate complex choices regarding communication modes, such as sign language, auditory-verbal therapy, cochlear implantation and assistive technologies, all while coming to terms with the emotional implications of the dual sensory loss.

Early intervention and the parental role

Just as the diagnosis of Usher syndrome has progressed over the decades, so too has the importance and evolution of early intervention programmes. Our access to technology has certainly improved, including those used to test, diagnose and manage hearing loss. Another notable change over the past few decades is the advice given to parents based on the outcomes of children who are aided or implanted. No longer is an auditory verbal or sign language approach mutually exclusive, with the literature showing that both spoken and sign languages can be developed simultaneously, giving the child exposure to rich language in the early years. But the fragmented services and lack of co-ordinated, multidisciplinary approaches is still causing unnecessary burdens on parents today. The mental health and well-being of parents, particularly mothers, of children with disability are well below that of parents with typically developing children (Bourke-Taylor et al., 2010). Parents need to become experts themselves and are constantly educating healthcare professionals involved in the care of their children about the condition and how it impacts

in various settings. This can heavily influence the developmental trajectory of children.

Parents assume the role of the Usher 'expert', and are constantly exhausted by the continual explanation of the condition, taking on the role of case manager, co-ordinating multiple providers and advocating for supports. This is often described as 'advocacy fatigue', a form of emotional and physical exhaustion arising from repeated efforts to educate, negotiate and secure appropriate accommodations within healthcare, educational and social systems (Basas, 2015), and I am yet to meet a parent of a child with Usher syndrome, whether their child is 6 or 60, who doesn't resonate with this.

As the condition is considered rare, it is often poorly understood and the parent is constantly in a position of educating others, whether that be in their places of education or employment, health professionals or government systems. Also, the invisible nature of Usher syndrome leaves parents having to really prove the impact of the dual sensory loss and continually fight for necessary accommodations and modifications.

This advocacy fatigue would be greatly reduced if there were better policies and procedures in place as well as increased awareness and empathy from healthcare professionals. Understanding the burden carried by parents can be alleviated in small ways by assisting them to fill out forms and advocate on their behalf, writing reports, initiating communication with other healthcare professionals working with the family and just going the extra mile for families to try and avoid some of that advocacy burnout and fatigue.

Even perceived simple tasks like having a sleepover with friends, attending a school camp or sporting event can involve extraordinary planning, advocacy and emotional cost. The 'disability tax' further reinforces this parental burden, a term derived from the constant financial, logistical and emotional costs incurred from assistive technology needs, co-ordinating appointments and navigating complex health and education systems. To illustrate the point further, following is a list of healthcare professionals typically involved in the care of a child with Usher syndrome:

- GP (General Practitioner) for referrals for other specialists
- Developmental Paediatrician monitoring gross motor milestone development
- Audiologist may become involved with the early fitting of hearing aids
- Ear, Nose and Throat specialist if the child has type 1 and the family are considering cochlear implantation
- Speech pathologist
- Ophthalmologist
- Orthoptist
- Optometrist (glasses may be needed for a refractive error which is not related to Usher syndrome, just a benefit of early and regular monitoring!)
- Physiotherapist, particularly those with type 1 to compensate for the vestibular dysfunction and reduce safety risks of trips and falls
- Occupational therapist
- Orientation and Mobility Specialists
- Teacher of the Deaf
- Teacher of the Vision Impaired

Co-ordinating the care of all of these specialists as well as having a baby is a huge burden on parents! No wonder we are all exhausted!!

Psychological and social impact on parents

In previous decades, parents were often left to navigate the enormous challenges of raising a child with Usher syndrome privately, in an era where disability and mental health were rarely discussed openly. Today, with the rise of the internet, growing peer networks and support organisations, parents have the opportunity to connect more than ever. Yet, although psychosocial research acknowledges the parental burden of raising a child with Usher syndrome, we still have some way to go to reduce the isolation often described by parents today. The *Theory of Possible Selves* (Markus & Nurius, 1986) provides a valuable framework for understanding the psychosocial experiences of parents when their child is diagnosed with Usher syndrome.

The theory describes three forms of 'possible selves', which are future-oriented aspects of the self-concept. *Hoped-for selves* represent who we want to become, or in this context, the hopes and dreams parents hold for who their child might become. *Expected selves* reflect the realistic outcomes parents believe their child will achieve, while *feared selves* capture the worries or anxieties about who their child might become. These imagined selves shape motivation, identity development and coping strategies.

For parents, this process can be complex and emotionally challenging. There may be some initial reshaping of the possible

selves after the initial hearing loss diagnosis. Parents then may come to terms with the identity of their child as deaf/hearing impaired/hard of hearing, only to have to reshape this identity when vision loss is added to the diagnosis. This dual sensory loss forces a rethinking of the possible selves, both for the child and the family. Parents may grapple with questions such as: What will the future look like for my child? Will my child be able to live independently? Will they experience isolation? Where will they belong if they do not fully identify with either the Deaf/hard-of-hearing community or the blind community?

This uncertainty creates a sense of being in limbo: the child has useful vision now, but we know further losses are likely. Alongside this comes anticipatory grief: grieving the losses yet to come and mourning the version of the child we had once hoped for. Parents may need to revisit some of their earlier dreams and adjust to a new, evolving vision of their child's future. But how can the parent do this when there is so much uncertainty about the future and the progression of Usher syndrome?

This process of adapting and re-establishing identity is ongoing, as parents navigate the challenges of supporting a child with a degenerative condition while managing their own emotional responses and expectations. It is important to consider whether we focus on the limitations Usher syndrome presents or on adaptive strengths to help develop resilience to cope with future uncertainties.

Siblings and family dynamics

The experience of siblings, both with and without Usher syndrome, adds another complex dimension to parenting and the family dynamic. There are clear practical implications of having

more children, but the emotional demands intensify if there is more than one child with Usher syndrome in the same family. Parents describe the compounding impact of co-ordinating multiple medical appointments, educational supports and therapy sessions while also managing the progressive nature of Usher syndrome in more than one child. Despite this, siblings with Usher syndrome can also provide mutual understanding and support, sharing strategies, empathy and companionship in ways that parents value deeply.

For siblings without Usher syndrome, the experience can be complex and emotionally fraught. They may experience guilt for not being affected as well as frustration or jealousy at the additional attention their sibling receives. The additional attention is inevitable though, as parents juggle the demands of appointments, managing therapies or co-ordinating school supports. Do we drag the siblings along and include them in the therapy? Or do we leave them being cared for by family and friends? Siblings often assume informal caregiving roles, including communication facilitation or advocacy, which can influence their own development and emotional well-being. Usher syndrome can certainly shift family dynamics, create subtle but persistent tensions and requires careful navigation to ensure that all children feel supported, valued and included.

Navigating school and education

Selecting and entering school are significant milestones for children with Usher syndrome, and can bring about mixed emotions for both parents and the child. There is an element of 'letting go' from the parent, often after years of being the case co-ordinator,

advocate and therapist for their child. None of this stops, mind you, but trusting educators to provide safety and support while also advocating for accommodations that address the combined effects of hearing, vision and vestibular losses just adds another layer to the journey.

For Diana, mum to Claire and Emma, she recalls the difficulties both girls faced with communication with their hearing friends at school. They both attended an oral/aural school, boarding four nights a week, but ultimately, sign language became their main method of communication.

For our family, attending a school in our local community was a priority for us. Although we had a school with a hearing unit (specialist support from teachers of the deaf based at a mainstream school) about 15 kilometres away, the practicalities of having two children at two different schools made me exhausted just thinking about it. The local school was supported by a visiting teacher of the deaf and then a visiting teacher of the vision-impaired in the later years.

Teachers of the deaf and teachers of the vision-impaired can provide specialised support, including orientation and mobility training, communication adaptations and classroom modifications. Their frequency of visits to the school can vary, so parents still play a critical role in liaising with school staff, setting goals and advocating for functional support.

Specialist schools for the deaf or vision-impaired may offer additional expertise such as classroom instruction in both spoken and sign language, but often don't fully address the dual sensory needs of students with Usher syndrome.

Adolescence and independence

The teenage years introduce new challenges as night and peripheral vision losses may become more apparent. At a time when young people are exploring identity formation, autonomy and social participation, those with Usher syndrome are contending with the realities of progressive sensory loss. They must navigate peer relationships, self-advocacy and disclosure of their condition.

Just as parents experience a shifting 'possible selves' (Markus & Nurius, 1986) following their child's diagnosis, teenagers are also beginning to develop their own ideas around who they might become. The way information about Usher syndrome is discussed during this time plays a critical role in shaping this process. Delayed or sudden disclosure, as was common just a few decades ago, creates anger and confusion in this developing identity as the imagined future self no longer aligns with reality. Therefore, it is vital that we give our children the opportunity to develop a healthy sense of identity.

The early disclosure of Usher syndrome gives them the opportunity for a gradual integration of a realistic yet positive *valued hoped-for self*. How might they use assistive technology to ensure access to the school curriculum? How can we modify a community sport to ensure they don't have to give up the activities they love? Having access to mentors and role models with Usher syndrome is important during this period, particularly those who work, study, travel and thrive socially. It allows both parents and children to see positive, attainable futures and expand *possible selves*. But, sometimes, it can be

confronting to see people who are further along in their vision loss journey for both teens and their parents. Encountering someone using a cane or a guide dog, for example, can initially feel daunting, as it symbolises a stage of progression they have not yet come to terms with. However, there is an opportunity to reframe these moments and see what these vision tools are allowing the individual to maintain, such as travelling safely, confidently and on their own terms. For many young people (and parents!), the realisation about the possible future selves can be one of adaptation, agency and ultimately, empowerment.

This is where mentors and role models with Usher syndrome can be really helpful. By connecting with others who are further progressed in both their vision loss and their adaptation and acceptance of Usher syndrome, there is much to gain for everyone. By learning about career choices, studying at university or overseas travel, young people are able to comprehend that, although life may include some obstacles and barriers, there is no reason that passions and dreams need to be abandoned just because of an Usher syndrome diagnosis.

Driving with Usher syndrome

For teens with Usher syndrome, the progressive nature of vision loss will require early conversations about safe transportation options and the consideration of driving. Parents play a critical role in these discussions, exploring alternative transport, adaptive technologies and strategies to maintain independence while prioritising safety. Driving discussions and ultimate decisions are not straightforward: they highlight the very nature of

adolescence, where growing independence and safety highlight the ongoing burden of anticipatory planning.

The following information was produced by UsherKids Australia and has been adapted with permission. Although it is targeted towards parents, healthcare professionals involved in the care of children and young people with Usher syndrome will benefit from an understanding of the issues and complexities individuals with Usher syndrome and their parents are faced with each and every day.

Helping teens navigate life is difficult enough, so when the extra challenge of Usher syndrome exists, it's essential to understand the unique considerations surrounding driving and transportation.

Understanding the licensing process

Individuals with conditions affecting vision, including Usher syndrome, are usually required to disclose their condition when applying for a driver's license in their local area.

Disclosing the condition may trigger a medical review process, where the individual may be required to undergo assessment by an ophthalmologist or optometrist. These specialists will conduct tests based on the guidelines in the local country to assess the individual's vision and overall fitness to drive.

Outcome of medical review

Based on the report provided by the ophthalmologist or optometrist, the local driving authority will make a decision regarding the individual's eligibility for a driver's license. This decision may include granting a license with specific conditions, such as

wearing glasses or refraining from night-time driving or denying the issuance of a license altogether.

Ongoing vision monitoring

There are currently no specific guidelines on how frequently individuals with retinitis pigmentosa or similar conditions should undergo vision rechecks. The frequency of vision monitoring is typically determined on a case-by-case basis, guided by the individual's needs and recommendations from healthcare professionals and will vary in different jurisdictions.

Navigating the process together

When bringing up the subject of driving with teenagers who have Usher syndrome, it's essential for parents to approach the conversation with sensitivity and understanding. As you and your teen navigate the process of considering applying for a driver's license, it's important to stay informed about the guidelines and procedures outlined by your state or country authority. Maintain open communication with healthcare professionals to ensure that your teen's vision is regularly monitored and any changes are addressed promptly.

Here are some recommendations for discussing driving with teens who have Usher syndrome:

Provide Information: Begin by educating your teen about Usher syndrome and its implications, including the progressive nature of the condition and its impact on vision, balance and hearing. Help them understand the potential challenges associated with driving, such as decreased night vision and peripheral vision loss.

Discuss Safety Concerns: Emphasise the importance of safety for both the teen with Usher syndrome and others on the road. Highlight the potential risks and dangers associated with driving with limited vision, including difficulty in detecting obstacles, pedestrians and other vehicles.

Encourage Open Communication: Create a supportive environment where your teen feels comfortable discussing their concerns and feelings about driving. Encourage them to ask questions and express any anxieties they may have about relinquishing the idea of driving.

Explore Alternative Transportation Options: Explore alternative transportation options that can help your teen maintain independence and mobility without compromising safety. This could include public transportation, ridesharing services, walking, biking or relying on family and friends for transportation.

Discuss Adaptive Technologies: Explore adaptive technologies and resources that can help individuals with visual impairments navigate their surroundings more safely. This may include talking about assistive devices, mobility training programmes and orientation and mobility specialists who can provide guidance and support.

Highlight Positive Aspects: While discussing the limitations associated with driving, also emphasise the numerous opportunities and activities your teen can still engage in despite not driving. Encourage them to focus on their strengths, interests and abilities beyond driving.

Seek Professional Guidance: Consider involving healthcare professionals, vision specialists or counsellors who have experience working with individuals with Usher syndrome. They

can provide valuable insights, guidance and support tailored to your teen's specific needs and circumstances.

Encourage Independence: Help your teen explore ways to maintain independence and autonomy in their daily lives, even if they are unable to drive. Encourage them to develop problem-solving skills, self-advocacy and confidence in navigating the world with Usher syndrome.

Ultimately, the decision about driving with Usher syndrome should prioritise safety and well-being while also considering the individual's desires and capabilities. By approaching the topic with empathy, understanding and open communication, parents can support their teens in making informed decisions about driving and explore alternative avenues for independence and mobility. By understanding the licensing process in your local area and advocating for safe and responsible transportation options, you can support your teen in maintaining independence and mobility while prioritising their safety and well-being.

Conclusion

We warned you Usher syndrome could be complex and challenging, but if well informed, well resourced and well supported, then there are more upsides than downsides, to borrow from Chapter 8. Chapter 6 continues the discussion on parents of Usher syndrome children but also explores the complexities of being a parent who lives with Usher syndrome themselves.

6
Parenting 2: Parenting, psychosocial needs and perspectives

Introduction

We continue to look here at lived experiences of parenting a child with Usher syndrome, and later, of being a parent with Usher syndrome. There are complexities and challenges with both but also great capability and much success. First, Emily shares aspects of her parenting journey and corollary research. Then, Emma and Annmaree will share some lived experiences of both lows and highs. Interspersed with all of this are some perspectives generated by current research.

Emily

As a leader of a national support organisation for families of children diagnosed with Usher syndrome in Australia, I am privileged to speak with many families in their early days post-diagnosis. As they grapple with making sense of this new future for their child, the question I get asked the most is 'when?' 'When will my child's

eyesight start to deteriorate' and 'when will there be a treatment to stop this deterioration?'

And while clinical research aims to answer both of these questions, we need to be cautious in how we approach treatments and cures. I had the same questions myself back in 2014 when I visited the International Usher Syndrome Symposium in Boston shortly after my son was diagnosed. The message from researchers was clear: in 'single digit years' there will be an effective treatment. But fast forward 11 years, and there is still so much work to be done. Clinical trials take years, and we need to help those with Usher syndrome and their families live well *today*. While clinical research into Usher syndrome is progressing and providing great promise, we can't sit with our lives on hold while we wait for science to one day provide a treatment. We need to know where to go for trusted support and we need to know how to live in a world that is not designed for those without their hearing or vision.

My colleagues, Emma and Annmaree, and I have been involved in various psychosocial research projects in our collective efforts in understanding the full impact of Usher syndrome on individuals and their families. These valuable insights guide the development of supports and health policies that are instrumental in enhancing the overall quality of life for those with Usher syndrome. I discuss a few of the research projects my own living experience has helped to shape, including:

- the clinical awareness of Usher syndrome among allied health professionals;
- the support needs of parents of young children with Usher syndrome;

- the experiences and support needs of youth with Usher syndrome;
- the research priorities of the inherited retinal disease (IRD) community in Australia; and
- a pilot multidisciplinary clinic.

The clinical awareness of Usher syndrome among allied health professionals

In collaboration with the University of Melbourne, Ayton et al. (2023) aimed to investigate the awareness levels of allied healthcare professionals. You would be hard-pressed to find anyone with Usher syndrome that hasn't experienced being more knowledgeable about their condition than the healthcare professionals themselves. Having to constantly explain the condition and how it impacts in various settings is exhausting. Findings from the survey of optometrists, orthoptists and audiologists revealed gaps in understanding crucial aspects of Usher syndrome, particularly the vestibular dysfunction and the need for interdisciplinary management, all of which help us focus our advocacy efforts for clinical education (including being involved in publications like this book!).

The support needs of parents of young children with Usher syndrome

Johansen et al. (2024) explored the support needs of parents of young children with Usher syndrome in Australia and the results were of no surprise to me as a parent myself. The qualitative thematic analysis unveiled four central themes: social needs, informational needs, practical needs, and emotional needs.

Social needs

Parents of children with Usher syndrome face isolation due to the rarity of the condition. They struggle to connect with others facing similar challenges and often feel alone in managing their child's complex needs. They described substantial benefits to connecting with other parents of children with Usher syndrome and identified support groups to be useful in facilitating such connections.

Informational needs

Parents reported dissatisfaction with the level of knowledge among medical professionals regarding Usher syndrome. They felt that they are required to educate healthcare providers about the condition and appropriate management strategies.

Practical needs

Managing Usher syndrome requires co-ordination among various healthcare professionals, scheduling numerous appointments and implementing treatment plans. Parents often need to navigate complex healthcare systems and seek funding for therapies. Case co-ordination and collaborative care can remove burden from parents.

Emotional needs

Parents experience significant emotional burden upon receiving the diagnosis of Usher syndrome in their child. They face grief, uncertainty about the future and pressure to engage in early intervention strategies to maximise their child's development potential.

The study's findings shed light on the challenges faced by parents of children with Usher syndrome and highlight the urgent need for tailored support services. Understanding the support needs of parents can lead to the development of evidence-based recommendations, effective tools and programmes to assist this vulnerable population. I believe the publication of this research was a significant step towards enhancing the quality of life for families impacted by Usher syndrome globally. Hollie and I were no longer just two parents sharing our experiences in an attempt to initiate change; we had contributed to a credible, peer-reviewed body of evidence that validated the experiences of families like ours and could be used to influence practice, policy and service delivery.

Both of these research projects highlighted the fragmented healthcare system that focusses separately on hearing and vision, and this 'siloed' approach was increasing the burden of parents.

The experiences and support needs of youth with Usher syndrome

This project is the natural progression of the parental support needs project, and it aims to give voice to young people (11–26-year olds) with Usher syndrome globally. At the time of writing, data analysis is underway, with the results to be published in 2026. It is a qualitative study using interpretive description and includes young people from Australia, New Zealand, the USA and UK. Data were collected through focus groups at an Usher syndrome youth camp in Australia in July 2025.

One early emerging theme is a *sense of burden* that having Usher syndrome has on our young people, including being a burden

on those around them – their family, friends and support staff at school, and having difficulties accepting help. This is a push/pull of wanting to fit in and not stand out, but also needing some accommodations and modifications to be able to keep up with their peers. But the burden also comes from having to constantly explain themselves and their needs to those around them: to healthcare professionals who are unfamiliar with the condition; to teachers who may underestimate the impacts; and to friends and family who struggle to understand. This burden and fatigue, on top of living with dual sensory loss, deeply shapes the lived experiences of our youth, and can have a significant impact on their identity during such a transformative period in their lives. We look forward to sharing the published results once available.

Research priorities of the inherited retinal disease community in Australia

There are a number of research projects occurring alongside clinical developments that aim to ensure laboratory advancements are in line with what the community actually wants. This increase in the living experience narratives in clinical research is evidenced by the Priority Setting Partnership (Robertson et al., 2025), a project that developed the top ten research priorities of the inherited retinal disease community in Australia. The project brought together a range of stakeholders, including those with living experience, their caregivers, parents, community organisations, healthcare professionals and researchers. Although the top priority remains to develop treatment to either prevent vision loss or restore vision, this was balanced by the need for improved psychosocial support for individuals diagnosed with an inherited

retinal disorder and their families and to address the gaps in current health service provisions.

A multidisciplinary clinic approach

The research, lived experience of children and families, and the advocacy of UsherKids Australia has driven the establishment of the Melbourne Collaborative Sensory Clinic, a pilot study between the University of Melbourne and the Australian College of Optometry. This pilot study brings together a range of allied health professionals in a multisensory clinic setting to determine if a co-ordinated care approach can improve outcomes for both children and adults with Usher syndrome and their families. This takes patients off public hospital wait lists, with expedited pathways back into the system for those requiring specialist clinicians, including geneticists, genetic counselling, ophthalmology, and more.

The clinic model also includes allied health student placements to embed collaborative care into our future clinicians and remove that 'Usher syndrome? Never heard of it' experience that so many people with Usher syndrome experience.

As I am the clinic manager, having that link between the clinic and community supports is central from connecting the results from the clinical assessments to tailoring community resources to meet the specific sensory needs identified in the clinic. This includes assisting the patient to apply for NDIS (National Disability Insurance Scheme; Australia's disability support system); connecting them to orientation and mobility supports to manage the deteriorating vision; and applying for financial supports, including blind pensions and public transport concessions.

Parenting as well as living with Usher syndrome

Being a parent with Usher syndrome is to be on unsteady ground. The progressive nature means that often you have just adjusted to one deterioration when the next is upon you. Some have said that 'Usher syndrome rules the family'. From our own lived experiences and those of peers, finding ways to enjoy family life that both accommodate the sensory losses and enable the family to have adventures and fun will prosper. Lived experience research on parenting with Usher syndrome is scant. From wider parenting with deafblindness literature, common themes include lack of supports, lack of information, broad lack of awareness of complexities and challenges and fragmented health and social care systems.

Facilitating factors for people and their families with Usher syndrome include:

- accessible information
- Readily available human and financial supports
- Social connectedness
- Attention to the psycho-emotional health of partners and children as well as the person with the Usher syndrome
- Peer support: engagement with other parents, spouses, siblings and carers, and sharing experiences, support and knowledge (Intini et al., 2022; Anderzen-Carlsson et al., 2024; Ehn et al., 2019).

A collaborative care model is also important for getting families and parents the individual supports and resources they need, when they need them. Usher syndrome, as you have read

repeatedly, is progressive and this will mean there will be periods of crisis; stretches, sometimes prolonged, of adjustment; and a need to pivot to newer methods of communication at times.

Parents need to be able to advocate for themselves and their families, and to communicate goals and needs to health and social care professionals, supporters and service providers as well as their own wider social networks.

Annmaree

Being a parent with Usher syndrome is a lot of swings and roundabouts (both literally and figuratively). I have three children and a host of godchildren who spent a lot of time in Casa Annmaree. What I have learned is that:

1. I would do things differently knowing what I know now! But we still did an okay job and all three are kind, compassionate and supportive big people now. We don't have to be perfect, just safe and loving and having occasional fun.
2. While I believe the children are not my carers, they are however intrinsically bound into the Usher journey with me. I strongly believe that as a parent with Usher, you cannot keep them separate. The children need to be part of your communication, orientation and mobility team. If I fall over or down or into something deep, sharp or steep, I can get injured. That then makes our family situation worse. So, the kids are part of the team but not full-time care workers.
3. Children can be creative problem solvers too: my youngest developed a tactile communication method for when we were out and about, as he realised I couldn't hear when there was traffic noise around us. So, he made everyone

learn how to let me know to stop, turn and tread carefully over uneven ground or when there are upcoming stairs. This made a huge difference to our ability to go on adventures and outings.

4. Learn orientation and mobility with the pram and stroller. Use the information imparted by the front wheels to tell you when the gutter starts and begins again when crossing a road, when there are steps (wheels aren't on a firm surface) and when you are on a different surface (e.g., grass, footpath, road etc.). I had a great teacher from Guide Dogs for this.
5. It is frustrating to have to deal with single sense organisations. I have occasionally been lucky to get trainers knowledgeable in dual sensory impairments, but mostly I have not. We are exhausted from teaching people the complexities of Usher syndrome.
6. You can't change yourself and your family by trying to do everything yourself. Get help. Tell social service providers your Usher makes it difficult (not dangerous or impossible) to be a parent sometimes. We hired a nanny to do the things I found tricky, like supervising three children in a park with one degree of vision. This meant the kids got to go out to play and were supervised by someone with good eyesight and hearing.
7. I also had more rules than some other mothers, such as no eating and running around the house or garden; we all sat down together. This way, if someone choked or had an issue, I was at hand and could help. I worried I wouldn't hear them if they were all over the place. I also went outside and sat in the garden when they played in

the garden. This way, I was nearby if there was a fall, a fight, an injury or an irate neighbour. When out and about, the kids had to let me know explicitly if they left my side, so that I knew where they were. My youngest was distractible and often went off without letting anyone know where he was going. We have loads of family stories about his adventures as a 3- and 4-year-old running off. After these experiences, I would explain why they couldn't run off and would institute a time out or a favourite toy confiscated for a week. This sounds harsh, but it was a reality for our family to keep us all safer; we needed these rules.

8. I also had a rule about rules; if I fell, it was NO ONE'S fault, we would give thanks for all the times I didn't fall (which outnumber the falls by thousands and thousands).
9. Sometimes, we all have to remember that it's stressful having to keep an eye on a parent with sensory loss/es, and relax together with takeaway if a day has been difficult, or ice cream breaks or fun activities to relieve tension.
10. Build and maintain connections with supportive people for the children. Make sure they each have trusted adults who will help them, listen to their frustrations and worries and act when needed. One example is: I found it really tedious to watch cartoons or animated movies, as I couldn't lip-read and there were no subtitles or captions. The children's uncles, aunties, grandparents and family friends would take them so that I didn't have to, and this fostered bonds the children have in adulthood as well.
11. We travelled a lot as my sight diminished, even if it cost a lot, because there were so many memories I wanted to make with everyone while I could. Now, I know that even

with small amounts of hearing and tiny tunnel vision, I can still travel: smells, taste and touch become important conduits of information about different parts of the world.

12. Use social supports that are available; don't be too proud. You don't need to keep your children at home to help you, send them out into the world knowing how to ask for help and how to receive it graciously.
13. Have an emergency plan: I woke up one morning unable to see out of my (best) eye. I had a retinal hole. I was able to contact a friend who came to help us manage this emergency, to get me to the ophthalmologist and the hospital.
14. Above all, it is a joy being a parent, even with Usher syndrome, and the kids have many funny stories about mishaps, miscommunications and missteps. Humour is essential in any family journey with Usher syndrome.

Emma

As a person with Usher, I faced additional difficulties and challenges when becoming a mother. Taking my two young children to water babies classes was not easy, navigating the class and being in the pool with two children is enough in itself with their being so close in age. I also joined a mother and baby group. This was fundamental for my Lucybelle and Thomas, who became immersed in a hearing environment with hearing mothers and children to engage with, but it was difficult for me to engage and communicate with the other mothers. I did always have a communicator guide with me, who could help to fill any gaps I had missed. If I took the children out shopping, I would use a double buggy, which was useful, but rather cumbersome once you also add in a guide dog. People always offered their assistance.

I remember a bad experience of crossing the road at a zebra crossing where a van failed to stop. The van caught the front wheels of the buggy and jerked it to one side out of my grip. I was very lucky it wasn't worse, and no one was hurt. The driver fled the scene, but many people rushed to help.

Another time sitting on the tram with my baby son, he reached out his hand – as young babies do – and grabbed onto something. It turned out that the emergency cord had been pulled. The driver came over and was very angry with me, although I didn't know what was going on. I remember seeing someone walk past me just before the driver came, so I wonder how my son could have been strong enough to pull it, or whether we just received the blame!

There were often incidents when out, as public transport and public shopping centres are busy places to navigate. I remember one time when I was pregnant and got on the train with my guide dog. I chose to sit near the window but when I went to sit down, I was on somebody's lap! I hadn't seen the man sitting there in a black coat. We had to laugh about the situation! Another time I was out shopping with Thomas. While I was looking at clothes, he disappeared. I searched the shop, running up the stairs and asking a shop assistant for help. We found Thomas, upstairs with another staff member who had lifted him up and sat with him while they chatted, realising he was lost. It turns out he had been drawn to the escalators and made his way up alone. Luckily all was well.

Although my children could sign and understood my needs, I never wanted to rely on them to help me. I remember difficult

times taking the children to and from school during the winter, labouring to see when it became dark early. Children don't want to slow down and walk closely alongside their parents, so this was always a struggle. Now that they are teenagers, they are very independent. Lucybelle is studying for her A levels (Advanced levels, a prerequisite for University study in England) and Thomas is completing his GCSEs (General Certificate of Secondary Education). I look back and am proud of what they are achieving. It is a reflection on me and my husband's parenting skills that gives me much fulfillment. Lucybelle is now learning to drive. This is an exciting time for her; I'm sad that I'm not able to teach her, but I'm proud of her.

I had fun raising my children, taking them to art exhibitions, museums, swimming, children's activities and other places, as parents do. Having Usher wasn't a barrier to us enjoying our lives together as a family. We use sign language to communicate at home, and taught our children, who are both hearing, to sign from a very young age as my husband Clive is deaf and we are both BSL (British Sign Language) users. As the children have grown and my vision has deteriorated, we also occasionally use hands-on BSL and regularly use social haptics too to communicate more easily.

For example, if a family member wanted to let me know they were just taking the dog out for a walk, they would use the BSL sign for walk and 'draw' walking footsteps up my arm or on my back to quickly convey this information. Likewise, to let me know they are just leaving the room to go to the toilet, they can draw a line up and down on my back or arm and I am aware that they are going to the toilet and will be back. Another example is if

my husband asks me for a tea or coffee; this can be conveyed quickly by drawing a 'T' or using a closed fist handshape in a circular movement on the arm for coffee. Social haptics are useful as information can be conveyed quickly through touch without the need to interrupt focus, say when working or doing the washing up, but a meaningful exchange of information can still take place quickly and effectively.

Conclusion

Health and social care professionals and practitioners can do so much to facilitate psychosocial health and well-being for parents of children with Usher syndrome, and parents with Usher syndrome and their families by:

- Providing information in accessible-to-the-individual formats
- Expediting founding and continued growth of peer support programmes
- Assisting parents and families to navigate health and social care systems
- Taking a person-centred approach in practice and service delivery.

Chapter 7 features the voices of siblings, spouses, offspring and supporters.

7
Siblings, spouses, offspring and supporters

Introduction

In earlier chapters, we briefly alluded to the wider family and social networks that live with people who have Usher syndrome. Their lives and experiences, too, are complex and challenging at times, while simultaneously being barely visible on policy, health and social care radars. We give you stories of the living realities for some who live in the homes or work in the houses of people with Usher: one perspective is the sibling, the next is the partner, then the child of a parent with Usher syndrome and the last is a supporter and friend. Thank you to Khloe, George, Tom, Jess and Liz.

Siblings

The research acknowledges that:

- Siblings of deafblind people experience psychological and social challenges, including feelings of neglect, resentment, embarrassment, jealousy, anxiety, exclusion and insecurity

- Siblings experience communication difficulties
- Siblings take on significant responsibilities within the family (Arcous et al., 2024)

Khloe

'Okay Khloe, we're taking Angus to the doctors again, you're going to stay at Lisa and James' for a while', said my parents for the 10th time that week. It became a routine. 'Yay, aunty Lisa and uncle James!' I'd do anything to be that ignorant and unaware of the real world around me again. Things aren't like that anymore.

I was just 2 years old frantically running around the house, making all the noise toddlers do, my loud squeals reflected against the walls, while my newborn baby brother was nearby completely unbothered. It was as if he was in a different world, unaware of a single thing going on around him. No reaction to anything from a quiet whisper to an echoing yell.

Four years later, on a cool evening in July 2013, was when it all began. I was 6 at the time and my little brother Angus was 3. Clouds moved in quickly as the rain started to pour against the roof, and a dreary feeling began to sour my family. My parents received the call. The call everyone is afraid of. The call that changes lives. My brother had just gone through genetic testing and the results were supposed to come back anytime now. By this point, my parents knew that it wasn't going to be good news. They could sense something wasn't quite right from the beginning. And on that blue night, the news that was bound to come, came. Angus was diagnosed with Usher syndrome, a rare genetic disease that causes both hearing and vision loss. A wave of guilt quickly filled the house. I was still too young to

fully understand what it meant, but anyone could pick up that after that night, things weren't ever going to be the same.

My childhood completely flipped. I was now spending days and nights with my aunty and uncle or any babysitter my parents could find while they were going all over the place taking Angus to a bajillion different doctors' appointments. My aunty Lisa and uncle James became my second parents, and I spent just as much time with them as I did with my own. Each day, I would sit in the back of the car on the way to their house with my eyes locked out the window watching cars go by, confused, disconnected and oblivious, but always glued on the thought of my little brother and if he was okay.

The first few years of this new thing that we were all adjusting to was hell. The doctors informed my parents that Angus's vision was going to get worse and worse every year until he reaches the age of 10, when he is going to go completely blind. He is fully deaf, but he has cochlear implants that provide him with a sense of sound. But there is nothing you can do to overcome blindness. You can't buy new eyes.

Obviously, it hit very hard, and immediately my parents panicked. They started planning holiday trips, booking random activities and signing up for anything that came to mind without any second thought. Initially, what my parents were trying to do was stressfully squeeze in as many significant experiences as possible in the short time frame before Angus would turn 10 years old. To me, this appeared totally awesome. All of a sudden, I was so excited and overwhelmed with all this new stuff we were doing. Helicopter rides over the city, driving in fire engines, catching planes to go on holidays. It was every kid's dream.

But now, I realise that in reality, this was just simply an unhealthy coping mechanism for my Mum and Dad. They were sheltering their grief and anger by doing these fun things as a family, while they were aching with pain inside, refusing to show it to anyone.

As I started to mature more, I could understand how critical Angus's condition really was and saw the struggles that it brought to my family as a whole. I realised that it would influence everyone, but in differing ways. And the older I got, the more I could notice it tackling me mentally.

Throughout the end of primary school and beginning of high school, I would constantly be torn between trying or not trying at all. I hated having to choose between the guilt and the pressure. Every day was a choice. *Do I make my brother happy, or do I make my parents happy?* I was very hesitant around my own personal achievements because I felt like I was stealing it from my brother in a way. I never wanted to make him feel like he's not enough because he doesn't have all the abilities or opportunities that I have in front of me. Yet, the other half of me stressed about *what will mum and dad think?* Although the pressure wasn't placed there by them, I always put pressure on myself to perform well in everything I did because I have 'no excuse' to fail. I felt like I had to be successful because Angus can't exactly be successful in the same ways that I can be.

Endless nights were spent silently crying myself to sleep in fear. In fear of pleading for more attention. I never admitted it, but a small part of me was jealous. Angus spent so much more time with Mum and Dad than me, and all of our friends and family would constantly check up on how he was doing or how my

parents were doing. 'Oh, your Mum and Dad must be going through so much', 'your poor brother', 'is Angus okay?' But the sister is never recognised. Sometimes, it felt like I was forgotten. Like I was a problem. Like I was just a waste of space and that I made it more difficult for everyone. I grew tired of being unseen. It was not that I was invisible. It was that people had become used to not seeing me. *And if no one sees me, am I really there at all?*

The future worried me too. Will he stop being able to see? Will he forget what I look like? Will my kids be like him? Who's going to take care of him when he's older? My breaths were sharp and frantic, my mind running a million miles an hour, heart pounding and visions raged. This process repeated itself often. I had immeasurable questions. I was mentally fatigued. It felt unfair. I didn't understand why this was happening.

But there is always time. Time allows us to learn. Time allows us to overcome. Time allows us to change and mature. Over time, my family started to put the pieces together and acknowledge that it will never stop being hard. That ups and downs are a part of life. That if we stick together and trust each other, then nothing else is important.

I began to feel more included and got the love and support I needed from my parents. They noticed me struggling, they helped me and always asked what I needed. I began to feel more important and my self-esteem bloomed. I still have occasional worries and concerns, but accept it as a usual thing being the older sister of someone with a disability, which has also taught me independence. I still spend a lot of time with my aunty and uncle, who have grown to be very valuable people to me that I feel safe around.

Our family used to spend all of the time worrying about Angus. Every day was a timer ticking for his future. But he is thriving in life. Currently, my brother is 13 years old and has good eyesight. His vision is still getting worse, we are unsure of what will happen, and he has picked up some balance issues with his disease, but he is in a very good place. He plays cricket, has lots of friends and is doing well in his first year of high school.

We have learnt to try to shift our focusses to accepting it, embracing it and learning to be flexible around it. The process was long, and we are still learning more about Angus's condition each day. Relying on one another has been so crucial, and without providing support to each other equally, we know that we would not be where we are today.

I would be delusional to say that it's going to be a smooth road from here, but I know that if our family has made it this far, then we can overcome anything if we stick together.

Partners and family members

Spouses/partners and other unpaid carers experience health and well-being consequences that are rarely studied and lack visibility in policy and service settings. They must navigate their own emotional distress due to the progressive nature of the condition, and take on increasing care and communication workloads, such as doing all the driving and supporting their partners' communication in family and social events and exchanges.

- Non-deafblind partners often compensate for the losses caused by deafblindness, placing their own needs second.
- Families and partners may need tangible support to manage everyday life (e.g. finances, practical tasks; Björk, 2020).

George

Living with someone with Usher syndrome is a challenge at times. The first 20 years less so than the last 10 as my wife's hearing and vision deteriorated. Life was great, we had three children and some help for some things; some things she simply couldn't do like driving, and some she shouldn't like internet shopping. She once ordered six apples in an online shop, but 60 kilogrammes arrived. But because she could still communicate and be communicated with, it wasn't as stressful as later on. I should point out here that the various organisations that were supposed to be helpful weren't; we didn't have an organisation for adults that understood the combination of hearing and sight loss. And we had an extended family with no oxygen left, because we weren't the only ones dealing with Usher.

Communication became difficult and she lost her ability to lip-read (and I didn't help this by growing a beard which made it harder). The worsening of her condition also made learning a visual sign language impossible for us, since she couldn't see them. She really struggled with tactile sign language too, as she was learning the signs herself but had to also learn them in reverse action as she had her hands on the person signing opposite her. Looking back, I see that we were latecomers to sign language and that we should have started learning as a family much, much earlier.

We also tried lots of tech stuff: microphone pens, special hearing aids and so on, but none of it helped, especially in group conversations. With the availability of iPhones and iPads, we could use speech to text, but for a long while they weren't very accurate.

Today, it's better, but from our point of view, technology fails miserably.

The simplest of questions could become a screaming match:

Do you want a cup of tea dear?
What?
(louder) Do you want a cup of tea?
What what?
(louder still) DO YOU WANT A CUP OF TEA
(louder and frustratedly) What what are you saying?
(screaming) TEA TEA TEA
(very loudly) Stop screaming at me
(very loudly) I'm not screaming at you. I GIVE UP.

Both of us were frustrated and cross. Something had to change. Our colleagues Russ and Riitta in Finland introduced us to using the body to send quick messages. We started to learn. We had a meeting with Russ and Riitta and then some lessons on zoom. We have a sign language teacher and also learned fingerspelling and sign shortcuts. We made up some touch messages for things we needed, such as watching television. My wife can't tell the goodies from the baddies or who is doing what by the subtitles alone, so we have a system for this now. B1 is baddie one, G2 is goodie two, FF means they are fighting or KK is kissing. We haven't worked out what to do when there's a double-cross and B1 becomes G4.

Slowly, our communication began to improve, but I am not without fault. I am slow to learn. Ros helped us consolidate and to learn sign shortcuts to help with longer sentences. My wife would say some of the trouble is because my spelling is atrocious,

so using the manual alphabet takes longer than it should. That is probably true.

Sometimes, what seems the smallest of social-haptic messages can bring a lot of pleasure. My wife has really missed the joys of knowing what is on her plate and where it is located. I draw a circle to represent the plate on her back sometimes, or more usually, on her palm. I tell her verbally or with some fingerspelling that the chicken is in the middle and the various vegetables are located around this. In a restaurant setting, it just looks like I'm being romantic and holding her hand.

One of the lessons I can give to people in our situation is this: try to come to touch messaging earlier, before you really need it, so you can get a pattern and a rhythm going. From our perspective, the more methods you have that can fit together, the better. We use social-haptic communication (SHC), manual alphabet and sign shortcuts, and lots of made-up ones that our family, friends and my wife's support workers know. They all go together. You might think we don't need them all, and that we can just keep using speech to text or hearing aids or whatever. But with Usher, you won't always have the hearing and sight you have right now. And there are some places you can't use the tech, such as swimming pools and operating theatres. There are other times when all the tech doesn't work: device fails, no internet, run out of battery, the device is slow to respond, you forgot the charger and more; so, having another method to change to is important. Learn early, you will thank us later. These communication methods are like a bridge between full-on sign language and having nothing. We've had more than 30 years together and are gunning for 30 more.

Children of parents with Usher syndrome

The offspring of parents with deafblindness, including those with Usher syndrome, describe to researchers some of the consequences of living in an Usher family:

- Children explained that going anywhere required a lot of planning and time due to their deafblind parent not being able to drive.
- Children tended to dislike the attention of having a parent with deafblindness, strangers pointing or commenting when walking down the street and a cane or guide dog also drawing attention.
- Some children stated they had to take on responsibilities and an adult role to manage their everyday life due to the parents' deafblindness; for example, cooking, cleaning, laundry, and washing dishes. Children talked about how this made it difficult to relate to other children, who would play when at home.
- Children talked about having difficulty managing schoolwork and homework compared to their peers who had more assistance from their parents.
- Children also talked about financial strain as a result of one parent being unable to work.
- Children described feelings of frustration due to miscommunication or the need for extra patience.
- In addition, children expressed increased empathy and compassion as a result of having a parent with deafblindness.

(Huus et al., 2022)

Tom

I didn't know my mum was different from other mums until I was about seven. I realised then that she didn't drive, and all my friends' mothers drove everywhere. We caught buses and taxis or my dad drove us. We also got a nanny to take mum and me to sport and sport practice and any other places we needed to go. But our dog worked it out within a week of living with us – Tarquin knows there was no point leaping up at her, barking or getting in her way. Now, he follows behind and waits until she is sitting down before putting his paws on her lap for a scratch behind his ears.

I worry about her a lot, but she works hard even when the people she works with are mean. We get help at home from the National Disability Insurance Scheme, Australia's disability support system (NDIS), and that is great as mum has become a terrible cook the blinder she gets, but it feels like sometimes our house is full of people who don't really belong there. There is a good side though: if I say something stupid or rude, she goes, 'what did you say?' and I say, 'ahh, nothing.'

Over the years, the things we do together have had to change: we used to go to movies together, but mum can't see now, so even with the subtitles taking up half the screen of a TV she can't understand what is going on. Now, we learn sign language and alphabets. This helps us a lot. COVID-19 was good because all the restaurants did meals you could get delivered or make at home, and that meant we didn't have to go to a noisy place with mum where she couldn't hear anyone. We have kept doing that after lockdown, and that is nice. We laugh a lot as some things mum misunderstands are really funny.

Supporters and carers

Professor Tom Shakespeare, a social scientist from the UK, talks about how people with disability can form not just service agreements with support workers, but also social connectedness. There is very little research that we could find on these relationships. The body of research in the Usher syndrome space indicates significant support and paid care needs for children, families, adults and older people living with Usher syndrome. Older people ageing with Usher syndrome can have both established strategies for coping, and the emergence of additional disability and conditions that impact upon existing ways of adjusting.

One study exploring the needs of carers of people with rare syndromes (of which Usher syndrome is one) reported that carers experienced:

- Suboptimal interactions with healthcare providers;
- Largely absent psycho-emotional support for themselves;
- Financial stress;
- Low levels of awareness in the community about their caring role and the rare condition/s.

  (McMullan et al., 2022)

Jess

I'm 29 and have been working with a deafblind person for 3 years. In this time, we have had lows that are usually caused by other people or government departments as well as highs.

Low Moments

Other people are disrespectful or discriminatory (and it happens a lot more than it should, sometimes almost every day).

We witness this and this upsets us too. For example, when I'm sighted-guiding them on a narrow path and someone walks straight into them instead of walking around. Or when we're in a meeting and someone continues to talk over them because they can't hear that others are speaking.

Other people do not always provide accessible means, resulting in a greater workload for me. My job is complex and there is a lot of multitasking involved. When others require me do the extra work to make everything accessible, it means less time and energy supporting the immediate needs and priorities of the deafblind person.

Other people have also been dismissive of my role and contributions. Sometimes, I'm seen as 'just a support worker'. My role as a support worker is funded through the NDIS. Being dependent on NDIS funding impacts my job security, if there are changes to the plan or less funding is available.

High moments

The relationship that I have built with this deafblind person has been extremely enriching and rewarding. We have a little community that I get to be a part of. Because of all the support workers and the deafblind person working so closely together, we have developed strong connections and good friendships.

Learning new practical skills, like fingerspelling and social-haptic communication, has been enjoyable as well. It's great to have a skill that allows you to facilitate communication and have good conversations, especially when we are in social settings and get to subtly make funny comments to each other that no one else can hear.

Other useful skills like learning to be very adaptable and staying calm under pressure (like when technology breaks or things don't go as planned) have been very helpful to practice and apply to other parts of life. It's nice to know that if things go wrong, we always end up making the best of it.

A support worker's job is so varied that every day brings something interesting to tackle. Work is never boring!

The highs of this job absolutely win out over the lows. The satisfaction, sense of community, friendships with colleagues and all of the new and interesting work we get to do together make my work enjoyable and incredibly rewarding.

Liz

My carers journey was born out of a very long and deep friendship. While my friend Louise had obvious hearing and vision impairment when I first met her, she could see some things and hear a little. As time has gone on, the vision and hearing loss has increased to almost no vision and no hearing.

Since we were very good friends and our children were also friends, as she needed more help, she would ask me if I could give her a hand, and I was slowly thrust into becoming a guide/signer/carer.

I have worked in creative industries all my life. I am a disorganised, overly verbose, mad woman. New methods of communication needed to be succinct, clear and concise.

It was a relatively slow ease into where we are now, and in the early days, I would make up little signals to help her understand the environment and so forth.

In my other work, I have to be very visual, meet challenges of the most unusual sorts, be intuitive and creative. I am sure this has actually helped in many ways with our communication and sight guiding.

I've also had to be extremely observant. In the early days, observation was the most important tool I had. Because I knew Louise so well, I was able to see when she perhaps didn't understand something that was going on. I was able to know what she was looking for and I could assist when needed. I was also very aware of the fact that while I might be able to help and reform information that Louise needed, she is an extremely competent, intelligent, independent woman who likes to be able to achieve things by herself.

In the early days, I largely helped out with everything from fixing knitting boo-boos, getting lunch, going on outings and just sitting in the background and being present when meetings were going on. If things were terribly misunderstood, I was able to step in and clear the information.

COVID-19 struck about the same time that Louise's hearing and vision loss became greater. And, of course, there was no contact with anybody. There was difficult communication for everybody, but particularly people with vision and hearing loss. Because of masks and face screens and isolation, requests to meet your doctor or your accountant or your lawyer on Zoom became impossible for the dual sensory impaired.

One of the first tactics I realised was useful was to wear bright red lipstick. Louise had been able to lip-read and I could turn up looking like the wreck of the Hesperus but have full lipstick on.

I was often asked, 'why are you wearing lipstick?' My response was and still is, 'if it only adds one percent of information sharing for somebody who isn't able to get all of the communication, that's one extra little piece of the jigsaw puzzle that they get.' The lippy coupled with vocal tonality changes, environment changes, lighting changes and so on and so forth just gave me layer upon layer upon layer of information transfer, in the hope that Louise would receive the maximum amount of information that she could.

As time goes on, these techniques have changed with Louise's changing condition. The tactics that I adopted earlier, I still believe in, and everybody's circumstances change sometimes daily. I still believe that observation is one of the greatest tools I have in my tool kit. Looking at the environment, you may need to change the lighting (e.g. pull the blinds down or turn lights off). Some days, Louise is having a bad day and the light really irritates, other days it's okay.

During the COVID-19 lockdown, I had already learnt a little bit of social haptics via Zoom. I sat on my trusty iPad and took the social haptics course with Riitta Lahtinen and Russ Palmer. This has been exceedingly helpful, but it also gave me the knowledge that this is a new technique for me, but developed by Riitta, and it's an ever-growing technique. It's said that it's designed for the individuals to work out some of the best ways of environmental messaging that give the most distinct information.

I've never been particularly good with technology; this was a very swift learning curve for me on some technology. Thankfully, some of the girls we work with are very clever in that department. They used to scribe a lot of our meetings which we would put into a

Word document, change to white on black print and enlarge the print, so Louise had access to the information as time moved on. This has become slightly redundant now, because there is a lot more closed captioning services, and more recently AI voice to text which we use regularly. AI voice to text is not perfect. Usually in a meeting, I sit just behind Louise and I watch what it says on the screen. When AI says very incorrect things, I can let Louise know what is actually being said. Louise and her brilliant mind will work most of it out, but occasionally I just have to tap her on the shoulder and go 'no, no, no, that's not what the other person is saying' or 'I think you haven't received the question correctly so your answer has gone on the wrong tangent.' We need to start again. Of course, we also have our own special little haptic sign that tells Louise that the technology we are working with is having a fit. I still talk clearly and choose a tonality that I know is best for the pitch that Louise can hear, fully knowing she can hear very little of what I say; however, harking back to the 'one percent of information is better than nothing'. I give haptics signs when we need to. I use the manual alphabet, but at this point in time neither Louise nor I are very good at hand over hand sign. My standard Auslan (Australian Sign Language) signing is very basic and Louise's hands are not little clouds resting lightly on top of my hands as I sign; it becomes very difficult and very confusing. But we muddle along somehow.

Positives of living in an Usher family

To conclude this chapter, we don't want you to feel that life in a family with Usher syndrome is a predominately negative

event. Despite the challenges and adjustments needed, we, that is, Annmaree, Emily and Emma, know that the upside is a great side. We are capable, and we enjoy our lives and relationships. It's important that policy and services, health and social care support all of us with Usher syndrome and those who live with us.

Recent research affirms there are positives to a family living with Usher syndrome too. In an article about children stating the positives of having a parent with deafblindness, children said:

- They enjoy the benefits of skipping the queue at amusement parks and going to places where they could meet other children in similar circumstances.
- Children said that guide dogs and canes were helpful as they relieved some of the 'burden' from family members and enabled their deafblind parent to be more independent.
- Children talked about wanting to help their parent and having a willingness to protect their parent from harm, for example, making sure to pack away their toys so their parent didn't trip.

  (Huus et al., 2022)

Conclusion

We have looked at over the past three chapters the impact of Usher syndrome in a variety of familial and support relationships. What is clear is that everyone needs good information, better resources and guaranteed skilled supports. The next two chapters interrogate the health and well-being impacts of Usher syndrome broadly. Chapter 9 looks at Charles Bonnet syndrome

(benign visual hallucinations in people with low vision of any cause and intact brain function) in particular, as those with the combination of hearing loss with low vision have higher rates of occurrence. After these, we move on to better communication and health and well-being solutions.

8
Complexities of health and well-being for people with Usher syndrome, their families and carers

Introduction

Usher syndrome complicates health and well-being in multiple ways, impacting carers, partners, siblings and children of people with Usher. To live with Usher syndrome is to live with health and well-being complexities that shift over a person's life stages. When presented 'en masse', these challenges can look overwhelming, unconquerable even, as they're not widely recognised or supported.

Yet the difficulties – health threats, carer burdens and communication challenges – can be mitigated with planning, lifelong learning, good support networks and an inclusive society. To address health and well-being vulnerabilities, we need health

and social care students, professionals and practitioners to be able communicators, advocators and team players. We need you to recognise that not only is each Usher journey different, but individual goals and needs differ, and differ over the person with Usher's lifetime. For our health and autonomy, we need to be engaged in decisions on all aspects of our personal and social lives. (There is more on ameliorating risks to health and well-being in Chapter 13.)

Health and well-being challenges for individuals with Usher syndrome

We remind readers that Usher syndrome is a spectrum of diverse residual senses owing to the degeneration that occurs over time in vision for all, but hearing and balance are added in for some. This means that health and well-being risks generally tend to increase with sensory loss/es and decrease in the uncommon event of improved sensory function (e.g. a cochlear implant, cataract removal or improved assistive technology). Health and well-being risks may also increase with the addition of other conditions: ones that are additive to the existing sensory loss/es (e.g. retinal detachment or further hearing loss from recurrent infections). Risk may also increase in the presence of non-sensory conditions such as other illnesses and injuries. In those cases, the dual sensory losses of Usher syndrome may complicate function and management. Also, adding complexity to life, health and well-being with Usher syndrome are intersectional influences such as social disadvantage, ableism, gender, indigeneity, racism and ageism.

Challenges fall into the following broad areas: diagnostic, risk of communication failure, health literacy, social barriers, physical health threats, visual hallucinations, delirium, psychological impacts, cognitive risks, public health threats, ageing with Usher syndrome, complexities of multiple disability and conditions and the ever-present but rarely considered fatigue of degenerative dual sensory loss together with its disruptions.

Diagnosis: Challenges, changes and consequences

Diagnosis of Usher syndrome can occur at different life stages from prenatal, birth, school age and even adulthood. The life stage at which a person receives an Usher syndrome diagnosis will impact the services, supports received and life adjustments needed (e.g. educational support, changes in working life and social adjustments). In the previous chapters, we mention some of the challenges for parents and families when a baby, or very young child, receives a newborn screening test indicating hearing loss, followed by genetic confirmation of Usher syndrome. This occurs in the absence of visual symptoms and signs. When a person is not-yet-diagnosed and older, however, they can notice vision problems that they might initially dismiss: possible night blindness, an increase in accidents, bumping into things and also recurring falls, deterioration of driving skills and new struggles with reading.

In the UK, those with Usher type 1, generally profoundly deaf and often British Sign Language (BSL) users are diagnosed earlier than those with Usher type 2 or 3. People with Usher type 2 or 3 are typically diagnosed much later in life. As not all who received

a diagnosis of hearing loss from newborn hearing screening will have received genetic testing, presentations with vision loss in the first, second or later decades of life will continue to present diagnostic challenges.

When an adult is diagnosed with Usher, things may become difficult and uncertain, especially if a person diagnosed is already in a relationship, marriage and/or on a career path. Diagnosis can be deemed a time of grief requiring emotional processing. As mentioned, because Usher syndrome is not a stable condition, constant changes in hearing and vision (and sometimes balance) occur over a lifetime, each bringing new demands on personal relationships, families and work or educational settings.

On average, people with Usher seek out support and advice every 5–7 years at crisis points, when a person is no longer able to manage as before with their Usher and needs further support or adaptations. These changes in vision and hearing have an immense effect on a person's communication, access to information, getting around and independence. This can lead to increased isolation from the world and people around them.

Diagnosis is an emotional journey

Receiving an Usher diagnosis can be distressing.

If we have been through an emotional journey when we receive a diagnosis of Deaf/have a hearing loss, a second diagnosis confirming dual sensory impairment can be devastating. A diagnosis of hearing loss is a time of processing, adjusting and learning how to deal with the changes of expectations. After this period of adjustment, a second diagnosis can be a blow, knowing we

have to process again and readjust. Not knowing what to expect and imagining what it would be like to not see or hear is scary. Common reactions are shock, upset, denial, anger, depression and resentment. Sometimes, people blame each other, that is, parents receiving diagnosis for their child. These emotions are normal for people with Usher and their families.

Every person is unique. Their experience of Usher is unique. Even siblings with Usher syndrome can have markedly different experiences and life attitudes. There is no time frame for 'accepting' we – or our child/family member – have Usher and for living with Usher. It can take months, or longer, to accept the diagnosis. However, the more information a person with Usher syndrome has, the more understanding they have, and the more we can feel in control of life with Usher. It is then that support can be put in place to help us continue to live a fulfilling life on our terms.

For some parents of children with Usher, guilt can be difficult to let go of, carrying heavy emotions for some time before accepting the Usher diagnosis. Other parents want to move on living ordinary lives and to watch their children with Usher grow up leading their own lives.

Usher is a forever-changing condition.

Communication risks

Usher syndrome is principally a disability of communication, access to information and orientation/mobility/safety.

We don't get good health literacy and shared decision-making in health if there is poor communication and a lack of accessible

information. Research shows that effective healthcare communication results in:

- Shorter hospital stays.
- Fewer hospital readmissions.
- Reduced emergency room visits.
- Closer treatment adherence.
- More effective medical follow-up.
- Less unnecessary diagnostic testing.
- Fewer medication misadventures.
- Improved healthcare outcomes.
- Reduced healthcare expenditure.
- Good communication is strongly associated with better patient experiences, and better patient experiences are linked to better health and well-being outcomes.

Poor communication especially is associated with poorer health and well-being outcomes such as:

- Increased risk to patients of negative events and misadventures, patients living with communication disability (such as Usher syndrome) experience higher rates of poor outcomes in hospitals.
- Deafness and/or blindness add considerably to risk in hospitals (Bartlett et al., 2008).
- People living with communication disability are not often able to question, criticise or report episodes of poor communication and care because the support needed to report these experiences is the same that is needed for good communication.
- Invisible data – we don't know as much as we need to know because patient experience evaluations are rarely in accessible forms.

- Communication failures underpin nearly all complaints about healthcare in Australia.
- Non-compliant consent – where consent forms and processes are inaccessible to the patient with hearing and/or vision loss. In a 2021 study, 0% of people with Usher syndrome and 0% of people with deafblindness received an accessible-to-them consent form (Watharow, 2021).
- 'Not knowing what is going on' for patients with hearing and vision loss can result in negative feelings, anxiety, panic, stress and poorer health outcomes.
- Negative communication experiences can lead to avoided or delayed health services.
- Huddle et al. (2016) reported, 'addressing sensory impairments could have substantial implications for public health given that these impairments are highly prevalent, undertreated, and amenable to treatment with established, low-to-no-risk rehabilitative interventions' (p. 1736).

Health literacy for people with Usher syndrome

Reduced access to information and communication challenges impact health literacy. Health literacy refers to an individual's ability to obtain, process and understand basic health information and services so they can make informed decisions (US Department of Health and Human Services, Office of Disease Prevention and Health Promotion, n.d.). For those with dual sensory impairment, barriers to health literacy can be especially pronounced. Consider a typical doctor's appointment: the environment is often crowded, noisy or visually difficult (too glary or too dark for lip-reading). Staff might speak quickly, wear masks

(limiting lip-reading) or present important health forms in tiny, dense print. Any of these factors can severely limit access to critical information.

Low health literacy can have negative impacts:

- Miscommunication of diagnoses and treatments: If you cannot hear clearly or see well, you might misunderstand instructions regarding medication dosages or follow-up tests. This can lead to serious medical errors, such as overdosing on insulin or misunderstanding a needed procedure.
- Delayed care: Frustrations with poor communication or inaccessible environments can drive some individuals to postpone or avoid medical care. This may result in worse outcomes, as untreated health concerns and diseases progress without treatment.
- Stress and anxiety: Not knowing what healthcare professionals are saying, or feeling disoriented in a clinic or hospital, can heighten anxiety and reduce trust in medical professionals.

The following excerpt is from Scene Two of Annmaree's drama *Harms Way*, a play that arose out of verbatim testimonies of people with Usher syndrome.

ROBERT: Where am I?

BETTY enters, and places hearing aids in ROBERT's hands.

BETTY: Put your hearing aids in love. You're in the hospital. The Nat. They say you took too much insulin. Such a fright you gave me. Lying there on the Turkish rug, not moving.

ROBERT: So noisy in here, isn't it?

BETTY reaches out and moves ROBERT's face towards hers and speaks slowly and clearly.

BETTY: You're in hospital. You took too much insulin.

ROBERT: I tried, I did, to do the right thing. I couldn't hear her! The diabetes lady at the hospital last time. I heard her keep saying that I need to take it all seriously, but I didn't hear how much I was supposed to take! Three. Thirteen. Thirty. They all sound the same. But I thought she probably said thirty. The more the better right?

BETTY: (affectionately) Silly old fool. I'll ask if I can stay when the diabetes person comes. Although there are signs everywhere saying visiting is restricted because of another COVID wave.

While the characters are fictional and composite, Robert's and Betty's experience is not rare. Miscommunication can have devastating consequences.

Social health threats

The health of people with Usher syndrome is not only impacted by poor health communication but also by the social structures and environments they live in: the negative attitudes, ableism, microaggressions and discrimination they experience impact psychosocial well-being. Social isolation that can result from poor social support is strongly associated with very poor health. These may be accompanied (and often are) by intersectional associated disadvantages, including gender-based threats.

The World Federation for the Deafblind reported in 2018 that women with deafblindness experienced higher rates of violence than men with deafblindness, and these rates were higher than other disability and higher still from a non-disabled population. Reduced healthcare access and knowledge and the risk of

hospitalisation are but a part of the complexities of health and well-being risks of living with Usher syndrome.

Physical health threats

- Decreased ability to self-manage chronic conditions and seek effective treatment.
- Single-sensory services that do not account for dual impairments, resulting in complexity of needs being misunderstood.
- Higher risk of accidents or misattribution by health staff of symptoms (e.g. stumbling attributed to intoxication rather than balance or vision loss).
- Increased vulnerability to environmental hazards (e.g. not hearing fire alarms or horns).
- Low vision can make food preparation and hygiene difficult, increasing risk of illness.
- Decreased overall quality of life if needs go unmet.

Psychological and cognitive risks

Usher syndrome is well known to be a condition of ongoing losses. These losses in sensory function require psychoemotional adjustment, and many describe living with Usher as being a series of bereavements. In one recent study, two Usher syndrome participants mirrored each other by saying the condition is 'grief, grief, grief' and 'loss, loss, loss' Additionally:

- There can be difficulties diagnosing psychological and psychiatric conditions accurately for people with Usher syndrome. Diagnostic tools and screenings are not validated for dual or even single-sensory loss.
- Mental health assessments may not take into account tactile and visual sign language and other communication needs.

- Misattribution of symptoms may mean misdiagnosis. A classic example is visual hallucinations, which may, in fact, be benign but the person with Usher receives an erroneous diagnosis of psychosis.
- Deafblind people experience reduced access to mental health services and information generally. Sign language users (visual and tactile) particularly experience a lack of linguistic and culturally sensitive services.
- Increased loneliness, social isolation and reduced community participation negatively impact health and well-being, and is a global concern with the World Health Organisation more broadly.
- Increased rates of depression, anxiety and suicidality have been reported in people with Usher syndrome.
- Hearing loss, low vision and social isolation are recognised, often remediable risk factors for cognitive decline in older age; this is what makes good psychosocial support critical for people with sensory losses, including Usher syndrome.
- Delirium (defined in detail in the next paragraph) is common in those with single and dual sensory losses, including Usher syndrome, especially in older age.
- Visual hallucinations are common, and around one-third of those with low vision of any type may experience these. However, awareness and supports are low, resulting in few being diagnosed and consequently reassured as to the benign nature of their experiences. This is known as Charles Bonnet syndrome (detailed in the following chapter). Visual hallucinations are distressing and commonly misattributed to psychiatric or cognitive conditions. It is important that health and social care professionals and practitioners are aware of Charles Bonnet syndrome and offer referrals, investigation, support and reassurance.

Delirium

What does it mean when we see things that aren't there and we are not well? When we are sick or hurt or in pain? Delirium is a state of confusion and decreased awareness that can come on quite suddenly. It is a condition where alteration in attention, awareness, perception and cognition is experienced. Delirium commonly complicates care in the acute setting of hospitalisation after falls, fractures and infections. People with sensory loss/es, such as Usher syndrome, experience higher rates of delirium when ill or injured than people without sensory loss/es. Delirium may be the first sign that something needs emergency attention. In delirium, people may experience:

- Acute confusion
- Disorientation to time and place
- Memory loss or changes, especially short-term memory
- Agitation
- Anxiety
- Behavioural changes
- Poor concentration
- Seeing, hearing or feeling things that aren't there (hallucinations) but believing them to be real

Delirium is unfortunately more common if you are ill, injured, physically stressed and have sensory loss/es. These stressors can be from a fall, an illness, a fever, strong pain relief medicine or an anaesthetic. The brain can be affected for a little bit until there is recovery from the cause. Families and carers should treat this as a medical emergency and seek help as soon as possible. Treatment will vary according to the cause.

Annmaree

My children love to tease me about the time I saw pink fluffy clouds after an operation when I was on strong pain relievers. Apparently, I said to them when they came to visit:

> Oh look! There are pink fluffy clouds on the ceiling.

I was certain these were real. The doctor stopped the medication and the clouds went away. Shame really, as they were quite pretty.

It's important when we are sick and seeing things that aren't real, our families take us to see a doctor and get us sorted out.

Public health threats

Public health emergencies present challenges for people with Usher syndrome due to accessibility barriers, including:

- Difficulty hearing/seeing public health messaging if it's not delivered in accessible formats; this was noted during the COVID-19 pandemic.
- Less likely to be involved in emergency and disaster planning, response and recovery.
- According to the Australian Bureau of Statistics COVID-19 impact survey, one in four carers of people with disability had difficulty providing care (Australian Bureau of Statistics, 2022).
- People with deafblindness reported mixed COVID-19 pandemic experiences, with some feeling more excluded as it seemed everything, including health, moved online. Others reported that working from home meant less stress on the journey to and from work – navigating difficult environments, microaggressions and more.

Growing older

People with Usher syndrome age. The syndrome itself doesn't limit life expectancy but the 'normal' events that happen to everyone, also happen to people with Usher syndrome. These may be harder to navigate with sensory losses, but, alternatively, having adapted to Usher syndrome, what comes next may be managed with equanimity, using skills acquired in life with dual sensory impairments. It is important that health and social carers know that people with Usher syndrome may be carers of their family members and partners. We need to break down the ideas around the binary of the 'cared for' and the 'carers', as people with Usher may be both.

Multiple disability and co-occurring conditions

Usher syndrome is a multiple disability already, but people with it may acquire other injuries, illnesses and chronic conditions. The dual sensory loss means that it can be complex to manage these conditions and navigate healthcare systems, as mentioned earlier.

Emma

In 2011, when my children were only a few years old, we went through a very difficult time. After losing a good friend of mine, who also had Usher, to cancer, I, too, received a cancer diagnosis. Everything then became critical. My life perspective changed, and for the first time, I had thoughts around death. It was a difficult journey in which I was very poorly, life was difficult and I relied on the support of my family. After a year of treatment,

I recovered to the point where I could return to work, but at this time my perspective on Usher had changed. It had become less important; what was most important was living. This was a period of adjustment and recovery for me. When I became well enough, I decided to set up a Deaf Cancer Support Group. The importance of supporting those with Usher through this complicated journey helped me refind my passion.

Annmaree

Everyone I meet in healthcare likes to categorise me as an Usher syndrome patient. But I lost the sight in one eye (and my best eye at that) from an unrelated condition that I was unlucky to get. This means I lost binocular vision and depth perception, which were at times challenging to deal with, on top of what I already had. This new setback prompted us to renovate the bathroom, as I kept misjudging the distance getting in and out of the shower bathtub. This resulted in a few falls and three fractures. So, the bathtub went and a high contrast bathroom was designed and installed.

Having found that the occupational therapists I'd consulted had no expertise in my specific needs, I'm grateful for the Deafblind Ontario Service group as their staff wrote an electronic book *Accessibility Guidelines for Sensory Loss* (2020) about home renovations for people with sensory impairments. So the new bathroom has: motion sensor lighting (no more groping for the switch at two a.m.); non-slip flooring; simple high contrast mixer that is located well away from the shower head so that there is no chance of jumping, slipping or falling when cold water hits you before it has heated up; easy to see – for me – black toilet

seat and rails; and a wall niche so that soap and more are easy to find. Touch wood, no further falls.

Complexities from balance disorder

Balance can be affected in people with Usher, mostly with Usher type 1, although people can notice issues with their balance across all types. This is because we get our sense of balance from different factors:

- Eyesight – what we can see around us
- Hearing – what we can hear around us
- Proprioception – our sense of space, our connection and understanding of where we are and what is around us

This is why a person with Usher will experience a worsening of their balance as their vision and hearing deteriorate. The environment can have a significant impact on someone with Usher and their ability to control their balance. If the terrain is rocky or cobbled, or has many bumps and an uneven surface, the person's balance will appear much worse, as they try and navigate irregular surfaces without the full use of these important senses. To control their balance, people often start walking with a different gait, a wider stance perhaps, they could appear to walk very clunkily or in zigzags. Also, they can reach out to grab something to steady themselves.

Those who have Usher syndrome in addition to vestibular dysfunction have heightened risks of:

- Falls due to loss of balance – injuries may be severe.
- Reduced activity and decreased fitness.

- Depression and anxiety.
- Psychological and cognitive threats.

Fatigue and fluctuation in function

As there is a significant increase in effort for someone with Usher to process the signals in the brain through their residual sight and hearing, fatigue is common. The increased effort on the senses means that the person tires out quickly, even when being supported; so, the condition can cause variable levels of function throughout the day. Lighting plays a role. When lighting conditions inside and outside are not favourable, the ability to complete daily tasks is severely limited. Bright, sunny days can cause issues with glare and reflective surfaces, while darker days make it harder to define objects and differences in floor surfaces such as the gradient and where steps and pavement edges are.

Fatigue syndrome

Fatigue syndrome is a condition where people continue to feel tired quickly after periods of concentration.

It is common for people with Usher to experience fatigue syndrome.

Due to the combined sensory loss of both sight and hearing, extra concentration is needed for everyday tasks and interactions. Intense focus is needed regarding where people with Usher syndrome must look, what they can hear and in trying to process extra information, such as changes to environment, an uneven pavement or a lack of contrasting tones on flooring/

surroundings and light areas and dark areas interfering with what can be seen. There is a constant consciousness of avoiding tripping and bumping into things, reassessing the surroundings and possibly also struggling with poor balance. Looking for simple objects, such as a phone or keys, can lead to fatigue; if it has been moved or is outside of the field of vision even slightly, concentration is needed to rescan the environment and process what is around and where this object may be, even if it is close to hand.

Impact on the family: Living with someone with Usher syndrome

We look at health and well-being for the carers and families of people with Usher syndrome. The complexities extend beyond the individual. No one escapes the challenges (and the joys) of sharing family life with Usher syndrome; partners, parents, children, spouses and siblings all experience stresses and strains that are neither widely recognised nor supported. Few jurisdictions recognise the realities of being in familial relationships and/or caring for people with disability generally. The latest Australian Census has found that one in ten adults are informal carers (unpaid spouse, family members or friends). Informal caring is associated with:

- Decreased physical well-being
- Increased experience of psychological distress
- Deceased access to support
- Decreased time for looking after own needs

These are general observations on carer well-being, but as explored in previous chapters, those who live with someone with Usher syndrome have specific complexities to navigate as well.

Carer well-being

Carers often experience decreased physical well-being, psychological distress and reduced time for self-care.

In Sweden, there is some recognition in law of the needs of the family of people with disability and their support needs. Disability support for carers is enshrined in the 'Law Regulating Support and Service to Persons with Certain Functional Disabilities' (LSS), which provides supplementary support to individuals with significant and long-term functional impairments, including support for family members acting as carers. This law essentially allows carers to access services that help them manage the caregiving responsibilities they have for a disabled person in their family; services such as respite care, counselling and information to help them manage the demands of caregiving. The law also covers children of parents with disabilities, including Usher syndrome, as being entitled to support too. It is our hope that such recognition of family needs is enacted elsewhere.

Conclusion

In closing, Usher syndrome – while presenting significant and ongoing challenges for both individuals and their families with the complex layers of shifting sensory losses, communication hurdles and related health risks – can be addressed through proactive strategies and collaborative frameworks. By recognising the contextual nature of vulnerability, prioritising strong communication and forming 'super teams' of healthcare professionals, educators, families and community networks, it becomes possible to mitigate threats. A conscious and inclusive approach

to care, one that values lifelong learning, social support and respect for each person's unique journey, can transform the lived experience of Usher syndrome from one of isolated struggle to one of purposeful adaptation and enriched quality of life. In chapters 10–14, we examine strategies to improve communication, collaboration, health and social well-being as well as caring for the carers. Chapter 9 explores visual hallucinations in detail, as the general lack of awareness surrounding these means that people with low vision in combination with hearing loss suffer needlessly and can be misdiagnosed and mistreated. We want to raise the general awareness to reduce/eliminate these negative impacts.

9
Complexities of health and well-being: Seeing things that aren't real

Introduction

There is a funny juxtaposition that a person losing their sight can't always see the things that are there, while some of them sometimes can see things quite clearly that definitely aren't real. We, who live with Usher syndrome, may fall over toys on the floor, knock over garbage bins in the street, plunge down steps we didn't see or stumble off kerbs.

There is a widespread lack of awareness generally about visual hallucinations in people losing their sight, especially for those with combined hearing and vision loss, such as in Usher syndrome. There is no study of benign visual hallucinations, also known as Charles Bonnet syndrome, in Usher syndrome populations. There are case reports such as one where a patient who

had postoperative bleeding, a precipitating stressful event, who developed visual hallucinations. These were resolved following a transfusion. This case is not typical of the majority of experiences that people with low vision of any cause can have.

Visual hallucinations

Seeing things that aren't there – that no one else can see – is called having visual hallucinations. Visual hallucinations are very common for people:

- Losing their sight
- Who may have high fevers, illnesses, drug or medication side effects or other health stressors such as injuries and surgeries. These medically related visual hallucinations can happen to anyone, but are much more common in people with sensory loss/es and/or older age. Alcohol intoxication and alcohol withdrawal may also be responsible. These visual hallucinations occur as part of delirium, and you can read more on that in the previous chapter.

Visual hallucinations are often experienced by people with neurological or psychiatric disorders such as:

- Parkinson's disease
- Dementia/cognitive impairment
- Seizures
- Sleep disorders
- Psychosis and other mental disorders
- Brain tumours
- Migraines
- And many more conditions

This then means that the benign, frequent visual hallucinations experienced by some people losing their sight are easily and often misattributed, misdiagnosed and mistreated. In this chapter, we focusing on the visual hallucinations that occur in people who have intact brain function, but with conditions producing low vision of which Usher syndrome is one. This is known as Charles Bonnet syndrome. The data on the syndrome is poor, but it is known that having both hearing loss and low vision increases the incidence of visual hallucinations (Pang, 2016).

About Charles Bonnet

Charles Bonnet (1720–1793) was a Swiss scientist, renowned from a young age. He was a fellow of both the French Academy of Science and of the Royal College, London, by his 20s. His grandfather, Charles Lullin, had advanced cataracts and vision loss. Lullin kept a diary detailing his visual hallucinations as they came and went. Bonnet himself was partially deaf since childhood, suffered progressive vision loss from his 20s and had visual hallucinations, like his grandfather, in later middle age.

When Bonnet observed his grandfather's struggles with vision loss and the apparitions his grandfather had carefully documented, Bonnet also noted the absence of any impairment of Lullin's brain function. In 1760, Bonnet wrote *Analytical Essays on the Faculties of the Mind* (or 'the soul', depending on the translation of *l'Ame*). Here, he described the visual hallucinations as 'fictions of the brain' that his grandfather had diarised in detail, writing:

> …I should tell about a strange case that would be considered fabulous if not supported by testimonies of the highest credibility…I will simply say that I know a

> respectable man full of health, of ingenuousness, judgement, and memory, who, completely alert and independently from all outside influences, sees from time to time, in front of him, figures of men, of women, of birds, of carriages, of buildings etc…. All these visions appear to him in perfect clarity and affect him as strongly as if the objects themselves were present (Hedges, 2007, p. 112).

This establishes the triad of features in what Georges de Morsier, a French physician in 1936, will call Charles Bonnet syndrome: patients with low vision, no cognitive or mental illness, experiencing simple or complex recurring visual hallucinations.

While a variety of realistic illusions may appear, they may be simple (i.e flashes) or complex (people, landscapes, objects etc); they are not accompanied by sound effects. Bonnet ascribes the origin of these phantasms as 'in the part of the brain that commands the sense of sight' (Hedges, 2007, p 113). So, his reasoning is that the brain of a person losing their sight (and therefore deprived of visual stimuli), emits visual images where there are none to be seen. Bonnet's own phantasms began in his 40s. He had the comfort of knowing, from his observations of his grandfather's spectacular apparitions that no harm or cognitive decline would accompany them.

For the rest of us, such knowledge is sparse. There are many case examples and cautionary tales in the literature about the havoc that these hallucinations wreak: people diagnosed with mental illnesses they don't have because the visions are seen as evidence of psychosis or dementia. Those seeing the unseen can end up in psychiatric institutions and are believed to have diminished capacity for self-care and determination. Prior to 2010, the

research literature is thin, with a major increase in interest and articles appearing since then.

We still don't know *exactly* what causes the phenomenon, and we don't know *exactly* how common it really is, because a lack of awareness and the presence of stigma mean doctors under-diagnose and patients under-report these symptoms. We know however that visual hallucinations cause problems for many who live with hidden fears of psychiatric or neurodegenerative conditions, who struggle with quality of life and disruptions to sleep, education and work (Jones et al., 2021).

Risk factors

Basically, any condition that can cause vision loss, can precipitate visual hallucinations. We don't yet fully understand why some do and many don't. Known risk factors include:

- Increased age
- Social isolation
- Low cognitive function
- History of stroke
- Poor bilateral visual acuity

Stressful life events and natural disasters can precipitate or worsen hallucinations in at-risk individuals. There are a number of case reports from the COVID-19 pandemic where people with low vision reported first onset hallucinations, new hallucinations or increased frequency of existing hallucinations.

Hallucination incidence is higher in people with dual sensory impairment (such as Usher syndrome) than those with low vision only (Pang, 2016).

How common is Charles Bonnet syndrome?

Badcock et al. (2017, p. 3) who cited a figure of 37% in healthy older adults experiencing vision loss also report visual hallucinations. It is widely believed that this number is underestimated, since many who experience visual hallucinations do not report them for fear of being labelled mad or dangerous. We have been unable to find data of the syndrome's prevalence in patients with Usher syndrome, but we know the addition of hearing loss increases incidence. Thus, it is likely that at least one in every three people with Usher syndrome may see things that aren't there.

Not knowing what visual hallucinations mean is a major life stressor for most. The low levels of awareness in the community and among health and social care professionals and practitioners means that people keep quiet about the things they are seeing that aren't real. Even healthcare professionals can get it wrong when they experience hallucinations themselves.

Let us give you an idea of what visual hallucinations can be like by telling you about Annmaree's experiences. She has been seeing things that aren't there for a very long time, a few decades, and of more than one type.

Annmaree

When I was in my mid-20s, I was walking along a darkened street with my friends when a strange man jumped out at me.

'Oh, oh. Sorry', I said.
I thought I had nearly bumped into him.
He was tall,

He had a dark suit on,
He wore a white shirt,
He had a red wine colour tie,
His suit had shiny gold buttons.
'Who are you talking to? my friends asked.
'That man over there', I said but I looked and
there was no one there.

And then I realised:

He didn't have a face and he didn't have any shoes or feet even.
He simply wasn't real.

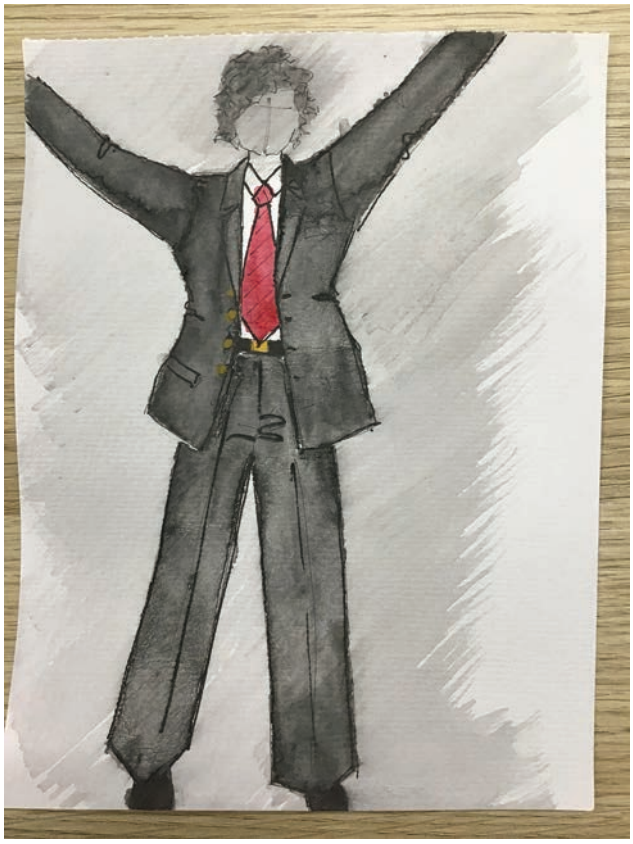


Image: Artist's drawing interpretation of 'the man in the dark suit'.

I was studying to be a doctor and I know that seeing things that aren't there is NOT good. So, I worried about this. And because I didn't want anyone to say I was mad, bad or dangerous, I TOLD NO ONE.

And no one has ever asked me if I see things that aren't there: no doctor, no nurse, no worker from any of the places that provide support.

I worried in secret and hoped this man would GO AWAY.

But the man in the dark suit kept on jumping out at me.

In restaurants, especially if there was a cute person taking me out and there were candles on the table.

In dark hallways at night where I worked at the hospital.

In streets.

In Sydney, Melbourne, Canberra.

In London, France, Hong Kong and New York.

After I had my first baby I kept on seeing the man in the dark suit. So I finally told a doctor I met who worked with the Deaf community. I said, 'I am really frightened that I might be psychiatrically unwell and hurt the baby'. He laughed and then said, 'It's perfectly okay, it's called Charles Bonnet syndrome, or visual hallucinations, or simply seeing things that aren't there, and it is very common in people losing their eyesight'.

Such relief. I have a medical degree. In classes and training, no one mentioned it. I had Usher syndrome, no ophthalmologist mentioned it. I attended two single-sense support organisations, no one mentioned it.

So, the man in the dark suit stopped stressing me so much when I saw him. I saw him a bit less when I was relaxed and happy, but I couldn't make him pop up or go away. He did what he liked, when he liked, and no one else could see him.

Then one day when I was very busy and had just gotten off an ultra-long-haul flight (23 hours travelling), I woke up in the morning. Looked at my watch which was analogue and had big numbers and hands that went around. ELEVEN O'CLOCK. This meant I was very, very late for an appointment. I checked my watch again. HUH, it now said seven o'clock. Then I checked again, the watch now said three o'clock. And none of these times were correct. It was actually one in the morning. Ok, I thought, I am having a stroke. This is not like the man in the dark suit who doesn't change. So, it must be something different. Luckily, I had an appointment with a new eye doctor the next day. I told him about the watch with weird times. He said, 'You are simply seeing things that aren't there'. So, no stroke. Charles Bonnet syndrome again.

Image: Three images of watch faces, showing 11 o'clock, 7 o'clock and 3 o'clock.

That's good. But it was difficult managing to be on time when I could never trust my eyes. I started checking the time on my mobile phone because that digital time was never weird.

Later, I bought a digital Apple watch with the loveliest big numbers and fall detection. I wear it now, and the funny times disappeared. I think it is because it is digital and not a round numbered clock face. I know instantly if I see the clock hands and numbers that it's not real, so don't worry.

More recently, I was in New York for a meeting and was having drinks. All of a sudden I saw little palm trees on the bar walls, in the air, on seats. My first thought: my drink's been drugged! But then, I saw palm trees in the lift and on the road. I realised THEY ARE NOT REAL. Let me tell you it can be tricky crossing a road when you are deafblind and there are palm trees everywhere, even if they are not real.

Image: Artist's interpretation of the palm trees in the bar, on the walls, in the air and on seats

But not everything is a hallucination. Earlier this year, my husband took me for a meal in a posh hotel. I looked around and saw … PALM TREES, all on one wall.

'Oh dear palm trees are here again', I told him. He laughed, then got his phone out and took a photo and enlarged it so I could see that there were REAL palms on REAL wallpaper. So, NOT Charles Bonnet syndrome.

Image: Palm trees printed on wallpaper

Over time, I see my suited man, watch faces and palm trees less and less. Knowing what is going on helps, and having health and social care professionals aware of my Charles Bonnet syndrome also helps. But most people experiencing these kinds of hallucinations alongside their vision loss don't have these comforts. This is why this book has a big section on Charles Bonnet syndrome.

Nature and forms of visual hallucinations

The hallucinations experienced in Charles Bonnet syndrome are many and varied; they can be formed or unformed, simple or complex, moving or stationary. Visions can be black, white or coloured. They usually last seconds to minutes. The hallucinations can be distressing and disruptive, sleep can be affected in some cases. Some hallucinations are frightening. Others may accept the hallucinations as part of a 'new normal' life with low vision. They can recur over days, weeks, months, years or even decades. They don't necessarily stay the same, they can change form, occur more or less frequently. New ones may come and old ones may go. Each Charles Bonnet journey is different.

Simple hallucinations can include:

- Flashes
- Lights (sometimes called photopsia)
- Dots
- Shapes
- Patterns like tiles, known as tessellopsia

Complex hallucinations are more formed and can include:

- Faces, known and unknown
- People, known and unknown
- Animals, trees, flowers and more
- Objects
- Houses and buildings
- Landscapes
- Letters and writing
- Roads and cars
- Gods and goddesses
- Devils and religious deities (these may terrify or calm, depending on what is seen)
- Anything and everything

The images may be stationary or moving. Moving hallucinations are called oscillopsia. One participant described seeing faceless men moving around their bedroom which was terrifying. Hallucinations can be realistic or they can be deformed. Another participant in research tells of seeing a child in a red coat with black buttons and a grotesque face deformed by thick black lines. This is known as prosopometamorphopsia, which is when facial features become grossly distorted (a note to readers: if you intend to use that lovely long word in a presentation, you will need to give interpreters time to manually fingerspell this long word and have it on a PowerPoint slide to assist with the spelling).

Interestingly, some types of visual hallucinations of Charles Bonnet syndrome have been shown to correlate to activity in specific related parts of the brain. Minakaran et al. (2019) reported

that functional MRI (magnetic resonance imaging) studies may demonstrate an association between the type of visual hallucination and activity in a specific part of the brain (2019). For example:

> Hallucinations of surfaces are associated with activity in the collateral sulcus, an area that responds normally to visual textures. Hallucinations of unfamiliar faces correlate with the left middle fusiform gyrus, and objects with the right middle fusiform gyrus, again as in normal visual processing (and) perceived motion of visual hallucinations is processed by cortical areas V5 and V6, as for normal vision. (Minakaran et al., 2019)

Adjustment

The little evidence we have suggests that most people over time, come to feel impartial about their hallucinations, with one study putting this figure at 70%. Adjustment is helped by knowing what is going on and what it all means. This requires knowledgeable health professionals to assess, diagnose and reassure.

However, approximately 25% felt their visual hallucinations were unpleasant or distressing. A low 5% found the hallucinations pleasant (Cox & Ffytche, 2014).

Mutability

Hallucinations are not always static, identical or singular for an individual. Some worsen as the day progresses. Hallucinations may change over time, from simple to complex; additional hallucinations may occur, as in Annmaree's description where the changing watch faces, the moving palm trees are in addition to her faceless suited man. Jones et al. (2021) note, 'The reports

suggest that more extensive visual loss is associated with more complex and enduring hallucinations' (Jones et al., 2021). However, regression and cessation of hallucinations as vision becomes severely or totally limited are also well noted. External events and life stressors may precipitate or amplify visual hallucinations, for example, being evacuated from a bushfire. One case report describes worsening hallucinations in a patient with Usher syndrome after surgery with heavy bleeding. Other stressful events include reports that COVID-19 infections worsened and increased the frequency of or precipitated visual hallucinations (Jones et al., 2021).

Causal mechanisms

There are many theories as to the causal mechanism of the syndrome. At present, the most accepted is, 'deafferentation, whereby increased activity in the visual cortex occurs following reduced sensory input from the eyes' (Jones et al., 2021). Some have likened the visual hallucinations to post-amputation limb pain: 'Severe vision impairment leads to the production of *de novo* images from the visual cortex causing visual hallucinations. This phantom vision theory could be similar to phantom limb syndrome' (Nair et al., 2014). Another putative mechanism is the neuromatrix theory, which posits that throughout the brain, a network of neurons, called the neuromatrix, can spontaneously produce visual phenomena experienced as hallucinations (Nair et al., 2014). The lack of a solid mechanism of causation has implications for treatments when needed for refractory and distressing hallucinations, as no single drug therapy has been found to consistently ameliorate the condition.

Challenges

Challenges for health and social care professionals in the diagnosis and support of those living with visual hallucinations include:

- Low levels of awareness among clinicians, especially general practitioners, patients and the community.
- People with low vision not systematically being told about visual hallucinations **before** they occur.
- Diagnostic challenges arise as the causes of visual hallucination are many and varied as outlined earlier. Misdiagnosis is not uncommon.
- The requirement for 'intact cognition' can be 'too hard' to disentangle diagnostically, as, for example, dementia and neurodegenerative conditions are associated with visual hallucinations without as well as with ocular pathologies. It makes no sense that only those who are able to recognise the hallucinations as not real after that fact are 'allowed' to have Charles Bonnet syndrome, when surely the same mechanism of reduced visual input to the brain applies to people with low vision and cognitive impairment?
- No evidence-based pathways or guidelines for managing those who need relief from the hallucinations and their impacts.
- Single-sense support and service organisations have low awareness and recognition of the impacts of dual sensory impairment, including visual hallucinations. This is the case in Australia, where deafblind-dual sensory impairment support (including Usher syndrome) is often fractured, with no dedicated government and policy recognition of the distinct disability status and complex needs of combined sensory losses.

Management

Management of visual hallucinations is multifaceted.

Educating Professionals:

- Educate the health and allied health students, professionals and practitioners about the syndrome.
- Ensure trainees in specialty and subspecialty training are Charles Bonnet syndrome aware.
- Include sessions on the syndrome in continuing professional development for medical, nursing, optometrists, orthoptists, audiologists and allied health professionals.
- Social care professionals who work with people and families with Usher syndrome also need education.
- Workers in residential aged care facilities, group homes and those providing in-home support to younger and older people with Usher syndrome.

Raising community awareness:

- Support disabled people's organisations, Usher syndrome and rare syndrome networks, older people's support organisations and sensory impairment support organisations to reach the wider community as well as known at-risk groups, including those with Usher syndrome.

Forewarning patients at risk (and their families):

- Professionals and support organisations should provide people with low vision, including those with Usher syndrome, with education about Charles Bonnet syndrome before the onset of hallucinations.

Funding support organisations to assist them to 'spread the word':

- Governments and social care funding bodies to assist in raising awareness and providing support and resources.

Reassurance and explanations.

Professionals to:

- Ask proactively, listen, investigate and diagnose. Doctors need to be more alert to the possibility that their Usher syndrome patients may be experiencing hallucinations, but fearful to disclose them.
- Pay close attention to differential diagnosis, take good histories and form trustworthy relationships.
- Provide reassurance and explain to the patient and family the benign nature of the condition.
- Emphasise that having Charles Bonnet syndrome does not herald incipient insanity, psychosis or cognitive decline.
- Advise patience, as the progression of vision loss often leads to reduction or elimination of the hallucinations.

Use diagnostic tools as an adjunct to clinical assessment (e.g. QR-SCB):

- French-language *Questionnaire de repérage du syndromede Charles Bonnet* Charles Bonnet Syndrome Screening Questionnaire (QR-SCB; in English).
- Aims to identify the syndrome and to locate those patients who need further intervention (see Cantin et al., 2019). Requires time to administer and covers eight dimensions: screening; characteristics of hallucinations; psychological impact; psychopathological origin; coping strategies; context and appearance of hallucinations; time-related matters; and psychological support.

Provide referrals to support services and organisations:

Referral to support groups and societies, for example:

- Esme's Umbrella – Charles Bonnet Syndrome (UK)
- Charles Bonnet Syndrome Foundation (Australia)
- Macular Society (UK)
- (See the resources appendix for more information on these organisations)

Behavioural strategies and therapies:

- Suggestions include frequent blinking or rapid eye movements or changing the light levels to increase visual input and alerting/distraction techniques (Jones et al., 2021). Exercise, reading, listening to music and changing a person's physical environment have also been found helpful to some.

Counselling:

- Should be offered, because living with vision loss generally is associated with higher rates of psycho-emotional difficulty, and living with visual hallucinations can be a source of marked anxiety and distress.
- Extra support may be needed in times of high stress, given that these may precipitate, increase or alter the nature of hallucinations.

Carer support:

- Families and carers need to know about Charles Bonnet syndrome as well. It can be complicated living with and supporting people with Usher syndrome and the degenerating sense/s.

Optimisation of vision:

- Keep the vision as good as it can be by scheduling regular eye reviews.
- Treating the underlying cause of low vision where possible, for example, remove cataracts when needed (rates of cataracts in

those with Usher syndrome are thought to be 55–65%, with higher rates in Usher syndrome type 1)
- Treating other eye conditions quickly, such as injuries, diabetic retinopathy and macular degeneration. Just because a patient has Usher syndrome doesn't mean they don't (and frequently do) get other conditions impacting their vision.

Support better living with low vision:
- Orientation and mobility training, aids and accommodations.
- Stimulate the senses with exercise, walks, reading, music, audiobooks, social engagement with others, puzzles and games.
- Living with Usher syndrome is at times complicated and good health and social care support is essential to living a good life (with fewer hallucinations, if possible).

Reducing social isolation:
- Good social support to keep networks and relationships.
- Good communication support to help those with Usher syndrome keep in touch with families and friends and to navigate new social relationships.
- Some researchers have suggested it's important to focus on the social interactions the person wants instead of creating myriad new clubs, classes and contacts.

Pharmacotherapies:
- Considered in refractory cases where hallucinations are causing disruption and distress.
- Possibilities include antipsychotics, anticonvulsants, antianxiety and SSRIs (selective serotonin reuptake inhibitors). Note the absence of large clinical trials, so case reports and small studies remain the mainstay of trial and error approaches.

Collaboration:

- Work together to keep people with Usher syndrome healthier and safer.
- Well-informed teams of professionals caring for patients with the syndrome make a difference in coping resilience and outcomes (Jones et al., 2021). We recommend creating a 'super team' of all the people involved in care/support of the person with Usher syndrome to learn from each other, provide ideas for support, write the reports to secure funding and provide co-ordinated approaches not only to visual hallucinations but all aspects of life with Usher syndrome.

Fund, foster and participate in research on Usher syndrome generally and visual hallucinations more specifically:

- Grow the body of knowledge about visual hallucinations in people with low vision, including Usher syndrome at all ages.

Conclusion

Seeing things that aren't there is reasonably common for people with low vision of any cause, including Usher syndrome. Charles Bonnet syndrome is the triad of low vision, visual hallucinations and intact cognition. Over one-third of people losing their sight may experience this syndrome. Most, with good information, support and reassurance, will adjust. A small number will find the hallucinations distressing, sleep may be interrupted and life impacted. We don't yet have expert guidelines and pathways for treatment; it is still very much trial and error and there is a need for more research. As mentioned, this book has a specific chapter on Charles Bonnet syndrome because of markedly low levels of

professional and practitioner awareness. Being aware will enable you to inform, investigate and reassure people experiencing visual hallucinations, preferably before their onset. No one should have to endure fear and anxiety over this largely benign condition. The next chapters are solutions-oriented: how to be good communicators, what kind of tactile messaging systems might be useful as well as how to live better with health and everyday solutions. We begin with Chapter 10.

10
Communication 1: Relational aspects of language and assistive devices

Introduction

Before we discuss relational, language and assistive aspects of communication with people with Usher syndrome, we would like to remind readers that Usher syndrome is a vast spectrum of diversity. Where one person with Usher sits on that spectrum changes over time: with degeneration of one or both senses that is lifelong; sometimes there can be movement towards better functioning, such as with Cochlear or brainstem implant or with the removal of cataracts, hearing and vision can improve.

But Usher syndrome is principally a degenerative condition and much adjustment is required in all life domains over the life course. Central to the Usher life is communication and access to information alongside safer mobilising. We live in an age of information overload, yet some people may struggle to access any of this because of the limited residuals of sight and hearing.

The digital divide is very real. A transition to tactile methods is common, especially if a bicultural approach hasn't been taken in childhood (learning oral and written language alongside signing, plus or minus Braille). Emma has learned sign language, unlike Annmaree, and both are learning Braille now as the loss of reading looms very near. Russ Palmer, who is a contributing author in the following chapter, swears by his cochlear implants that have vastly improved communication capacity. However, no matter how great and wonderful the technological aids and devices we have, don't mistake us; people with Usher syndrome live long and fulfilling lives, and work in a wide range of fields (although employment rates are lower, mainly due to prevailing discriminatory social attitudes and failure of support by employers).

There remain, however, situations of great vulnerability where our aids and devices may not work or be allowed. Healthcare and hospitalisations in particular are spaces where tactile messaging can be crucial to reducing anxiety and better patient experiences. We – Russ Palmer and Riitta Lahtinen (his partner and researcher) – have a lot more to say later.

For now, we examine the basics of communication. Before we do this, we offer some explanatory notes.

The following definitions are all related to deafblindness/Usher:

- Adapted equipment – equipment with adaptations to read or hear information.
- Audio device – equipment for listening.
- Auditory Brainstem Implant (ABI) – a surgically implanted device used to bring a sense of sound in someone with profound hearing loss by directly stimulating the brainstem.

Candidates may receive an ABI when a cochlear implant is unsuitable.

- Auslan – Australian sign language, a recognised language since 1987. This has its own cultural norms, grammar and is performative with both hands, facial expressions and gestures. Like other major sign languages such as American Sign Language (ASL) or British Sign Language (BSL), it is not signed English.
- Block (Block Alphabet) – Using a finger to write capital letters on the other person's hand to communicate. This can also be called 'print on palm'.
- Braille – Raised dots used to convey written information in a tactile format. The person can read the information with their fingertips, either on paper or using technology.
- British Sign Language (BSL) – A recognised language with its own grammar and structure that uses both hands to sign, body language and facial expression.
- Cochlear Implant (CI) – A surgically implanted device used to bring a sense of sound to someone with severe to profound hearing loss.
- Communicator guide – Someone who supports communication and guides a Deafblind person around their environment.
- Cued speech – using a set of handshapes near the mouth when speaking to represent the phonemes/sounds used to aid lipreading.
- Deafblind manual alphabet – Communicating using a modified version of the BSL or Auslan alphabet on to the deafblind person's hand. There are only a few differences in the manual alphabets of BSL and Auslan, but major differences in their visual and tactile sign languages.

- Hands-on sign – A deafblind person who uses BSL will rest their hands on another person's hands while being signed to, to receive information. This is called 'hand over hand' in Australia.
- Handwriting to text – Technology that converts handwritten text into typed format.
- Interpreter – A person who relays or interprets information from one language to another.
- Lip-reading – Receiving information by following somebody's lip pattern while they are talking.
- Lip speaker – A person qualified in relaying information through their lip pattern, without sound, so that someone can lip-read clearly.
- Live captioning – Text transcription of spoken audio onto a device, provided in real time.
- Moon – A system of raised shapes that is a form of reading information through touch.
- Notetaker – A professional notetaker takes notes for the person with Usher from things such as meetings and training sessions.
- Objects of reference – Receiving/feeling information from an object as a form of communication, that is, feeling the keys means going out.
- Own family signs/home signs – Families create their own signs to use at home, which are not part of a recognised sign language.
- Palantypist – A person qualified in typing spoken information live into written text.
- Sign supported English – Sign language following the grammar and structure of spoken English, to support a person with Usher to follow spoken English.

- Social-Haptic Communication (SHC) – Information is 'drawn' or signed on the person's body, such as their back or arm. Often used to convey additional information such as about the environment, mood or to convey information quickly in an emergency or shorthand. This must be agreed beforehand with the individual. See Chapter 11 for a fuller explanation and examples.
- Speech to text – A device used to pick up speech and convert to written text.
- Subtitles – Text on TV, video and other devices to relay information in text that is spoken or signed.
- Tactile sign language: As a person with Usher syndrome loses vision, even restricted frame and reading lips becomes impossible. Tactile Auslan is a communication method where Auslan signs are interpreted through touch rather than sight. It's used by Deafblind individuals to understand Auslan, and involves the Deafblind person placing their hands over the signer's hands to feel the shape, movement and location of the signs.
- Types of pen and paper – Either thin or thick black pens on white paper (or other preferred colour) can be preferential for people with Usher when writing information down.
- Visual frame – For a person with Usher who uses sign language but has loss of their peripheral vision, signing is giving in a smaller space, as if in a 'frame'. This can sometimes be called restricted frame as the person with Usher's visual field reduces.

Communication is a broad topic; however, for people with Usher/Deafblindness, there is a lifelong need to adapt to different communication methods. The method/s used depend on whichever

way suits them best in terms of their ability, skill and available resources.

Every person with Usher has their own unique communication skills and abilities in terms of their level of vision, the effects of retinitis pigmentosa, fatigue and changes to environment and settings such as ambient light. As a person with Usher's vision deteriorates, their communication skills may change, depending on where their vision and/or hearing is. For example, if they have narrow vision, they will look straight at the person, as if looking through a telescope.

Annmaree

One thing I noticed from my 20s was that the blinder I got, the deafer I became. I was very dependent on visual cues to support my hearing residuals to make sense of what people were saying and meaning. Conversely, too, as I lost decibels of hearing later in life, I was unable to use my lifelong lip-reading to support my social encounters because I had, by this stage, lost the ability to see faces and read lips. These combined to make a transition to tactile forms of communicating an imperative.

Receptive/lipreading/facial expression/handshapes

Deaf and Deafblind people rely on lip-reading, facial expressions and handshapes when communicating.

When a person with Usher receives information from an interpreter, they may be able to process this language in much the same way as their Deaf peers; however, they may have blurred

vision due to their retinitis pigmentosa, which means that they are no longer able to see the person's lip pattern when speaking, the facial expression of the person (conveying much emotion and intent) and the handshapes used. This is because the effects of retinitis pigmentosa are constantly changing. They also may have 'good' and 'bad' eye days. In some cases, a person with Usher may be able to see the whole picture of the interpreter's face, chest and arms. However, at times, their vision may reduce to just the face or mouth movement due to many factors, including lighting, fatigue, the effects of retinitis pigmentosa and peripheral vision changes.

Emma

I remember being able to see a friend well enough to communicate early in the day, but as the room got darker in the afternoon, I would only be able to see my friend's mouth, not their expression or handshapes being used.

Annmaree

I have a friend with Usher syndrome, who has an acronym BDBD that they use to describe a 'bad deafblind day'. This phenomenon is common to people I know with Usher. Some days you wake up and can't see and/or hear enough to make it to work or to the social gathering or even out of bed. Sometimes, I would struggle to find my glasses or my hearing aids, so if no one else was around to be my 'eyes and ears', I would get back into bed and snooze. In my experience, BDBDs are related to co-occurring fatigue. The effort required, as Usher progresses to navigate the day-to-day activities, is enormous. We have to be vigilant, always, to avoid

falls, social catastrophes, domestic disasters – I am the person who almost took a hearing aid battery instead of a Panadol tablet. This intense alertness and close focus are fatiguing. This makes bed, sleep and understanding people our good friends.

Modifications to communication

Due to changes in our vision, which can even happen daily, a person with Usher and the person supporting them will need to constantly adapt and modify their communication to fit the current situation. This pivot can be because of changes in vision and hearing, the presence of fatigue or the failure of technology being used. One situation that poses increased risk and need for pivoting is the healthcare encounter.

Within healthcare settings, there are many different environments to navigate throughout a hospital or GP surgery. These might involve different lighting conditions and different kinds of tests and examinations that may be carried out, so that a variety of modifications to communication will occur.

Some examples of pivots needed include adjusting the layout of people and professionals in the room, visual frame signing, hands-on signing and use of social haptic communication (SHC) where information is 'drawn' or signed on the person's body, such as their back or arm. This is often used to convey additional information, such as the environment, mood or to convey information quickly in an emergency, in shorthand. This must be agreed upon beforehand with the individual.

There may be many changes throughout an appointment, encompassing an array of things, such as changing from signing

to Braille and to social haptics and back to visual frame signing and so on. For example, when entering a consultation room, the lighting may change drastically from the waiting area. There is a time of adjustment for someone with retinitis pigmentosa, as it takes the eye much longer to cope with such changes. This could also mean a change in communication from visual frame to hands-on signing.

The layout of interpreter, patient and doctor may need to be adjusted so everyone can see and hear who they need to. If lights need to be dimmed or turned off, such as in an eye appointment, the interpreter must inform the clinician that the communication is going to be more difficult, slower or impossible. When the lighting is changed, and modifications must be made by the interpreter, the clinician, and the person with Usher, they must decide how they want to receive information at this time – it could be delivered in one go before lights are dimmed and assessment is carried out. Social haptics could be agreed as to when the person is to turn their head or eye gaze, such as 'when I tap your left shoulder, look left', so that there is no need for visual communication.

Even during an appointment where the lighting is good and consistent and the environment is well suited for communication to take place, a person with Usher may need to make their own adaptations due to fatigue, having a 'bad day' in terms of their vision or struggling with the complexity of the information. Interpreter/support worker/family and patient must work collaboratively together to ensure they can still communicate comfortably and to make decisions on whether information needs to be limited, written down or mode adjusted.

Emma

When I attended a hospital appointment in the winter, it was very dark, difficult to see and my interpreter and I used hands-on BSL before entering the consultation room. This room was brightly lit, and I was then able to see more and managed to receive information from my interpreter with visual frame signing.

Annmaree

I want people to know that there is no universal set of adaptations for all people with Usher syndrome. When I was in hospital having my children, I was at a stage where I needed lights on and blinds up to maximise the light in the room to improve my ability to understand what was being said and what was going on. But now, I need blinds down, lights dimmed, as my eyes hurt in bright light and communication definitely doesn't happen. And if the doctor sits in front of a window, forget it. I can't cope with that level of direct sunlight. So over time, what I needed to navigate was not just healthcare encounters, but social situations, educational settings (whether I am learning or the one giving the lecture) and any shopping/banking/appointments had to change. More is done by proxy now (partner, support worker, accessibility assistant) and we use more tactile methods (deafblind manual alphabet, social-haptic communication, tactile sign shortcuts and basic tactile sign language as we are still learning this). We use more speech to text and dark mode is our closest companion.

Not all interpreters are deafblind communication skilled

There is a shortage of uniquely skilled interpreters who work with Deafblind people and those who communicate using hands-on

BSL and other specific modes of communication. There are plenty of BSL interpreters available, but sadly they are without the specific skillset to meet Deafblind people's needs, which is challenging and frustrating.

We would like to remind health and social care professionals and practitioners to ASK each person with Usher (or any deafblindness) what kind of interpreter they need and who would be best. Tactile communication methods require a high level of trust, and every effort should be made to book the person's preferred interpreter.

Interestingly, in Australia, there are moves to formally credential interpreters for the deafblind with the intention that these interpreters would be paid a little more for the extra qualifications. It is expected over time that deafblind interpreting credentials will be needed for official interpreting. Individuals can still at this time use their NDIS plans to choose their preferred interpreters. This is an evolving policy in practice situation. Monash University is running the first course for deafblind interpreting in July 2025 (see the Australian resources appendix for more information).

Reduced vision, reduced signing frame

When a person with Usher has remaining useful peripheral vision, they will be able to see an interpreter more wholly, as if on a large screen, when they are talking and signing. When a person with Usher's vision becomes narrow, they may use visual frame sign language, meaning that the interpreter will be signing in a much smaller space or frame, like seen through a small television or 'frame'. Having a loss of peripheral vision means that if the

interpreter were to sign in their natural signing space, the person with Usher would not be able to see both the interpreter's lip pattern and signing/handshapes, which is why an adaption to visual frame signing helps, as the interpreter brings their signing space inwards. This means they are signing in a smaller, higher space, nearer the mouth, so that more information can be taken in and communication is more comfortable to process without 'gaps'. Sometimes, sitting further away from the interpreter helps, as tunnel vision means that they can then see more of the interpreter in their narrow field of vision.

If a person with Usher has a very small amount of vision remaining, or no remaining vision, they use hands-on BSL. This means that a person with Usher using this method of communication will need more time to receive and process information.

For people with Usher using an interpreter, communication will be modified throughout the conversation in order to meet the person's understanding. The interpreter will have to change their style and register of communication throughout the appointment, focusing not only on the person with Usher, but also listening to the conversation/topic that is being discussed and relaying this to the person appropriately, along with any important environmental information. The person with Usher will respond in sign, which can then be relayed through the interpreter back to the clinicians.

Communication methods and equipment

There are different methods of communication used depending on the individual, what suits them best and what resources and

services are available to them. A person with Usher may wish to use lip-reading, or cued speech, changing to the deafblind manual alphabet as well as using equipment and technology as their vision deteriorates. For a person with Usher who uses sign language, they may use visual frame sign language and later may use hands-on sign language as well as social-haptic communication.

Individuals may choose to use more than one type of communication at any one time, depending on the environment, situation and other factors.

Examples of equipment that can be used to aid communication include:

- Braille
- Audio device
- Palantypist
- Speech to text
- Subtitles
- Types of pen and paper

Good practice when working with Deafblind people in health and social care setting:

- ASK (acquire specific knowledge) a person with Usher what their communication preferences are and if there is a preferred interpreter they would like to work with.
- Acquire skills and knowledge of your own: Learn the deafblind manual alphabet (doesn't take long), discuss some social-haptics touch messages with the person and where on the body it is ok to touch. Be ready to pivot to another method if the one being used isn't working. This means if

you have an accent, own it – don't make deafblind people feel frustrated – and move to another method.

- Ensure the interpreter booked has the right qualifications and experience to work with deafblind people. Depending on the appointment, it could be vital to have specific skills –eye surgery, for example – someone who is appropriately trained and can adapt to the changing communication needs.
- Remember that no two people with Usher have the same communication requirements. This is also true of siblings with Usher.
- Allow extra time to communicate with a deafblind person. Double appointments are always a good idea.
- Clarify and make sure that the person has understood the appointment and/or treatment.
- When having an interpreted appointment, remember to look at the person with Usher, instead of looking at the interpreter.
- Communicate with the patient who has Usher; do not share information or ask questions to family members, interpreters or support workers.
- Respect that the person with Usher has the same rights as anyone else when accessing medical appointments or social services.
- Don't sit/stand with a window behind you due to glare; it is better for the person with Usher to be in this position, or to change the lighting to brighten the whole room. If this is not possible, a more accessible room might be necessary.
- Ask them where they would like you to sit/stand, whether it is between four to six feet distance or closer/further back.
- If you are not sure, it is ok to ask questions.

- If the person you are treating is using hands-on sign language, deafblind manual or social haptic communication, it is vital to get enough chairs and allow them to be moved around to ensure smooth communication.
- Wear a plain top (without pattern) in a contrasting colour to your skin.
- Allow extra time for the appointment.
- Be patient.
- Do not make assumptions.
- Book an interpreter in advance.
- Do not interrupt the person with Usher when they are speaking/signing.
- Treat the patient with Usher with the same dignity and respect as you would any other patient.

Communication failures rife in health and social care

Many people with Usher, who use a variety of communication methods, have had negative experiences accessing health and social services due to a lack of communication provision. Some people with Usher will also have difficulty accessing information, such as reading letters or accessing online booking systems due to their level of English (if their first language is BSL) or due to little or no reading vision.

Examples in the health sector

Example 1

For an upcoming cataract operation, a lady requested a BSL interpreter qualified and experienced in using visual frame, with the

ability to use hands-on BSL and social haptic communication, in order for the surgery to go smoothly.

A specific interpreter was requested directly by the patient to the hospital, in order that these communication needs would be met. The hospital refused to book the requested interpreter due to a factor of cost and cancelled the upcoming surgery the night before it was due to take place, despite the interpreter's availability for the proposed date.

Example 2

A person with Usher requested a hands-on interpreter for his upcoming hospital appointment. The hospital provided a communicator with level 2 BSL, who was not a qualified interpreter and could not use hands-on BSL. This had a negative impact on the individual and compromised the effectiveness and clarity of understanding at his appointment.

The importance of redressing communication failures

Addressing communication failures goes beyond meeting disability legislation requirements. It represents a fundamental commitment to human dignity and equitable health and social care. The diversity of communication needs within the Usher community requires health and social systems to move beyond one-size-fits-all approaches. The consequences of failing to provide good communication cost everyone: individuals with disability in social isolation and poor health and well-being, families in increased care provision and distress and health and social care systems in increased costs.

Redressing communication and care failures means systematic change: adequate funding for qualified deafblind interpreters, recognition that communication access is a clinical and social necessity and policies prioritising the needs of people with Usher syndrome (and all other communication disability) over cost concerns. Health and social care staff, families and individuals need education about deafblind communication principles and creating accessible environments. Individuals and families need support, training, devices and human supports. Society is responsible for the provision of these:

> Deafblindness limits activities and restricts full participation in society. According to the Convention on the Rights of Persons with Disabilities (UNCRPD) participation is a given right for all human beings. Thus, in order to enable the individual to use their potential capacity and resources, society is required to facilitate specialised services.
>
> The individual and their environment should be equally involved, but the responsibility for granting access to activities lies with society. An accessible society should at least include:

- Available competent communication partners.
- Available specialised deafblind interpreting, including interpreting of speech, environmental description and guiding.
- Available information for everyone.
- Human support to ease everyday life.
- An adapted physical environment.
- Accessible technology and technological aids.

> A person with deafblindness may be more disabled in one activity and less disabled in another. Variation in

> functioning might be the consequence of both environmental and personal factors.
>
> Specialised competence related to deafblindness, including an interdisciplinary approach, is vital for proper service provision. (Nordic Welfare Centre, 2024)

Conclusion

Successful communication is possible when providers are willing to learn, adapt and collaborate. We must also learn to pivot: when one communication method doesn't work, swap to another. As Usher syndrome is a degenerative condition, communication needs and capacities change over time, so there is no one time solution for everyone, or indeed, anyone. When we get communication right, we reduce individual distress and isolation, we improve compliance with health and social instructions and we save money, as better health and social care is associated with better health and well-being. In short, we create more inclusive health and social care systems that benefit everyone. As one participant told us, 'Communication is the first act of care.' Chapter 11 takes communication methods into tactile messaging, in particular social-haptic communication.

11
Communication 2: Healthcare and the role of social-haptic communication

Introduction

Healthcare encounters are fraught with communication failures with the potential (and all too real) consequences of harm. The 2024 Consumer, Carer, Professional and Practitioner Survey of the Needs of Adults with Dual Sensory Impairment-Deafblindness, which Annmaree led, had a number one top theme for those with lived experience of deafblindness or dual sensory impairment, including the Usher syndrome subgroup, of 'communication failures are rife', especially in healthcare.

> "It's more important that the professionals change the way that they communicate and work, not the deaf-blind people."
> "Talk to **me**, not my support worker."
> "I didn't understand anything the doctor said" was common, or the specialist, audiologist, optometrist, etc.

Other research (Watharow, 2024) has shown a poor report card of negative healthcare experiences, not knowing what is going on, uninformed consent, abuse and neglect. Not knowing what is going on extended to the post-discharge space as well, and this has implications for ongoing health, well-being and increased costs. Huddle et al. (2016) concluded that better detection, management and support of sensory losses generally would save substantial sums: 'Addressing sensory impairments could potentially carry substantial implications for public health given that these impairments are highly prevalent, undertreated, and amenable to treatment with established, low- to no-risk rehabilitative interventions. If individuals with DSI {dual sensory impairment} are at greatest risk of adverse health outcomes, then strategies targeting this at-risk cohort may be particularly impactful' (pp. 2–3).

Good communication confers health and well-being benefits as well as cost savings to healthcare institutions.

Research shows that effective healthcare communication results in:

- Shorter hospital stays
- Fewer hospital readmissions
- Reduced emergency room visits
- Closer treatment adherence

- More effective medical follow-up
- Less unnecessary diagnostic testing
- Fewer medication misadventures
- Improved healthcare outcomes
- Reduced healthcare expenditure
- Good communication is strongly associated with better patient experiences, and better patient experiences are linked to better health and well-being outcomes

Chapter 13 has more on this.

In the interest of avoiding the dire consequences of poor communication for patients and families living with Usher syndrome, we showcase some strategies and solutions. In this chapter, we examine social-haptic communication and its particular role in improving healthcare communication. We have asked two experts with lived experience, expertise, knowledge and research (Lahtinen, 2008) to share their insights.

Russ Palmer has living experience with Usher syndrome, practicing, receiving and promoting social-haptic communication, and Riitta Lahtinen has lived knowledge as a partner and expertise as a researcher, professional and practitioner. They have been involved in documenting, systematising and promoting social-haptic communication for over three decades.

Communication, healthcare and Usher syndrome

By Dr Russ Palmer and Dr Riitta Lahtinen

In this chapter, we, Russ and Riitta, focus on some of the challenges in learning to deal with a deafblindness condition, such

as Usher syndrome. This includes how one's communication may change from spoken language to using touch-based methods such as social-haptic communication to enhance or augment spoken language communication (e.g. when one is visiting hospitals or having specific treatment).

We shall use the word deafblind, and also dual sensory impairment interchangeably, to represent people with combined hearing and sight loss. Deciding on an identity or a name each individual uses for the combination is a very important process to go through when experiencing changes over their lifetime due to progressive hearing and sight loss. Furthermore, using the word deafblind (or dual sensory impairment) may allow individuals with hearing and sight loss to obtain more services in their own geographic regions from their local service providers.

For me, Russ, as a deafblind person due to Usher syndrome, I've had to progress over time to implement and develop new communication methods. These include deafblind alphabet, print on palm using block letters and visual frame to hands-on signing (sometimes called hand over hand). In my case, when I met my Finnish wife Riitta Lahtinen in 1991, we began developing social-haptic communication. This has since become the subject of Riitta's PhD thesis and her life career.

Russ

One of the challenges in dealing with a condition like Usher syndrome over the years is learning to deal with the constant changing of one's vision and hearing. Since my late teenage years, at the age of 21 and after my diagnosis, I realised I had a problem with my sight. Initially, this had a profound effect on the

way I felt, how my career and personal lifestyle would need to be addressed. The doctors at Moorfields Hospital in the UK basically said: by the age of 50, you will become blind and completely deaf. This had a deep impact on how I felt about my situation at that time.

I was born severely deaf and was wearing two high-powered hearing aids, used spoken language and lip-reading and starting to realise how difficult it was to see in bright light and dazzling conditions. I also had night blindness. One had the attitude of every time the sun goes down one had to make sure you were in a safe and well-lit place to avoid hitting obstacles. Later in life, at 66 years of age, I am totally blind, have been since 2015 and have two cochlear implants, which has made a big difference to my life and the way I communicate and do things. I use a white cane and have to use a guide at all times outside my home environment as I do not feel confident or safe enough to move around independently. When I was first diagnosed all those years ago, I was driving a car and was totally independent and did not have to rely on others guiding me.

Looking back, I was also motivated to push on with my computing career to become qualified as a computer analyst programmer. Before taking early retirement at the age of 30, I achieved the post of project team leader where I had four people under me designing a property database. Also, I was going through my personal identity crisis, going from a hearing-impaired person to a person with a visual impairment, later registered blind and finally deaf and blind. All these changes were difficult to accept in the small time period. Learning to deal with these issues is another thing. In my case, I had to deal with them myself with

only a little professional help. Following taking the early retirement, I decided to change my career in trying to become a music therapist, which for someone with my challenges, with my hearing and sight disability, was a little bit unusual to say the least! But I had decided I wanted to work with people. Since I was an active musician playing guitar, piano and writing songs, this seemed a good idea. So, I ended up for 2 years at Dartington College of Arts in Devon, UK, in preparation to do a music degree. This was followed by a 1-year visit to Norway, where I did a music programme with special teachers who were working with deafblind children who had profound learning disabilities. This started my journey with music therapy. In 1999, I qualified in Sibelius Academy in Helsinki, Finland, as an International Music Therapist. If one considers that all the course work was in Finnish with only some lectures in English, then this was a good opportunity to be introduced to the interpreting services for the deafblind people in Finland. This did not stop any of my independence; it actually increased my possibilities to do a wide range of activities. What I am trying to say here is that one has to try to accept one's condition and take on different services to be able to function independently. Defining oneself as deafblind will allow one to receive more services in their own geographic regions from their local service providers.

This chapter explores various communication techniques that may be useful for both families and different healthcare professionals. We discuss different solutions that the deafblind person may find useful when identifying their personal communication elements and needs. When consulting a doctor or having an operation, there are certain key elements that will improve

communication between the professionals and the deafblind patient.

We advise all deafblind people to have a support person with them if possible. All too often, the hospital protocols are very strict and do not allow 'unqualified' persons to be present in the operating theatre or treatment room. In that situation, it is critical that everyone is able to use other communication methods to alert, soothe and give information to the patient directly.

For deafblind patients, touch-based methods are easy to understand. Social-haptic communication has a group of touch messages that have been developed for this purpose in healthcare encounters. These are known as hospital haptices. They include a set of touch messages which allows the patient to feel safe and more relaxed, even if they are not able to use any of their hearing aid devices and other assistive technologies. All people with deafblindness have the right to information about their healthcare and treatments and to be involved in shared decision-making.

Hospital haptices

The need for hospital haptices arose when I, Russ, had a cataract operation. On this occasion, I had to take off one of my hearing aids. This resulted in my receptive communication capacity diminishing. I was not able to see lip-reading and was only able to listen through one hearing aid device. Before the operation took place, Riitta and I agreed on a certain set of touch-based messages called haptices. This would allow me to identify key stages within the operation. On this occasion, Riitta was allowed to be at my side in the operating theatre giving me information as and when these key points of the operation occurred. As a

result, I was very surprised by how much lower my blood pressure was and how relaxed I felt during the whole process. Usually, one never knows what will happen next, especially if you cannot hear or see so well.

Cochlear implant (CI) operation

During the time I had my first CI operation, I used two high powered hearing aids, and it took more time to understand and hear what was going on. I needed a lot of time to make sure I had clarified all the stages that will be happening and I was on top of understanding risks and benefits. I was very fortunate to have a good anaesthetist who was very patient and explained everything very precisely.

For this operation it was extremely important to have a support person as I was so nervous they would operate on the wrong ear, and even before the operation I could not see or hear anything. I was also lucky to have Riitta there to repeat things and complement what was said with touch-based messages, haptices if needed.

There was also an opportunity to explain to the post-op nurse how to give me my one hearing aid when I started to wake up in the recovery room to communicate with the nurses since Riitta was not able to be there.

We taught her some basic haptices, such as yes and no. (Riitta says that to communicate, the patient asks questions with a yes/no answer, e.g. did everything go well? Can I have something for the pain? The professional then can answer with haptices for yes or no.) One important point here is to remember to recharge

your hearing device batteries in advance or put fresh ones in. It would also be good to put the noise cancellation programme on if you have one as the recovery room is likely to be noisy.

In my case, I had a private room in the hospital where Riitta could stay. This made things much easier; I was able to communicate in a quiet environment and also Riitta made me aware of the head bandage I had on – I looked like a mummy. This allowed me to be much more relaxed and to recover without feeling anxious. In the UK, my NHS (National Health Service) hospital had a unique privileged arrangement of people with dual sensory impairment having their operation in a private hospital and having themselves a private room to recover in.

Sao Paulo, Brazil, 2015

If we visit abroad, we need to be prepared for any unknown eventualities, such as when I visited Sao Paulo in Brazil. On the day before travelling back, I had a fever and started to develop black blisters on one of my legs. It became obvious that I had caught an infection. This turned out to be septicaemia. Luckily, I had good insurance and I was rushed into a private hospital in Sao Paulo where I needed to have an immediate operation to remove the bacteria from my leg. I was informed that had I travelled, I could have either lost my leg or died on the way back to the UK; so, I had a lucky escape. Since most of the people spoke Portuguese and only a few doctors spoke limited English, the full-fledged hospital haptices system started to evolve on the go. These included injection, intravenous drip, blood pressure, medication, time, and others were introduced. These turned out to be extremely helpful in the situation where I was starting to

recover slowly, but was still too tired to use my cochlear implants to listen to spoken language or stay awake for too long to comprehend what was going on. Hospital haptices were also taken on board by the hospital staff who adapted them in their communication with me quite easily. That also made it easier for me to understand the staff during different shifts, so that everyone who came to me used the same system. Riitta made sure everyone was using the haptices in the same way. Luckily, I had my iPad to listen to my music to relax and distract from my pain, and this felt very healing and resulted in me feeling less anxious.

Using hospital haptices in healthcare settings

As a result of the Brazilian experience, Riitta adopted the idea of introducing a hospital bag, which included some basic but important items relating to deafblind communication and equipment. Information such as how to change batteries, how to charge the cochlear implant device, for example, the bag also included a deafblind wristband (see Figure 1), and an illustrated word list with basic hospital haptices.

This hospital bag is always located in a certain place in one's home, so that it is easy to find and also for the paramedics or family/carers if needed. On one occasion, it was necessary for me to have a treatment that involved me lying in a specific position with my cochlear implants off, and Riitta was not allowed in the operating theatre in that particular hospital. Thus, before the treatment started, both Riitta and I explained to the nurses and doctors that I would benefit from them using hospital haptices with me. On this occasion, it proved to be very successful as both

Figure 1: Three bright orange wristbands with DEAFBLIND, VISION IMPAIRED and HEARING LOSS printed on them in navy text.

nurses and doctors thought it was a good idea. I felt relaxed and safe in good hands and not anxious. This made everything go smoothly. Even though the subgroup is called hospital haptices, they can be beneficial in wider settings, such as personal care, family-related matters and social care.

Hospital haptices are part of nursing practice as their role is to provide information to patients as to what is happening or how

to respond by their own movements. Those who use hospital haptices need to understand how these methods are being applied. The use of haptices can be agreed upon by the patient and nurses in advance. Haptices are chosen according to individual needs. For the dual sensory impaired – deafblind people – social-haptic communication (SHC) is one natural method of receiving information when visual and auditive information is not possible. Hospital haptices is a subgroup of the social-haptic communication research which originated from the 1990s by Lahtinen (2008).

Social-haptic communication uses touch as an agreed way to give the patient information supporting communication because the patient's physical condition after the operations or during treatment can be unstable, or they cannot hear instructions or see their surroundings. Often, deafblind people are not able to use any devices and/or interpreters in these situations. While in the hospital, many different people can use haptices such as nurses, caregivers, interpreters, assistants and family members. Usually, these short, practical and describing messages are haptiering not only onto the hand or arm but also other areas are recorded such as leg, shoulder and head, to indicate one is not allowed to move during treatment.

Hospital haptices, meaning agreed touch messages onto the body, have been studied with deafblind people and hospital staff during treatment procedures such as cataract operations, various medical examinations, recovery and isolation rooms. Hospital haptices can help communication during various nursing functions, because the meaning of haptices gives information about the upcoming procedure or how the patient should

act during the treatment. Those who use hospital haptices need to know which one and how to adapt to them as communication in different circumstances (Lahtinen et al., 2016).

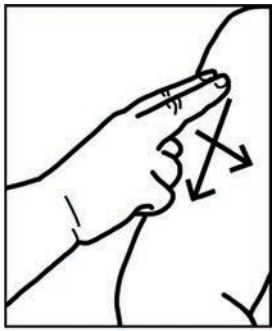
Effective communication is one important aspect of interaction in terms of good patient care. Interaction between the nurse and the patient can be hindered for many reasons when the patient cannot hear or see well. The patient's health may not be good, and the hospital environment is unfamiliar, it can be noisy and dazzling due to bright lighting conditions. These special hospital haptices used in the care or hospital environment were born from the need to give information in advance to the patient about upcoming procedures (Palmer, 2015).

Hospital haptices consists of 12 different haptices which were used by some dual sensory impaired people. They form six different functional categories. They are identified by professional role, confirmations (feedback), emotions, measurements, length of treatment and how to show movements (Lahtinen et al., 2016).

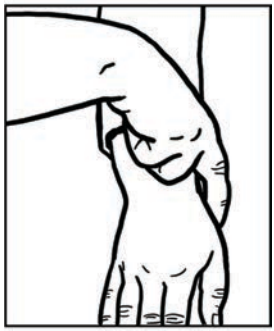
Identification of professional role

The first category is identification of professional role. When a person enters the personal area, they must state who they are. Usually, the person tells their name and profession. It is important for a deafblind patient to know who has entered next to them into their personal space. This can be identified either by a NURSE or DOCTOR – haptices (Figure 2).

NURSE communicated/haptiered with two fingers on the upper arm in the shape of a medical cross: a downward vertical line and then a left-to-right horizontal line.



nurse



doctor

Figure 2: NURSE and DOCTOR haptices.

DOCTOR communicated/haptiered with two fingers on the inside of the thumb side of the wrist, where the pulse may be felt.

Confirmation

The second category is confirmation. When talking to a member of staff, a deafblind person is speaking using spoken language, the nurse or doctor can answer by giving YES or NO haptices (Figure 3) onto the hand or arm. These are usually basic confirmations that require a question to be answered yes or no.

YES communicated/haptiered by tapping the hand up and down on the person's hand/arm.

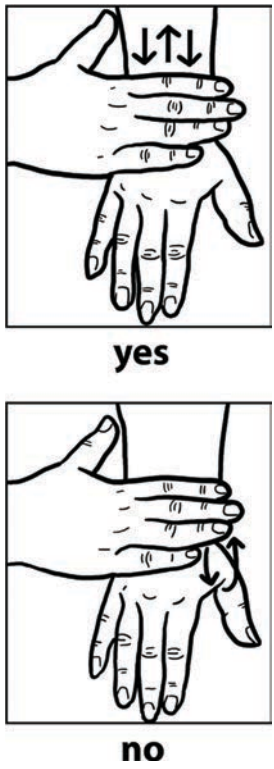


Figure 3: YES and NO haptices.

NO communicated/haptiered by rubbing the hand left to right on the person's hand/arm.

Emotions

The third category is emotions. The patient's condition is observed in many different ways, visual observation or by asking. Figure 4 shows how we can share 'HURTING' information if the procedure is painful. This way, the patient does not panic and knows to prepare in advance.

'HURTING' communicated/haptiered by touching all five fingers in a claw shape onto the top of the person's hand with intensity.

Figure 4: 'HURTING' and BLOOD PRESSURE haptices.

BLOOD PRESSURE communicated/haptiered by using the hand to form a 'cuff' around the person's upper arm.

Different measurements

In the fourth category are different measurements. The nurse can measure the patient's blood pressure or perform other health-related measurements or tests. Usually several times a day, including in the middle of the night, a nurse may perform a set of observations that include blood pressure, temperature, respiratory rate, heart rate, oxygen saturation and check wounds, intravenous drips and drains if they are present. The blood

pressure haptice shown in Figure 4 can be used to signify this set of observations.

Duration

The fifth category is length of treatment. This can be, for example, to tell now it is 'TIME' to do something which has been agreed earlier. In Figure 5, this is shown by tapping a couple of times the place where a wristwatch would be.

The sixth category is showing how and which direction the deafblind person needs to move or not to move. Figure 5 shows information 'WAIT, DON'T MOVE.' When showing this haptice, the deafblind person is in one place as long they feel the hand is showing the information by pressure, intensity.

TIME communicated/haptiered by tapping the tip of the finger onto the middle of the wrist.

WAIT, NO MOVEMENT communicated/haptiered by placing a flattened hand onto the wrist/arm.

Haptemes

'A hapteme is a grammatical variable related to touch, an element for building and identifying haptices and of separating individual haptices from each other. Haptemes of movements are recognised as the direction of movements, change of directions on the body, directions between people, pressure, speed, frequency, size, length, duration, pause, change of rhythm and shape.' (Lahtinen, Palmer & Ojana, 2012, p. 270)

Research and lived experience of social-haptic communication have shown that when using haptices, the most common

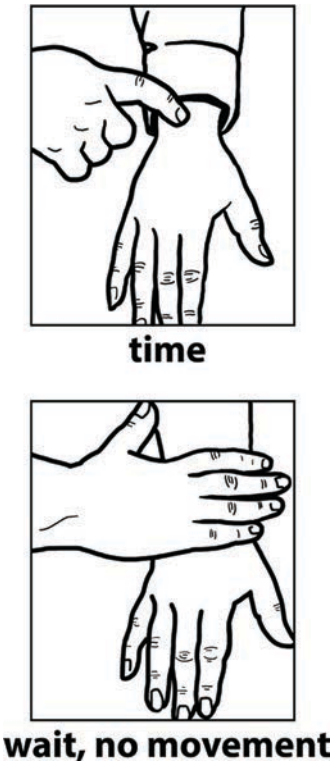


Figure 5: TIME and WAIT, NO MOVEMENT haptices.

haptemes were location, pressure, movements, speed, duration and handshape. Location was arm and hand. Pressure – intensity of touch – was normal or heavy. Movements were towards or surface of the skin. Speed was mostly calm. Duration was longest when haptiering the STOP, DO NOT MOVE haptice. Handshape variates from a flat hand, to one finger and a grip.

Harnessing our own capacities

A deafblind person can educate hospital staff on how to use haptices themselves. They can even have a hospital visit folder, which contains, for example, pictures of hospital haptices as

a dictionary. The nurses' feedback we have received so far is encouraging; haptices can make hospital experiences better. Along with speech, haptices can be used to provide predictive information about future procedures, and this can reassure and empower patients. Haptices can be used in different spaces, for example, in the intensive care unit and in the recovery room. Haptices can also be beneficial and reassuring for other patients such as culturally and linguistically diverse and those with other disabilities or language and comprehension problems.

The instructions needed in research are related to smooth communication, regardless of whether you can still pick up information visually or auditorily. Treatment situations are usually mentally stressful, so the use of haptices gives a feeling of attention. The need for touch is individual and is use by respect. 'When someone touches my upper arm, I get to know that my blood pressure is being taken, when I feel a heavy touch, the realization that I have to stay still', told by one deafblind person after using haptices.

Further considerations

During the recovery process, a person with deafblindness may find their energy supply very low and co-ordination of where objects are located very poor. This may be partially due to being in an unfamiliar environment. One's balance may be poorer, especially due to added tinnitus in some cases. Also blurred vision, such as after a cataract operation, may cause further balance issues. It may be a good idea to take more time to move around and find things. Otherwise, there may be more frustration and anxiety. Also, family members need to be aware of these

changes and that the person with deafblindness will need additional support and may need more touch-based communication methods, such as haptices, through the convalescence period also in the home environment. This was particularly the case during the cochlear implant recovery process, as I was only able to hear with one hearing aid, making communication extremely difficult. Another consideration could be the side effects of medication, including nausea or sleepiness, but also additional hearing or vision-related issues. Also, pain management needs considerations as pain tends to distract concentration and have a further effect on hearing and sight. In my case, I found listening to music through my portable CD (compact disc) player, and later my home hi-fi system played a contributing part to relaxing. This was also a stress relief and helped with fears and anxiety. Another contributing factor to my recovery process was to play some of the films on DVD (Digital Versatile Disc) that I could relate to and that could relax me. For that, I used my induction loop system with my hearing aid.

One-to-one communication by narrow vision field and using hearing aids

It is important to speak clearly and have suitable lighting for the environment when communicating with hearing impaired people, so they are able to lip-read. Because of the narrow field of vision, for instance, it is only possible to see the other persons face when looking directly at them. This situation becomes impossible when lighting conditions are difficult. An Usher person may sometimes need some form of confirmation that the

other person has understood their conversation. Hearing and sighted people do not have to use tactile communication. This can lead to misunderstandings, frustration and stress. During conversation, the repetition of words can be slow, and takes much energy and time.

Since hearing aids and radio microphones are only able to pick up some speech, and lip-reading can prove exhausting, it is important to understand that people with Usher have to use a large amount of energy to communicate because of the concentration required. This applies to both deaf and hearing-impaired Usher people who use sign language and lip-reading. The use of hands-on and tactile communication becomes the main source of communication, as vision deteriorates over time. However, the development of tactile and touch is a slow process if one is used to speech. Participation in group discussions may be necessary but impossible, and one-to-one contact and appropriate lighting conditions are important to ensure the Usher person is able to see and follow these interactions properly.

If lighting conditions are difficult, the deafblind manual alphabet can be used. For instance, use the alphabet for every first letter of a word, maybe B for bathroom. Confirmation and feedback can also be given through touch, tapping for yes, wrapping from side to side for no. During examination, this body contact allows the deafblind person to feel safe and to know where the other person is, for instance, touching the person's shoulder or leg. To indicate a direction of an object, their worker can guide the deafblind person's hand to the object or give the object directly to their hand. Items in the environment, such as furniture, utensils and objects, need to be kept in the same places. The healthcare person needs

to tell the Usher person where they are moving to in settings. Guidance becomes a necessary way of moving around.

Different forms of communication may have to be used in different environments. Main forms of communications are usually (Palmer & Lahtinen, 1994):

- English spoken language using different hearing aids
- Block letters onto the hand using first finger as a pen
- Spelling English deafblind alphabets onto the hand
- Signs into narrow vision field or hands-on
- Writing English language onto paper, computer, mobile phone
- Touch messages, haptices

To get attention, distance, and direction

Before speaking, you need to attract someone's attention. This will save energy, time and any misunderstanding:

- Get to that line of vision first
- Wait until the deafblind person has eye contact with you, and observe that they are aware of your position
- Touch them to show where you are positioned

During night-time, the person usually takes off their hearing aids and communication methods will change, using one of the methods mentioned earlier.

Conclusion

Following are the basic ideas for effective meeting and interaction from the perspective of a hearing and visually impaired person:

- As you approach, you can calmly touch the other person on the shoulder. You can call their name.
- State your name and your occupation. Recognizing a person by their voice can be difficult. Due to a visual impairment, the person may not always be able to distinguish themselves from the background or other professionals.
- Position yourself face to face so that lip-reading is possible. Be at an appropriate height level and distance. Do not cover your lips or speak with your head down.
- Choose an appropriate method of communication (speech, writing, sign language, block letters etc.)
- Calm and clear speech is easier to follow.
- Give the patient enough time to react and allow enough time for interaction.
- Check that the assistive devices are working (e.g. hearing aid devices are switched on).
- Check that the room has sufficient and good lighting.
- Eliminate background noise, such as TV, radio and more. If this is not possible, find a quiet room.
- Describe things and the environment, say what you are doing.
- Things can be repeated and clarified as an agreed method.
- Say when you leave the situation.

Chapter 12 explores communication from the sign language interpreter perspective.

Illustration credits: Sophie Hague, referenced from photos provided by Riitta Lahtinen

12
Communication 3: An interpreter's perspective

By Ros (Roslyn) Barnes

> Patience, a sense of humour and a positive attitude will go a long way to reduce the impact of a dual sensory impairment!
>
> Deafblind Information Australia

Introduction

As noted in earlier chapters, good communication is critical for the best patient outcomes, positive health and social experiences and better health and well-being more broadly. The strategies necessary to ensure clear communication will vary according to the type and level of sensory loss as well as the person's preferred communication method. Some people only need simple adjustments to general communication, while others involve a third party who work as an interpreter between two very different communication styles or languages.

This chapter focusses on better communication when working with interpreters for those who need them. It takes specific

training to communicate in-depth information in a signed language or a tactile language. The ways in which people with Usher syndrome communicate are not only myriad, but will likely change over time. People with Usher syndrome may be sign language users from an early age, or learn later in life. As a person with Usher syndrome's visual fields shrink, so does the capacity to see a visual sign language. A restricted (visual) frame to sign in, defined in more detail later, might become necessary. As the visual fields continue to decrease and central vision is compromised, forms of signing and communicating involving touch may be needed for communication. These forms include tactile sign languages, social-haptic communication as well as tactile alphabets, sign shortcuts and print on palm. We have asked Ros Barnes to give her perspectives as an interpreter with decades of experience.

About Ros

Ros Barnes started supporting blind children at school in a volunteer capacity in Sydney at the Royal Institute for Deaf and Blind Children (now NextSense) nearly 50 years ago, initially learning Braille and guiding techniques. Over the last 30 years, she has learned and taught Auslan, studied as an interpreter and became involved with the Usher and deafblind communities as interpreter, guide, communication guide (commguide) and teacher. She has worked extensively in New South Wales (NSW), Australia, also at national and international conferences, including two World Federation of the Deafblind conferences and Helen Keller World Conferences, and four International and European Haptics and Human Computer Interface Conferences. She now works as

an interpreter and educator using Intypreting (speech to Braille), Auslan, Visual Frame, Close Frame, Tracking, Tactile Auslan, Tactile Fingerspelling and Social-Haptics communication. This chapter is informed by Ros's expertise in the (NSW) Australian context, but there are many parallels with broader national and international health and social welfare providers.

Empathy in communication

We begin this chapter with communication's cornerstones: empathy, connection. To walk in the shoes of individuals with Usher syndrome is to see why clear, accurate communication is vital. Health policies require that qualified interpreters must be used for all people who are not fluent in spoken English or who are deaf. We explore the role of the interpreter in working with patients and healthcare/social work professionals. Because the vast diversity of losses in both hearing and vision among people with Usher syndrome gives rise to multiple communication styles, it is paramount an interpreter is experienced in the person's chosen communication style. Information and strategies are also provided to help health professionals facilitate better communication through working with interpreters.

In the shoes of a person with Usher syndrome

When you enter the door of a specialist's room, what do you see? Perhaps a specialist seated at their desk, an examination table, a chair, a procedure information form, a computer screen? Now, because you have Usher syndrome, you can only see these things dimly. Or you can't see any of them at all.

What would be the consequences of missing that visual information? What would be the impact on your ability to know where to go? How to manage moving safely through the space? What information would you miss if you could only hear the specialist speak? How would you know that what you were telling the specialist was being understood?

Notice, too, what you hear. In a specialist office, perhaps your name, a greeting and directions on where to sit, a request for test results, a nurse with a trolley preparing to take a blood sugar test or someone letting you know you dropped your phone.

What would the combined consequences be now of also losing the ability to hear these words and audio cues clearly, or at all? How would not seeing and hearing impact your ability to give informed consent? Your mental health? Your confidence? Your independence?

The combination of hearing loss and low vision has a significant impact for each individual, affecting the ability to move independently, remain connected within social relationships and communicate openly with other people. Balance disturbance – another complicating factor for those with some types and subtypes of the syndrome – also needs to be considered. Combined hearing loss and low vision contribute to fear and anxiety in unfamiliar surroundings, loss of identity, independence and knowledge of the world in general. In addition, a person's health literacy may be low as a result of fragmented information and restricted worldview.

Always use accredited interpreters

Using an accredited interpreter respects patient/client privacy and ensures the accuracy of information being conveyed. It

avoids health professionals talking to family members, sharing private information, rather than addressing the person themselves. The World Federation of the Deafblind Global Report 2018, 'Persons with deafblindness and Health', noted:

> Healthcare staff [often]…lack knowledge about the specific communication requirements of persons with deafblindness, which often leads to professionals talking to…guides or family members rather than the person themselves. This can have a serious impact, including misdiagnosis, as the person is unable to explain his or her symptoms. Furthermore, persons with deafblindness are unable to access information about proposed treatment, leading to a limited understanding of their own medical history.

For all important health and social work conversations with a person with Usher syndrome who uses signed and tactile forms of language, an accredited interpreter must be used. Conversations/consultations with a person with Usher syndrome, or indeed any patient for whom English is inaccessible or who is deaf, need to be through a professional interpreter who is experienced in the specific communication style used by the patient/client.

Do not use someone 'who knows a bit of sign' or a support worker/commguide or someone who is a family member. A strict Code of Ethics applies to all professional interpreters to ensure both confidentiality and accuracy of information, among other ethical considerations. Untrained family members or bilingual individuals generally have a limited ability to remain unbiased, or emotionally uninvolved or use interpreting strategies to make sure all parties are able to participate in the discussion.

Sarah

Sarah is a professional Auslan interpreter, booked for a consultation between a registered nurse and an older person with Usher syndrome during a routine dialysis treatment. Sarah arrived to find that the patient had been undergoing treatment for years but had never had an interpreter at a consultation before. Previously, only family members had been present and they'd organised the treatment on the patient's behalf. At the end of the consultation, the patient mentioned wistfully that they wished they knew why they had to come to the hospital all the time. Appalled, Sarah requested that the registered nurse return and asked the patient to repeat the question, which she interpreted. The nurse was also dismayed. They not only explained the current treatment and its importance to the patient, but also shed light on the diagnosis and need for dialysis. With Sarah, the nurse was able to answer questions the patient had been struggling with.

This lived experience example underscores the critical need for interpreter support for important conversations whenever a sign language-using person needs and wants one. Not infrequently others, including family, decide, diminishing patient autonomy and rights. In Australia, providing an interpreter is a free service provided by state health departments.

Law and policy

Globally, UNCRPD, specifically article 21, enshrines the right of all sign language users to an interpreter when needed. This establishes and supports freedom of expression of opinions and access to information (CRPD, 2006). Locally, in NSW, Australia, the

Ministry of Health stipulates that essential communication must be conveyed clearly and in a manner that allows the fulfillment of duty of care and informed consent.

> In any healthcare situation where communication is essential, health practitioners must engage healthcare interpreters for patients who are not fluent in English, including people who are deaf. This is critical to ensure accessible, safe and high-quality services. Health practitioners are responsible for assessing a patient's need for an interpreter and arranging an interpreter to assist. Members of the public cannot book health care interpreters directly. (2)

In the UK context, for example, the NHS health policy dictates that professional British interpreters must be used for deaf patients to ensure effective communication and uphold the (UK) Equality Act 2010 (NHS England, 2018).

There are Australian standards set by NAATI, the National Accreditation Authority for Translators and Interpreters. It is important that the health practitioner ensures that a professional interpreter is present at each consultation, and the interpreter/s is/are experienced in working with the specific communication used by the person with any single or combined sensory loss such as Usher syndrome (also known in the Australian Interpreting industry as deafblind, regardless of the level of vision or hearing loss).

People who need and use interpreters in healthcare settings do not always have access to them (Watharow, 2024). This is why this chapter has been specifically included in this book, so that

health and social care students and professionals understand the legal and ethical obligations to provide good communication and interpreter access, in addition to knowing how to work with interpreters effectively.

Usher syndrome patients and clients (and indeed, all people with dual sensory impairment or deafblindness) and their families have the right to free professional interpreters whenever they use public health services. The onus is on the health and social care practitioner to arrange a suitable interpreter to fulfill their duty of care, provide quality communication to and obtain informed consent from the patient. Simply put, asking for an interpreter experienced in the specific communication used by the patient is imperative.

Working with interpreters

Interpreters must be fluent in both the Source Language (the language used by the 'speaker') and the Target Language (used by the 'listener'). They must be able to find an equivalent phrase in each language, even when a direct interpretation does not exist, and they must be able to adjust their output to match the understanding of the recipient. For people with Usher syndrome who use Auslan (Australian Sign Language) or British Sign Language, for example, or a modified form of it, this also means the interpreter must be able to adjust the production of the signs in a way that the person with a hearing and or vision loss can still perceive their meaning.

When communicating directly with a person with Usher syndrome, there are differing preferred strategies, depending on

age, levels and types of loss, experience and previous access to support. We emphasise that all people should be asked about their preferred way/s to communicate.

Spoken communication strategies

There are many strategies that may be used by a person with Usher syndrome in isolation or in combination with interpretation. These can include:

Speaking – clearly without background distraction or at a closer distance. This may be augmented by technology (e.g. hearing aids or personal amplifiers).

Respeaking – someone listens to what has been said and respeaks to the person.

Speaking combined with lip-reading – speaking directly to and close enough to the person for them to be able to use the lip patterns during speech to complement sound reception to increase comprehension.

Lip-reading only – the person uses only the shape of the lips during speech to receive information; however, only 30–40% of English can be understood through mouth/lip patterns (Woodward & Barber, 1960) and understanding that balance is usually guesswork.

Speech to text apps – these are programmes usually on a smartphone or tablet that 'listen' to the voice of the practitioner and turn that into text on the device (e.g. Otter). The Usher patient reads the text in their preferred colours, contrast and size type. This technology is currently helpful in what it provides, but it is far from being reliably accurate, and much of the information on the screen is not exactly what the voice actually said

and sometimes is completely different! Speaking clearly and at a *slightly* slower than normal pace may help in this regard.

Speech-to-Braille apps – like the above apps, only the output is into a refreshable Braille display, which is read by the dual sensory loss person. Lines of type vary from 18–40 Braille cells. Once a line of Braille is read by the fingertips, the Usher person pushes a button and the next line appears. Again, the accuracy of the information relies on the app, so it is important to check that the patient understands what the practitioner is actually saying.

Esther

Esther heard what she thought was the ward nurse speaking to her, and using her residual hearing picked out of the ambient noise something like:

Oo_oo_o_a_o_o_a_i

By turning towards her and asking for a repeat, she was then able to lip-read the face now looking at her. She perceived:

Y__y__w_n__n_t_vv__r_r_w_j

Putting the two together, she worked out:

Yoo_yoo_won_tan_ tovv_ora_ra_wii

Then she had to consider which lip patterns looked similar that made more sense:

Doo_yoo_won_tan_koff_ora_sanwich

Applying context and common sense, she hazarded a guess at:

Do you want any coffee or a sandwich?

She replied in the affirmative, hoping she'd guessed correctly and this was what she was being offered.

Visual communication strategies

Few practitioners have enough fluency in Auslan to speak directly to the Usher patient or their family. This is where requesting an interpreter is vital and required by law, as stated by the NSW Health policy quoted earlier. Visual communication strategies used by experienced interpreters and Usher patients and families include:

Auslan – using a visual, spatial language rather than a spoken language. Auslan is a language in its own right and is used by the Australian deaf and deafblind community. It has its own unique structure and grammar and is not a visual representation of English. Rather than follow a linear framework such as English, it uses space and movement in 3D (three-dimensional) as well as facial expressions, body language and posture in combination with signs (specific handshapes, orientation, locations and movement) to express any concept available in another language.

Close vision – positioned in closer proximity for an Auslan user with more vision loss.

Visual frame – for an Auslan user with a more pronounced vision loss, Auslan is used within a specific spatial frame at a specific distance that includes the signer's face but excludes signs that are located outside this frame, for example, DOG (patting the leg), LAZY (located on the hip) or STARS (signed high above the head). These signs are modified or swapped with another sign located within the visual frame to convey the same meaning.

Tactile Communication Strategies

The use of touch in combination with other strategies is common and may include:

Tracking – instead of watching the signer from a distance, the person may hold the signer's wrists to help them keep the hands and face in view. This further restricts the signs that can be made, and more modification is required.

Tactile Auslan – with very low vision or none at all, the Usher person places their hands onto the signer's hands and feels the signs. Emotions and non-manual features must be conveyed through the hands as facial expressions are inaccessible.

Tactile fingerspelling (sometimes called the deafblind manual alphabet) – a tactile version of a spoken language where the words are spelled out into the hand. This is usually used by non-Auslan users, or people who require information or specific terminology in an English format (e.g. procedure or medication names). As with Tactile Auslan, emotions and non-manual features must be conveyed through touch while spelling the words.

Shortcuts – a system of touch cues on the hand that represents a sign rather than a letter. This is used in conjunction with Tactile fingerspelling (e.g. YES, NO).

Print on palm – a tactile version of the English alphabet formed by writing capital letters on the palm of the Usher person. This laboriously slow method of communication is sometimes useful when a person or a healthcare practitioner needs to communicate simple requests when no other method is known.

Social-haptic communication – a set of tactile cues used to convey real time messages such as environmental information, personal reactions in interactions, locations of services or amenities, and room/plate/visual layouts. For example, conveying the location of the bathroom in a ward, or the reactions of a doctor if they are nodding, frowning, pleased, etc. See the previous Chapter 11 for more on this.

While the examples used are Auslan, the visual and tactile communication strategies are also true of other sign languages.

Note on deaf relay interpreters

In some cases, the language used by the person with Usher syndrome may be of such specific nature that a second interpreter is essential to the chain of communication. This is likely to be a deaf interpreter, who is a native Auslan user and able to take the Auslan given by the 'hearing' interpreter, and further interpret into the more specific language required by the Usher person. This may be applicable in cases of brain injury or mental health challenges, or when the education of the Usher person was so inaccessible that only a basic concept of language was acquired.

Deafblind community and culture

It is worth understanding that Auslan users will consider themselves part of the Deaf community, and/or the Deafblind community. This is part of the rich cultural and linguistic diversity of society. Deafblind culture has its own language and sense of community with a cultural identity, a feeling of belonging to a specific community and observing social conventions, norms and etiquette. Interpreters are well aware of the etiquette and therefore can also act as a 'cultural bridge' as well as a linguistic bridge.

Some useful examples of appropriate etiquette include:

Appropriate attention-getting – waving in their visual space, a light touch on upper arm or forearm, stamping on the floor (wooden floor).

For the deaf community, flashing lights are a common way to get attention, but for a deafblind person this may be inappropriate due to a sensitivity to glare.

Keep conversation concise – the information in a health or social care setting can be overwhelming and the wider community etiquette of 'niceties' before the details can be a further unnecessary burden.

Let the deafblind person know who is present – a 'roll call' is essential to let them know who is in the room and where they are.

Sight and sound terms – the English way of asking questions, such as 'Have you seen the results?' Or 'Have you heard from your specialist recently?' is not offensive.

Understand the conveyance of visual information – the interpreter is supplying both what is seen as well as spoken, such as the specialist's perusal of a computer screen, preparing for a procedure, writing notes, so that the patient knows what is going on between conversations.

Taboos

Do not hold a signer's hands down – this is considered the equivalent of holding a hand over someone's mouth.

Do not say 'oh don't worry about it' if something is not understood – if it was worth saying, it is worth conveying properly.

Do not talk about them to the interpreter – the interpreter is obligated to interpret everything said and done, as the patient has the right to know everything available to a hearing-sighted person.

Setting up space for communication

Because of the different physical requirements of each communication strategy, the setup of the interpreter within the space will differ.

Auslan or other sign languages– with enough vision to see at that distance, a person with Usher syndrome needs the interpreter to be positioned beside the practitioner to allow them to see both the practitioner and the interpreter in order to access both the spoken information and the facial expressions and body language of the practitioner.

It is important for the practitioner to look at the person with Usher syndrome and ask about their communication preferences. Then, it is important to address them directly person to person. It is inappropriate to address the interpreter and ask the interpreter to 'tell them' the information – speak directly to the patient or client.

The sign-language-using person will be looking at the interpreter for the signed interpretation, but also watching the practitioner and addressing their signed replies to them. As they do this, the interpreter will speak aloud these replies in first person. For example:

Practitioner: Have you had any blood tests recently?

Interpreter: signs the equivalent in Auslan (RECENTLY YOU BLOOD TEST HAVE?)

Usher patient: signs a reply in Auslan (YES PAST FOUR MONTHS, DON'T-KNOW NOT-YET)

Interpreter: Yes, I had one about 4 FOUR months ago but I haven't yet seen the results.

Practitioner: I can retrieve the results and let you know – where did you have the test done?

Interpreter: signs the equivalent in Auslan (I-CAN COLLECT, LET-YOU-KNOW – TEST WHERE?)

Usher patient: signs a reply in Auslan (THIS HOSPITAL HERE, WANT-ME GO-DOWN ASK-THEM?)

Interpreter: Here at this hospital, shall I go down and get the results?

Practitioner: Ok, no I can access your results from here…

Note: the text in capitals is a crude, written representation of signs used in a 3D modality but without the nuances of facial expression, posture or movement.

Close vision/visual frame – for this strategy, the interpreter will likely need to sit closer to the patient, who may not be able to see the practitioner as well. However, the same procedure would be followed, so it is important for the practitioner to still speak directly to the patient, who will receive the signing from the interpreter, and reply in a similar manner, just sitting closer.

Tracking/tactile Auslan – for this strategy, the interpreter will need to sit close enough for the Usher person to touch and feel their hands. Usually, a small table is used to support the arms of both interpreter and person, especially in longer consultations or conferences. Again, the same procedure is followed, with the practitioner again speaking directly to the Usher person.

Note: Interpreting into a tactile language is draining for both an interpreter and a person with Usher syndrome. Shorter consultations may be attended by one interpreter, but for anything longer than approximately half an hour, two interpreters should be working together, swapping at 10–15 minute intervals to maintain clarity of communication. The Usher person will also need breaks, as assimilating new information through a tactile language is also challenging.

Communication strategies when an interpreter isn't present

Interpreters are human too, and unable to work 24/7. In reality, a person with Usher syndrome will not likely have an interpreter for extended periods of time, and so it's important to be aware of communication strategies that can be used for basic everyday care. Here are some strategies you can use when appropriate. (It's also important that if the interpreter is coming at 10 am that health and social care professionals prioritise that communication and are also present at that time.)

First meeting

The first moment that communication starts is likely when the person arrives at a facility or is in the waiting room. Simple strategies can help alert a medical practitioner to the communication required and make the interaction more comfortable for both people.

Adaptations to approach

Especially when someone is unfamiliar with a person who has Usher syndrome, some strategies in approaching the patient could include:

- Avoid calling them from a reception desk or from a distance.
- Try approaching them from the front, within any vision they may be able to use.
- Wave gently to ascertain if any vision is usable.
- Say hello, using their name and identifying yourself (e.g. Hi Jenny, it's Silvia, I'm a nurse).
- Speak clearly at a normal pace, without over-enunciating or shouting.
- If a sentence is not understood, rephrase rather than repeat the same sentence.
- Use eye contact, body language and gesture, pointing and facial expressions.
- Ask if a particular distance or direction is easier for them to see.
- If there appears to be no reaction to voice or vision, it is appropriate to ask permission to touch first, but if no response, try a light touch, usually on upper arm or forearm, with the back of the hand, to get attention.
- If an Usher person raises a hand in response to touch, they are likely to either raise palm upwards, requesting tactile fingerspelling, or palm downwards, requesting a tactile sign language. (If palm upwards, 'wave' on their palm, then try slow Print on Palm. If palm downwards, gently turn their hand over and do likewise, but understand the urgency of organising an interpreter.)
- Using paper and pen, a whiteboard, texting on a smartphone or tablet, or print on palm may be an option for slow, basic communication if the person does not have an interpreter or a support worker accompanying them but has sufficient vision. This is a temporary measure only.
- Allow them time if needed to process information.

- Check that they have understood key information.
- Limit prolonged concentration if they are struggling to understand.

Adaptations to environment

Some simple strategies can help increase comprehension when the sight or hearing loss is mild. These include, in consultation with the patient,

- Adjusting light levels, brighter for those using visual cues to compensate for a hearing loss, or dimmer for those for whom glare makes vision difficult or painful.
- Seating such that ambient light is coming from behind the patient rather than from behind the practitioner, which then makes their face harder to see.
- A simple and uncluttered background behind the practitioner, devoid of posters, equipment racks, flashing lights or screens.
- Plain coloured garments rather than 'busy' patterns or stripes.
- Consider wearing strong-coloured lipstick to help someone who lip-reads, and remove masks.
- Masks make communication difficult, both for lip-readers but also for Auslan users.
- Minimisation of background noise, such as fans, air conditioners, equipment hum, TVs or music, outside traffic or construction work.
- Soft surfaces that reduce echo and reverb, such as carpets.

Vincent

Vincent, a non-verbal sign language user, had been taken by ambulance to the Accident and Emergency department at his

local hospital. Vincent is able to do basic written communication via a computer tablet. Vincent had been left in the waiting room but didn't recognise any of it due to recent renovations. The next morning, via the tablet, he asked a passing nurse who was doing waiting room observations why he hadn't been seen yet. The nurse returned to type in reply that his name had been taken off the list because he had been called twice and not responded. This was despite the paramedics handing over the information that Vincent is profoundly deaf with vision loss. After an apology and being ushered in, there was no interpreter made available. Vincent struggled with the harsh lighting, the examination was brief despite severe abdominal pain, and he was misdiagnosed and sent home with incorrect information.

Sometime later, he had to return to hospital with the same symptoms. During the 6th day, exhausted after being moved 7 times without knowing why, missing dietary-specific meals because they were planned for a previous ward, unable to eat the allergen-loaded basic meals, he lay in a darkened room trying to rest. A new doctor came in wearing a mask, without an interpreter, flicked on the light, and was shouting loudly to communicate. Fortunately, Vincent's support worker had just arrived and was able to turn off the light, explain about the masks, the futility of shouting, and the impact of glare on the low vision. They found Vincent's sunglasses before the light was turned back on. But the consultation still went ahead without an interpreter, and Vincent had little understanding of the test he was told to undertake, as the support worker did not have sufficient language skills to function as an interpreter.

Communication strategies to try

Speaking/respeaking – clearly and repeating if necessary or rephrasing to use a different set of syllables and sounds.

Lip-reading – face the person, ensure you have a face clear of masks, facial hair, pens/pencils, food, hands, etc, and always check comprehension.

Writing – some Usher people can access writing, consider a thick black felt pen and large writing which may be more easily read. Having a supply of paper and pens on hand would be helpful. Writing on a tablet may also allow changing to a more easily read contrast, such as yellow or white type on black background, or black type on purple background – ask for their preference.

Print on palm – with your index finger, trace letters onto the palm of the Usher person using simple capital letters. Make sure the letters are large enough and the person understands what they are.

Tactile fingerspelling – based on the Auslan alphabet, this simple modified alphabet on the following pages is easy to learn and can be used with most Usher Auslan users.

Pivoting – If a method being used is not working, don't shout louder, don't get frustrated -pivot to another method

Room orientation – with the hand of the person with Usher syndrome on yours, you can gesture in a direction and state what is there, for example, (point) bathroom, (point) cupboard, (point) door, (point) nurse. This can also be used for food on a plate. Eg (point) bread, (point) meat, (point) mashed potato, (point) jelly, (point) fruit juice.

Social haptics – learning some simple haptics goes a long way in patient care, such as I'm here, who I am, yes, no, question,

blood pressure test, injection, medication, emergency evacuation and more.

Isabel

Isabel was admitted to hospital for surgery. When she arrived in the ward with her interpreter, the nurse on duty took the time to learn the best way to communicate with her, her preferred communication style both with and without her hearing aids. The anxiety about being woken by a stranger in a strange place for overnight blood tests was assuaged by an agreed strategy of:

1. A light touch on the shoulder to waken her.
2. A cross drawn on the upper arm to let her know it was a nurse.
3. A light pressure on the webbing of the thumb to let her know an injection was forthcoming.
4. A touch on the index finger let her know where the injection site would be, from which Isabel deduced it was a blood sugar test.
5. Isabel agreed to nod and hold out her hand, indicating consent. This is an example where hospital haptics can be used in conjunction with other forms of tactile and visual communication.
6. After the blood test, the nurse would draw a circle on her palm and a question mark, which constituted a question regarding any pain medication needed.
7. Isabel would sign a YES or NO.
8. The nurse would tap her arm a few times to indicate she understood.

This preplanned strategy was also noted in her file, conveyed to the other nurses on the ward and explained to the next shift

during handover. As a result, Isabel felt much more positive about her stay in hospital and was calm throughout the experience.

Seek to understand the person

Be person-centred: Read their files, ask them what is best for them, provide information and consent forms in accessible formats and be open to learning more about their experience. Be aware that a symptom may not be related to their disability.

People with sensory loss/es are often misconstrued or even misdiagnosed as being affected by dementia, being grumpy or difficult to manage, or are neglected usually through misunderstanding or lack of communication. Diagnostic errors can occur if communication needs are not met and an interpreter is not provided.

Robert

Robert had a mini mental state examination done in hospital. No interpreter was booked and the intern didn't even know Robert had Usher syndrome. As a result, Robert misunderstood most of the questions and received a very low score. The doctors informed his wife that Robert really needed to be in a supported residential aged care facility. Happily, his wife was sceptical that someone who could do a cryptic crossword in under 30 minutes could have dementia and she queried the result, demanding a repeat with an interpreter present.

Vance

As an Auslan user, Vance identified as being deafblind. However, the staff in the emergency department refused to believe he was

blind at all, as he was using text on his phone to try to communicate. They refused to type answers, spoke to him despite his requests to use text or writing, and refused to organise an interpreter. The next day, as this behaviour continued, Vance became more and more frustrated and agitated and refused to do any more tests not knowing what they were for or what to do. Now severely anxious and still in pain, he asked for discharge in the afternoon. The next day, the support worker arrived at Vance's home to find a canula still in Vance's arm.

Ben

Ben was in hospital after a mild stroke. While the nursing staff were aware of his being totally deaf and totally blind, no one informed the catering team. Each meal was placed on the table beside his bed and then was removed half an hour later untouched. When a friend came to visit, he complained that he had not been fed for two days. On further investigation, his friend found this was simply because no one had told Ben that the food was there. Catering claimed they were forbidden to touch a patient, even though they could have simply guided his hand to the food tray. Without an interpreter present, he couldn't ask, and slowly starved during his 'care'.

Neither did anyone explain to Ben's friend that the stroke had caused paralysis and numbness in Ben's left side, which happened to be the hand the Ben used to receive tactile fingerspelling. Finally, Ben's lack of response to questions made sense to his friend, as he had been spelling into a hand that could no longer feel the language he needed to understand. This also meant that until that revelation, Ben didn't know who was there.

Boosting communication skills

The best way to learn more about communication with people with dual sensory impairment such as Usher syndrome is to meet them, talk to others who know and attend courses in communicating in Auslan or other relevant sign languages.

Conclusion

Communication in our community is a fundamental right for all, including those with dual sensory impairment or deafblindness of any cause. Usher syndrome is a spectrum of loss of vision combined with hearing loss, and this can severely compromise the ability to communicate with the wider community. Strategies are available to minimise this communication loss, and the onus is on professionals in health and social care to provide these strategies to fulfill their obligations to duty of care and best practice. Book the right accredited interpreter for the person's communication needs; work with the interpreter to ensure access to information and answering questions. Pivot to other methods if an interpreter is not available. Also a fundamental right for all is better health and well-being. Chapter 13 showcases the ways health and social care professionals and practitioners can work collaboratively with each other and Usher syndrome people and families.

13
Better health and well-being

Introduction

Usher syndrome is a condition that involves varying degrees of both hearing and vision loss, sometimes with a balance disorder accompanying these. As seen in Chapters 4, 8 and 9, sensory losses impact health and well-being in complex and challenging ways. What can we do to reduce these risks and threats?

The triple threats

Three key threats to health and well-being for people with Usher syndrome (and indeed, any disability) are unenforced or inadequate protections, policies, systems and institutions; professional and practitioner and service failings; and finally, a lack or inadequacy of individual and family/carer support and resources that can, all singly or in combination, compromise health and well-being.

From the 2024 Consumer, Carer, Professional and Practitioner Survey of the Needs of Adults with Dual Sensory Impairment-Deafblindness (Watharow, 2024), the following quotes embody the lack of recognition of dual loss, the fragmentation of care, the communication failures and the lack of accessible information for individuals and family/carers:

- On lack of recognition: 'I went blind slowly and deaf slowly, but nobody explained that the two together would change everything.'
- On fragmented care: 'Eye clinic agrees I'm blind, audiology agrees I'm deaf—yet the system treats me like two separate patients.'
- On communication failures: 'It's more important that the professionals change the way that they communicate and work, not the deafblind people.' 'Talk to me, not my support worker.' 'I didn't understand anything the doctor said' was common, or the specialist, audiologist, ophthalmologist and so on.
- On useful, accessible information for individuals and families/carers: 'Give me a handbook that tells me how to keep cooking, not a PhD on retinal cells.'

The variability of types and subtypes and changeability over time in Usher syndrome means all three actors need to be flexible, creative and reactive in solutions building: Complicating health and well-being journeys is multiple disability (20–75% of all those with combined hearing and sight loss have additional impairments; WFDB, 2018). And then there are the grief journeys an individual (and their family) makes as they adjust to living with sensory losses. These are highly diverse, sometimes protracted, lived experiences of Usher syndrome.

An inclusive approach to better health and well-being: Eight pillars

We all have a part to play in better health and well-being: from systems and institutions; policymakers, professionals and practitioners; and individuals and families. The key pillars to a more

inclusive approach are recognition, communication, accessibility, collaborative care, preparedness, supporting individuals and families/carers, empowerment and breaking binaries.

We present these pillars with explanatory notes. We also include a special note of the grief journey and introduce the concept of ontological insecurity as a complicating factor to better psycho-emotional well-being.

1. Recognition

- Deafblindness (including Usher syndrome) must be acknowledged as a distinct and significant condition, not merely a subset of single-sensory impairments. This is a systemic issue. Australia doesn't have this recognition yet, although England and the European Union do.
- Awareness campaigns and policy reforms can highlight specific needs and secure funding for specialised support.
- Dedicated specialised diagnostic and support services to help people with Usher syndrome navigate the complexities and challenges. Collaborative care is better care.
- Resources, training and credentialled support for all life stages: early intervention, education, work, relationships and psycho-emotional well-being.
- Understanding the vast complexities and challenges of Usher syndrome is important so that services are specialised and credentialled. There is no one-size-fits-all or tick box lists that will generate better health and well-being.

2. Communication

From the 2024 survey of 223 people living or working with dual sensory impairment, including over 60 with Usher syndrome,

the principal request for professionals and practitioners was: be a better communicator.

The first step is always:

ASK: Acquire Specific Knowledge

1. How do you prefer to communicate?
2. What tools work for you?
3. What aspects of the environment help or hinder you?

THEN follow up with

- Learning basic tips, for example, face the person and speak at a moderate pace.
- Having interpreters booked in advance for known appointments.
- Knowing how to communicate with interpreters as relayers (see Chapter 12).
- Ensuring signage and documentation are offered in multiple formats.
- 'ASK' also means practitioners themselves must acquire the specific knowledge and skills to communicate effectively with people with Usher syndrome. For instance, undertaking online courses:
    - The DSI Project's Dual Sensory Impairment for Health Professionals module
    - Birmingham City University's Certificate in Professional Studies (Deafblind Studies) course
    - Deafblind International Resources
- (See resources appendix for more information.)

3. Accessibility

- Provide healthcare information in multiple formats as a standard practice.

- Provide alternative ways for people who can't use the phone to contact health caregivers.
- Provide alternatives to online registrations, forms and booking platforms for those who cannot use them.
- Provide accessible-to-the-individual consent forms.
- Ensure physical environments are well marked, well lit and, when possible, free of excessive noise or visual clutter.
- Implement robust training for health professionals on how to identify people with dual sensory impairment, including Usher syndrome and adapt communication methods.
- Low-Cost Supports:
    - Personal Amplifiers
    - Telephone Amplifiers
    - Captioning Services
    - Magnifiers and Bold Markers
    - High-Contrast Labelling
    - Screen Reading Software
- Orientation and Mobility Tools:
    - Long Canes or Support Canes
    - Tactile Surfaces and Markers
    - GPS Apps

An important note on relational aspects

Whether simple or more sophisticated, accessibility tools and devices for sensory losses should be chosen based on the individual's preference and abilities. Support and funding for acquiring devices, training and troubleshooting is often as vital as the technology itself. Family, support workers and carers need training too so they can assist as needed. These relational aspects of accessibility are incredibly important and invariably overlooked.

4. Collaborative care

Building 'Super Teams': People thrive when they have a network of practitioners, carers and supports who co-ordinate care and address challenges holistically. This network is sometimes referred to as a 'super team', comprising:

1. Healthcare Professionals: General practitioners, audiologists, ophthalmologists, mental health specialists and allied health workers (e.g. occupational therapists, orientation and mobility instructors, interpreters).
2. Social Workers/Case Managers: Individuals who understand social welfare systems can link the person with Usher syndrome to relevant benefits or programmes and co-ordinate support services.
3. Rehabilitation and Support Agencies: Local deafblind organisations or agencies focusing on hearing and vision services in combination are preferred. Single-sensory loss organisations need to do more to identify and support those with dual loss.
4. Family and Friends: Spouses, parents, siblings or friends who often provide day-to-day support, emotional backing and advocacy.

In addition, collaborative care is fostered by:

- Extended consultations: it is well established that relaying information in accessible-to-the-individual ways takes more time, hence consultations may need to be twice as long (Möller, 2005) or spread over a number of appointments.
- Involvement of persons with Usher syndrome in the planning, evaluation and feedback loops of care teams and services that affect them.

A key function of collaborative care should be to improve health literacy. Health literacy refers to an individual's ability to obtain, process and understand basic health information and services so they can make informed decisions (WHO, 2021). For those with Usher syndrome, barriers to health literacy can be especially pronounced, particularly in later stages, with very low vision and severe to profound hearing losses.

- Create communication-friendly consultation environments (e.g. reduce noise, eliminate glare).
- ASK what the patient needs and then provide it: Remove face masks where possible to decrease muffling and improve lip-reading; pivot to another method if there is communication difficulty, provide accessible information and multiple formats.
- Provide Multiple Formats: Offer documents in large print, Braille or digital text (can be converted to speech or enlarged on-screen). Include plain language summaries rather than medical jargon.
- Use Visual and Tactile Aids: For those who have some residual vision, pictures or diagrams in high contrast can be helpful. For those with little or no vision, a tactile model or a raised line drawing can communicate critical information about anatomy, procedures or assistive devices.
- Teach-Back Method: Encourage healthcare workers to use a process where they provide information, then ask the patient to repeat it in their own words. This ensures the message was delivered clearly.
- Create Clear Pathways in Clinical Settings: Ensure that waiting rooms and consultation spaces are signposted well, well lit, easy to navigate and not overly noisy. Mark floors, steps and other transitions with high-contrast and tactile indicators.

- Involve Disability Advocates and Peers: Trained peer mentors who also live with deafblindness can provide invaluable first-hand advice on how to interpret health information, access technology and self-advocate.

5. Preparedness

Systems and health professionals can adjust procedures and practices so that care is person-centred, disability aware and safer. Individuals with Usher syndrome, with support from family or professionals, can develop personal hospital kits, use wristbands, have personal emergency plans and sustainability kits, maintain updated medical summaries and use technologies that facilitate timely self-monitoring.

- Healthcare systems can adapt triage protocols to quickly identify and help people with Usher syndrome; offer interpreters or supportive technologies; and schedule longer consultations to avoid rushed and unsafe care.
- Preventative care is critical for long-term health and well-being. People with significant dual sensory losses may need support to make appointments, travel to healthcare sites, relay communication and access information on screening, immunisations and check-ups.
- Monitor vision, hearing and balance regularly.
- Use technology and other aids to assist in managing one's own health, for example, Smartphone apps that track heart rate, blood sugar or medication schedules can be set with vibration alerts or integrated with screen readers.
- Assemble a going-to-hospital kit.
- Use wristbands (see Figure 1):

  - Patient choice
  - Portable
  - Low cost
  - An immediate visual identifier for healthcare staff
  - Useful in other settings such as airports and shops
- Build a personal emergency plan and keep a sustainability kit (bottled water, disinfectant, a week's supply of medications and food, power banks etc.).
  - Link to Collaborating 4 Inclusion Person-Centred Emergency Preparedness website: https://collaborating4inclusion.org/pcep/
- Learn the social-haptic communication emergency sign (a large cross drawn on the back that signifies: 'come with me, hold my arm and I will explain later what the emergency is'; see Figure 2).
- Foster community networks with neighbours, family, friends and community services so that no one is left behind in a personal emergency or disaster.

6. Supporting individuals and families/carers

Health and social care professionals can support individuals and families/carers to minimise health and well-being risks. Individuals can harness their own capabilities too, helping not only themselves but their peers. In our survey, overwhelmingly, people living with Usher syndrome (and other forms of dual sensory impairment – deafblindness) wanted to know how to live a good life from those who were walking this same journey.

Figure 1: Three bright orange wristbands with DEAFBLIND, VISION IMPAIRED and HEARING LOSS printed on them in navy text.

- Peer Support: Peer support groups, individuals, physical meet-ups, online forums and local deafblind associations can provide a space to share experiences, tips and coping strategies. Joining these communities can be a powerful way to learn about new assistive technologies, health programmes or personal strategies that others use.

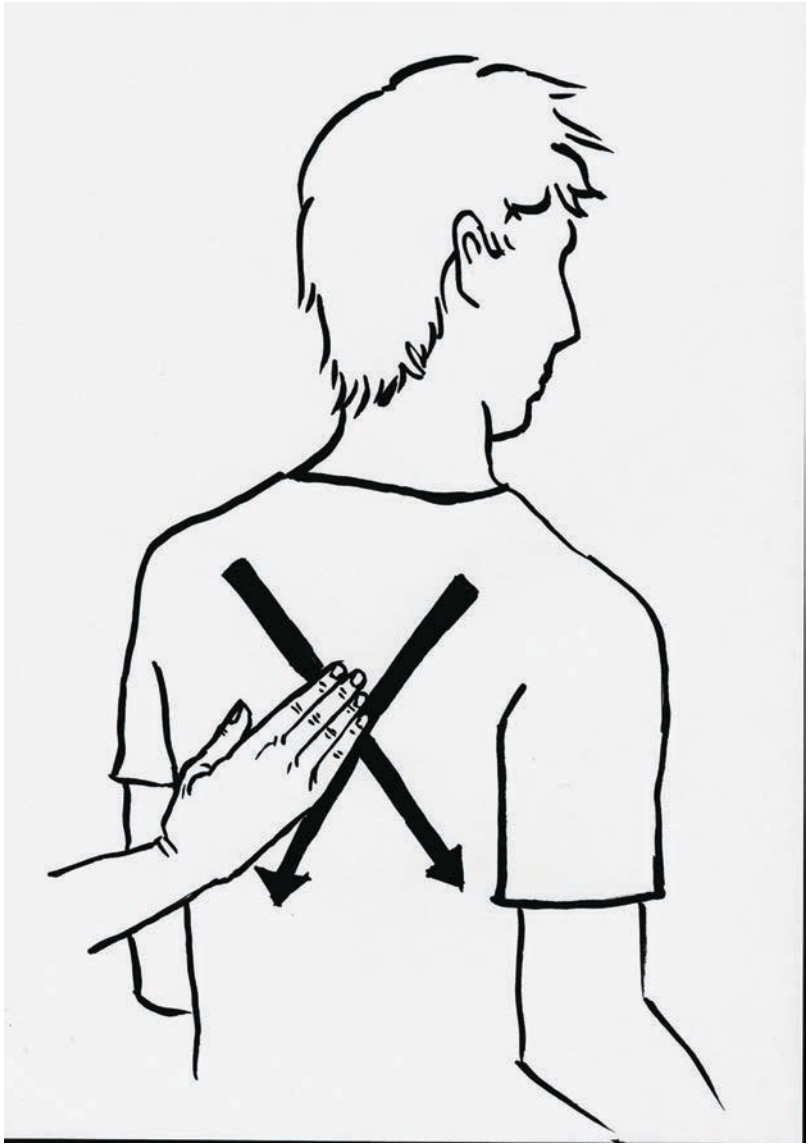


Figure 2: Social haptic communication emergency haptice.

Peer support also fosters an environment where the emotional aspects of living with DSI can be safely and openly discussed.

- Written or Digital Exchanges: Real-time, text-based conversations or speech to text solutions can be used, if that's more comfortable, such as WhatsApp. These can also be used for peer support and communication with disabled people's organisations and are a cheap, inclusive option for many too.
- Reliable Routines: Consistent schedules or daily habits help reduce disorientation.
- Clear Environmental Cues: Label spaces with tactile or high-contrast markings, keep furniture arrangements consistent and avoid creating sensory chaos with blaring background music or rapidly changing lights.
- Support Mental Health and Emotional Well-Being: Living with Usher syndrome affects more than just access to information; it also impacts emotional and psychological well-being. The grief journey can be anticipatory and prolonged, and adjustment to a new degenerative episode can occur before one has adjusted to the previous one (Gullacksen et al., 2011). Isolation, frustration or stigma can accumulate into chronic stress. Fortunately, there are strategies that individuals, families and professionals can adopt to safeguard mental health.

If someone exhibits signs of persistent sadness, loss of interest in usual activities or heightened worry, it is crucial to seek mental health support that is tailored to their communication needs.

7. Empowerment

- Encourage self-advocacy skills, knowing one's legal rights, local resources and how to express sensory and communication needs in new situations.
- Support mental health through counselling, peer networks and recognition of the psychological toll that isolation or stigma can impose.
- Respect each individual's identity and choices regarding devices, interpreters or communication modes, acknowledging that some prefer hearing aids while others do not and that some rely entirely on tactile sign or Braille.
- Support people and families to give feedback and critique care by providing accessible evaluation methods.
- If a patient with Usher syndrome or indeed a disability experiences poor service, negative treatment or didn't understand what was happening during treatment, give this feedback to the health centre, hospital or other institution. This is important data that tells the hospital the areas they need to improve on. Without feedback, the experience doesn't get counted, so the system doesn't change.
- If you have a great experience, give that feedback too. Tell the health centre, hospital or other institution what they did right. This increases the chances that they will keep doing the positive things.
- The supports needed to understand healthcare and treatments are the same supports needed to ask questions and critique care. Health and social care staff, family and carers, interpreters and support workers may be needed to support a person with Usher syndrome to be authentically part of shared decision-making and sharing their patient experiences.

And if a person with Usher syndrome is interested in being a part of making changes in the systems, they can:

- Join a disabled people's organisation.
- Get involved with local community hubs.
- Get involved with research as a consumer advisor, or a co-researcher or lead research yourself. So much needs evidence to show what works and what doesn't for people with a combined hearing and sight loss.
- Reach out to the Department of Health and ask to be advised of opportunities to be a consumer advisor on their committees and projects.
- Reach out to local hospitals and universities asking if you can be involved in teaching students in healthcare how to communicate with people with Usher syndrome.

8. Breaking binaries

It is important to note that people with Usher syndrome don't occupy exclusive roles as service users, patients, clients and the cared-for. We can be service providers, peer supports, professionals, practitioners or policymakers too. You see this in the authors of this book. Our lived experiences and lived expertise makes us important sources of knowledge in health and well-being research and translation. Annmaree is a medical practitioner, therapist, researcher and writer. Emma is a peer support leader, head of Usher services, writer, qualified trainer, assessor, and advocate. Emily has immense lived knowledge as a parent, a founder and CEO of a disabled people's organisation, manager of the first collaborative care clinic for Usher syndrome in Australia, researcher and writer.

A special note: Understanding and supporting the grief journey

The diagnosis of single and dual sensory impairment/loss is a crisis point for most. Usher syndrome involves progressive deterioration of sight and sometimes hearing. This means there will be many crisis points in a life, each needing the navigation of grief and then adjustment to the 'new normal'. And it is not uncommon for a new loss to present itself before the previous/current one has been adjusted to. The process may take weeks, months, years and even decades (Gullacksen et al., 2011). Grief doesn't necessarily begin with the loss or degenerative episode; it may be anticipatory: When will I go blind? What will happen? How will I cope? Who will help?

It's unpredictable

The grief associated with sensory loss/es doesn't just occur at the time of the loss. Grief can be triggered at any time by a memory, a sight, a sound, a lost capability or a new need to get assistance to do something that was once an independent action. Going to the movies and suddenly not following the action, even with the aid of subtitles, can be triggering. Being adrift at a party when you don't understand what anyone is saying can trigger sadness. Struggling to read larger and larger fonts. Sometimes you don't need a triggering action or event, but the mere thought of what lies ahead. We don't believe that we will cope with these yet to be experienced losses, but our peers around us will tell us that we can, and we do. It may take time. And sometimes, one of the things that people with Usher syndrome need, according to the

lived experience survey we conducted (Watharow, 2024), is for the carers, family and the supports around them to sit quietly and acknowledge the depth and breadth of the sensory loss/es. We don't always benefit from having 'solutions' and technology thrown at us. It may take time for us to regroup and gather ourselves one foot after the other, one day at a time, while thinking 'I can get through this.'

It's complicated

Grief reactions are contingent: We don't experience the grief of sensory loss/es in isolation; they can be layered with the practical difficulties of learning to live with sensory loss or deterioration; there may be changes in working life and relationships; not everyone will respond well to you; the stresses of a normal life still happen even when you are struggling with Usher impacts; health and well-being may deteriorate, for example, after an accident or fall. If you can't work anymore, then that can bring financial and social disadvantage alongside the health and well-being effects; violence rates are increased for people with disability generally and with dual sensory loss in particular (WFDB, 2018). The world may be perceived as hostile.

A person with Usher needs to navigate the practical realities of decreased access to information, potential communication difficulties and challenges to safe orientation and mobility. The people and society around them might be unaware or ill informed ('Why don't you just get a cochlear implant? That'll fix ya'), they might be prejudiced and unhelpful. One way to understand some of the forces that complicate the grief journey is to think about the additive impact of ontological insecurity. Ontological

insecurity is the loss of the fundamental sense of safety (Laing, 1965; Giddens 1991).

Ontological security: The sense of safety

This is the desired state of trust, reliability and predictability of people, places, things and information, which promotes the sense of knowing what is going on. Knowing what is going on makes it easier to feel calm, to solve problems and respond to emergencies, for example. Ontological security is therefore a fundamental sense of safety. Sensory impairments can by their very nature upend this sense of safety, trust and predictability.

Security of the self – with social relations, experiences and the environment – is tightly interwoven with trustworthy people, reliable information and predictability of the environment and objects around us. Erosion of this security puts people at risk of harm. It is not the sensory loss/es and impairments themselves.

Laing, in his landmark book *The Divided Self* (1965), introduced the sociological concept of ontological security within the context of people with mental illness. Laing reflects that a person who feels secure, safe and informed can navigate 'the hazards of life' (1965, p. 39). The consequences of an ontologically insecure position, however, are 'greater anxieties and dangers' (1965, p. 67). In *Modernity and Self-Identity: Self and Society in the Late Modern Age*, Giddens (1991) affirms that confidence; constancy; and trust in people, information and the environment are essential for ontological security. Without these, anxieties and fears prevail, and the world is experienced as unsafe and psycho-emotionally perilous.

People with Usher syndrome live with reduced information and increased uncertainty. This, in turn, negatively affects self-confidence, because people with impaired or absent sight and/or hearing cannot always trust what they see and hear.

Ontological security requires reliable information. Accessible-to-the-individual communication thus becomes critical to people with sensory loss/es to build and maintain ontological security. Failure to provide this results in what Giddens describes as 'chaos' and a 'flooding in of anxiety' (1991, p. 36).

For people with Usher syndrome trying to navigate grief reactions, these ontological insecurities are in addition to those of ordinary daily life, other intersectional factors, intercurrent illnesses and injuries. This accumulation threatens their safety, health and well-being in a myriad of ways.

The link between ontological insecurity and increased risk of mental health harms is the basis of Laing's (1965) work and is continued by Giddens (1991). Many researchers have since examined this connection in the context of sensory losses, showing that ontological insecurity can lead to anxieties, distress, depression, isolation and suicidal ideation. Communication difficulties, such as in certain head and neck cancers, can result in ontological insecurities and distress (Heine et al., 2002; Crossley, 2005). Deaf people can experience ontological uncertainties, depending on the contingencies of how they are interpreted (Young et al., 2019). For those living with Usher syndrome, the insecurities of sensory degeneration are also linked with depression and suicidal ideation (Miner, 1997) or psycho-emotional distress (Ellis & Hodges, 2013). Several authors note that the degeneration of senses, as seen in Usher syndrome, is accompanied by difficulties

in personal relationships and with an increasingly hostile environment, (Danermark & Möller, 2008; Hersh, 2013a; Miner, 1995; Schneider, 2006; Wahlqvist et al., 2015). Watharow (2021) demonstrated that poor hospital experiences perpetuated ontological insecurity and thus psycho-emotional harms. The threat of constant change unsettles security and necessitates frequent psycho-emotional adjustment (Gullacksen et al., 2011). It is our contention that grief reactions are thus complicated by the consequences of ontological insecurity.

This all means that wider society, its systems, institutions, professional, policies and practitioners must support those with sensory losses in ways that promote ontological security with:

- Improved communication, especially by healthcare professionals
- Better access to information
- Funding support services
- Removal of disabling barriers
- Peer support services (identified as high priority by people with Usher)
- Specialised psychological support, and don't forget the families, carers, partners and siblings and children
- Healthy coping'menus'

These will promote security and safety and support people with Usher syndrome to better navigate grief.

Simon

I'm Simon, and I live with Usher syndrome – a combination of significant hearing and vision loss. For me, blindness has been the most challenging part.

My disability means:

- Around 98% vision loss
- About 60% hearing loss
- Use of a guide dog and support workers is common in my daily life
- I miss roughly 30% of the words in conversation and can't recognise people by their faces or voices
- I visually miss about 90% of what's in my field of vision

Alongside the physical impacts, I've faced mental health challenges, including anxiety, depression and the fallout from domestic violence and assault. My upbringing wasn't ideal – family violence, abandonment and other traumas have all left their mark.

Impacts of Usher syndrome

Everyone has their own life journey, with challenges, hazards and moments of survival. Genetics, environment, parenting, work and social life all shape us. Add in the gradual or sudden loss of sight and/or hearing, and the path becomes far more complicated.

There's no clear manual for this. Mentally, it's a challenge on every front:

- What will my family expect?
- What will my employer think?
- How will friends and colleagues react?

Blindness and deafness mean missing most of the non-verbal cues (60–70% of communication) and a chunk of the verbal content too. For me, about 30% of spoken dialogue goes missing, and almost all visual information is gone.

While I make good use of my other senses, I can't claim the superpowers of Daredevil! My life has included depression, anxiety, violence, poor decision-making, grief and loss, but also loving siblings, lifelong friends, a wonderful daughter, fulfilling work and the support of the NDIS (Australia's National Disability Insurance Scheme which provides support and resources to those living with severe, lifelong disability).

Living with moving goalposts

Usher syndrome means constantly adapting, like playing every position on a footy team where the goalposts keep shifting. One day you're the ruckman, the next you're full forward. It takes emotional strength to cope with losing more sight and hearing while still trying to keep life on track.

Even well-meaning support systems can drain your energy. The NDIS has opened many doors for me, but finding skilled, trustworthy support workers can be a minefield. Large corporate providers often disappoint, so I now self-manage and choose individuals who treat my time and needs with respect.

The ongoing grief/loss cycle

Living with Usher syndrome is like being stuck in the grief cycle – over and over again:

- Denial: 'My eyes are fine, I can still drive, no need to tell anyone.'
- Bargaining: 'If I just get better glasses…'
- Anger: 'I hate people grabbing my hand. Stop telling me it's fine!'

- Sadness/Depression: 'Why me? I'll never be a good partner.'
- Acceptance: 'I'll do a marathon with a guide. I'll paint. I'll let the dog guide me.'

Unlike losing a job or having a setback that eventually fades, Usher is a constant change. You're always readjusting.

Coping strategies

Over time, I've learned practical ways to stay grounded:

- Grounding: Touch trees, fences, letterboxes or even lavender bushes when walking to calm my mind before stressful meetings.
- Practice 'blind days': Navigate my house with the lights off to prepare for further vision loss.
- Humour: Laugh off the inevitable 'blind days' and 'deaf days' – the shirt worn backwards, mismatched shoes or talking to someone who's already left.
- Lower expectations: Give myself longer timelines to avoid frustration when plans change.

Technology and support

The NDIS has been a game-changer for funding technology:

- Siri and voice assistants
- GPS apps
- Screen colour inversion for contrast issues
- Speech to text software like Dragon
- Password managers for online security

Support organisations like Vision Australia, Guide Dogs Australia and Achilles Australia have all been part of my journey.

Family and relationships

Raising my daughter was one of my proudest achievements. She learned resilience early, and when her school invited me to explain guide dogs, two whole grades came to meet my dog. It removed the 'disability' label and replaced it with connection.

My guide dog brought safety, independence and emotional support to our lives. When he retired, over 30 people came to his party – proof of how much he meant to our community.

Final thoughts

Living with Usher syndrome isn't easy. It means constant change, ongoing grief and the need to adapt over and over again. But it's also meant building resilience, finding good people, using the right tools and holding on to the belief that life can still be fulfilling.

If you're facing similar challenges, my best advice is simple:

- Advocate for yourself
- Accept help without relying on it completely
- Find the humour in the hard moments
- Keep your support circle close

The goal isn't to pretend everything's fine; it's to keep moving forward, even if the goalposts keep moving too quickly.

Conclusion

When services and institutions implement these strategies, they promote truly inclusive care. Health and well-being then become a collective responsibility, shared by health institutions, clinicians, support staff, care agencies and individuals themselves.

Usher syndrome can touch all domains of life: communication, education, employment, social relationships and of course, health. From daily tasks like reading medication labels to more complex processes such as navigating emergency services or hospital admissions, people with Usher syndrome can face unique hurdles that undermine safety, dignity and well-being.

Grief journeys deserve special consideration as adjusting to sensory loss and degeneration may take a long time. So wider social attention to removing disabling barriers, providing support patience, support, especially peer support, may be needed.

Yet, these hurdles are not insurmountable. With concerted effort at individual, community and institutional levels, we can transform the landscape of care.

Following on from these pillars of healthcare improvements, in Chapter 14 we turn to look at broader solutions for a better everyday life.

14
Supports for better everyday living

Introduction

In this chapter, we look at support for people living with Usher syndrome and their families and how this works in the real world.

First, we need to understand the protections and policies that exist to fund services and supports. For example, in England, this is the Care Act 2014 and in Australia, the National Disability Insurance Scheme Act 2013 (NDIS Act).

In England, people with disability, including Usher syndrome, receive support through their social services under The Care Act 2014. This works by approving funds so that people can book their own communicator guide to take them shopping, to leisure centres, meeting up with family and friends, all depending on the individual's wishes and desires, maintaining their independence. Funding and some services are administered by local councils and counties, so there is a lack of consistency across England (as well as between England and Scotland, Wales or Northern Ireland). Some people with Usher have difficulty accessing supports because some local authorities expect them to pay for services. This puts people with Usher at a disadvantage compared

to other people who do not need support, leaving a financial inequality for people with Usher who are paying out more for support and equipment that their peers without Usher do not need to spend on. This also creates inequity throughout various counties in England where some are better served than others.

People with Usher syndrome may be in receipt of disability benefits, but this is usually insufficient to cover all the expenses related to their disability. Under the NHS, costs are covered for interpreters and communicator guides. Emma however has noticed that the specialised and specific interpreter needs of people with a combination of hearing and sight loss are not always covered. In addition, other institutions such as opticians and dentist practices operate as private companies and refuse to provide interpreters, often failing to meet their duty of care under the Equality Act in the UK.

In Australia, the NDIS Act provides the funding and framework for the provision of support to people with severe, lifelong disability. This support is person-centred and goals-based, and resulting plans are funded and reviewed yearly or every few years. Participants have choices and control over the services and personnel engaged under their plans.

Emma uses her lived experience as a person with Usher syndrome and her lived expertise as a head of Usher services, researcher and a peer support leader to discuss common support requirements in a real-life scenario. People living with Usher (and indeed, any disability) are so often subject to oppressive forces such as stigma, negative attitudes and exclusion. By improving knowledge and skills (such as reading this book) and being more aware

of the support needs of people with Usher and their families too, you can help us all enjoy good lives and better health and well-being. Emma talks about:

- Support in everyday life
- Support at work
- Support for getting around
- Supporting reading
- Accessing support

Support in everyday life

As a working mother with older teenagers, I need support. I do not however want to be dependent on my children, because I want them to grow, develop and enjoy life rather than being young carers.

As my vision changes, my communication becomes more difficult. Mealtimes, for example, with family can be frustrating as I often miss out on conversations. I then have to ask others to repeat what was said. My husband converses quickly and deeply with the children, but often forgets to include me, especially if having heated conversations, which is not unusual with teenagers!

When I go to the supermarket I can shop, but it takes me ages to find the items I am looking for. I once took 45 minutes to find four items. Supermarkets and shops often change their layout. Shopping was especially difficult during the COVID-19 pandemic as I could not get a communicator guide to support me. This meant shopping by myself and taking a lot longer. With a communicator guide by my side, however, I can go shopping

more easily and faster too. The communicator guides explain the products and prices to me. Shopping is still time consuming, especially when buying birthday or Christmas presents. I can look at things online, but due to accessibility issues, I am not able to order them and must ask my husband or communicator guide to help me with actual purchasing. Ultimately, my preference is going out shopping and seeing people face to face, which I enjoy much more. Human interaction is so important.

When I prepare food, I cannot read the labels or the weight on the scales. I can't clearly see my induction hob controls; as the colour on the hob has faded over the years, I can't see the saucepan lines or switch setting. This means that I must ask someone to support me when cooking.

I used to love sewing, but can no longer see the needle, so I need assistance to sew, for example, my kids' scouts explorers' badges.

When I drop my pens or tablets on the floor, I cannot find them, so I must ask someone to help. This often takes them a few seconds, whereas I could be looking for over 15 minutes.

Having poor balance means I must think about which route to travel and whether I want to go out. It could be that I use my communicator guide to support the outing. If I feel anxious, tired or stressed, my balance is often worse, and I would then need my communicator guide to support me in my environment. Sometimes, I must rely on a family member. The paving slabs outside a friend's home are quite a challenge to navigate. These steps have no rails to hold on to and no contrast in colour. I have to ask my husband to guide me down the steps for my own safety. When I take my dog for a walk, I can lose my balance

but feel the pull of the lead which keeps me centred and stops me from falling.

In the past, I had a guide dog. The dog guided me around different environments and I felt independent. Since she retired, I am not eligible for another because I work remotely. This means less getting around and less independence. In the future, I would consider getting a guide dog again, depending on where I live and my work/retirement plans.

When I was single, I had a dual guide dog – one dog with two roles: a guide dog for the blind and a hearing dog for the Deaf. I found it brilliant to keep me company and prevent me from becoming isolated. It changed my life by getting me out to meet people back when I lived in London, which is a busy place to navigate.

When out and about now, I use a long mobility cane with a rolling ball on the end. It is coloured with red and white stripes which means that I am deaf and blind, although often people are unaware of the significance of the red. I find the long cane beneficial because of my poor balance and also to help me manage paving and roads. Sometimes, my communicator guide is not available as I get only 12 hours of support per week, and so my cane is indispensable.

I found food shopping with a guide dog particularly difficult, navigating the shop while holding my dog and purchasing food, with the added problem of how to hold the shopping bags and my guide dog. All this is more challenging with my poor balance. I found I had to adapt to a backpack to carry shopping home.

Since the new rule about charging for bags in shops, the assistants no longer put the items into bags, which means more work for me. As a person with Usher, I am already carrying my handbag, finding my purse and holding the mobility cane. When I make a contactless payment, the machine may decline, and I must enter my pin. This proves to be tricky, as I am not able to see the numbers. The screen appears blank to me, so I have had to ask my communicator guide at times to help, putting my trust in another when in a vulnerable position.

Having a guide dog, using a mobility cane and having the support of a communicator guide are very different and I have used them at different stages in my life. For example, when my children were very young, I found having the guide dog and using a twin pushchair quite a challenge.

My communicator guide helps to guide me and to communicate with the shop assistants. Since my vision has deteriorated and my lip-reading skills have vanished, communication with others is more complicated.

Having a communicator guide to support me when my children were small helped immensely. They were able to accompany me to take my children to various activities and for me to join mother and baby groups. Without them, it would have been much more challenging to access information, communicate and get around my environment. In short, without the support from social services under the Care Act 2014, life would be much more difficult.

When my children were only 2 and 3 years old, I had cancer and was very ill for over a year. It was a very difficult time, and I needed more support than usual. I managed to get a lot of support from

my communicator guides, my family and especially my husband as well as my mother who drove over 2 hours to help almost every day.

Work life

At work, I have Access to Work support (this is government-funded support to help people with a disability get or keep a job in the UK). This pays for my interpreters and communicator guide. This support is principally to translate information and guide me when needed.

I have worked in the field of deafblindness for over 20 years and can do my work because I have been provided with interpreters. We have flexible work tasks and protocols where my interpreters wear plain, dark tops (to receive sign more easily with a strong contrasting background) and they agree to communicate-guide me within my environment when out interpreting face to face.

This flexibility is needed as my Access to Work support doesn't cover all costs for all of the different kinds of support I require at any one time. For example, at a conference, the interpreters would translate all the information from the seminar from English to British Sign Language, and during breaks would guide me to the tea/coffee area because the environment may be too dark, unfamiliar or difficult to see. For this reason, I may also struggle to make myself a drink and they often support me. Networking is also a tricky scenario to navigate, as if I am holding a cup or a plate I cannot sign to communicate. If the area is too dark or too bright near windows, for example, then we would have to move to another area. During lunch breaks, there is also the difficulty of eating and conversing with others, especially as my interpreters

need to have a break and eat too. They would help me to get food and find a suitable table. Without them, I am also unable to access any conversations around me. This means my interpreters have extra tasks working with a deafblind individual. Many interpreters avoid working with deafblind professionals because they have extra needs. Some interpreters prefer to be there solely for translating the information and not taking a more supporting role.

A few years ago, my reading vision changed, and I struggled to access my computer to work. I requested a Deafblind Assessment in the workplace. They deliberately delayed the process of getting this assessment. I mentioned that my neck pain and headaches were getting worse because of leaning into the screen and not having the right equipment. They did not arrange for an occupational assessment. After over a year of struggling, I then moved to a different organisation, and they finally provided the assessments I required. Following the assessment, I received the appropriate equipment to support my needs, such as a computer stand, a desk that can be raised and lowered as needed, a large screen I can move and am able to see my interpreters on and Braille equipment as well as the right type of blinds to reduce glare and ensure I can see the screens when working. My work situation has greatly improved, and I have reduced headaches and migraines following this. These positive experiences have enhanced my life at home, with greater work satisfaction. As my vision may deteriorate further, I may not be able to continue the work I do now, needing to work face to face with my interpreters at all times and adapting my communication needs, possibly using hands-on sign.

I have had several negative experiences with my support at work. An interpreter who I worked with for many years started to change their attitude towards me, wearing inappropriate clothing that made it difficult for me to see their signing and ignoring messages. This interpreter caused me to be an hour late for a meeting on one occasion by failing to meet and guide me from the train station as was normally agreed beforehand without issue.

I found it very hard to deal with the attitudes of the interpreters who are no longer supportive and are disinterested in working with a deafblind person, particularly as my vision has deteriorated. These changes now mean I am unable to read written information, which becomes challenging and frightening. At that time, I felt very vulnerable. I believe safeguarding needs to be in place with reviews for deafblind professionals and their communication support, such as interpreters and communicator guides, to advance deafblind people's safety. Additionally, I did not feel that I had anyone to talk to about this circumstance, because the interpreters work freelance and are registered with the National Register for Professionals working with Deaf and Deafblind people (NRCPD). So, there is more need to provide support that meets the needs of deafblind people over all stages of life and work.

Getting around

When I was young, I was very independent and travelled solo; however, as my vision has changed, people tend to help me more or ask if I need help.

Recently, I went to a family party, and we needed to go to the kitchen to serve our food. A member of family asked me if I wanted to have my pudding served and brought to my seat. I replied with thanks that I would get it myself. However, I do appreciate the offer, as I am determined to keep my independence as much as possible without relying on others. I wanted to do this for myself. Plus, I like to see for myself what food is being served! Things like this can be taken for granted by those who are able to easily do so.

On the London Underground, I recently travelled on the tube without the assistance of staff; however, a member of the public asked if I would like a guide. I appreciated but declined the offer. At night-time, however, it is quite a different matter, and I would have accepted the offer, being too dark to see. Depending on the circumstances, it is important for people to ask me if I would like support, instead of assuming that I must have the support. On occasions, I have refused support and then realised that I need it. On a recent journey, I found myself arriving at the wrong station as I was unable to read the platform numbers on the screen and mistakenly took the wrong train.

Reading vision and Braille

When I first moved to Sussex, I contacted my local authority for the provision of Braille tuition. I had previously begun learning Braille when I lived in London, where it took around 2 months to have this in place. Unfortunately, my learning stopped prematurely when I became ill, and I wasn't able to continue. Here, in Sussex, I waited over 6 years before I had something set up. This time frame is extraordinary.

I started one-on-one rehabilitation support for Grade 1 Braille on a weekly basis, and a British sign language interpreter was provided to assist. It was positive and a lot to learn and adjust to. After some months, however, the local authority stopped the sessions, citing the high cost of the interpreter. I felt emotionally affected as well as demoralised. I stopped learning Braille for a few months, which negatively impacted on my learning and enthusiasm. As a working mother, I juggled everything, plus my studies on Braille, so it was extremely difficult. Eventually, the local authority agreed to continue the support. But then, the tutor retired and the sessions stopped once again. I was unable to find a private tutor. A social worker did give me details for an organisation I can contact but have heard no further. I must continue to learn on my own. Currently, this means I am doing remote learning, but without the support of a tutor I cannot clarify things that I don't understand. I have now completed Braille Grade 2 online. I have real concerns about the future provisions of rehabilitation for deafblind people, finding that many continue to get little to no support.

On a positive note, now that I have Braille equipment, I find that my Braille reading skills have improved. When I look back to last year, I was understanding only letters of code, slowly progressing to words. Now, I am managing to read whole sentences in Braille, picking up the sentence'strolled back to the hotel' in Braille Grade 2. I was elated at seeing this progress; it really feels like light at the end of the tunnel. It is a positive step, and I cannot wait to be able to access more information with my Braille device.

Another blow to losing my reading vision was watching television. Being unable to read subtitles anymore means I cannot

watch television, films and have a laugh as I once did. One day, using voice to text, I may be able to turn audio into Braille to feel the information from subtitles. The future will tell. A few months ago, I also found that I struggled to see people's expressions, and I find myself leaning on my husband for more support, asking him what has happened to the actors. He explains briefly to me to fill in some of the gaps, but I must be mindful that he also needs a rest in the evenings after finishing work.

Not being able to discern written letters anymore means I need someone to translate what has been said, with support from family, my communicator guide or the interpreters I work with. With my phone, I can access the device but need support by adjusting the accessibility settings. WhatsApp is a very popular platform to use for messaging, but I am now unable to read them, although I can use the photo and video function. This means to read a typed message on WhatsApp I must copy and paste each message on to a blank email and adjust the font to Arial, bold, size 24, which seems ridiculous. For my friends who are British sign language users like me, I ask them to send me their signed videos instead. This is great and much more accessible for me, although I can struggle if the background makes it difficult to see the lighting and clothes people are wearing. If these are busy/light in colour, I switch to my iPad to watch the videos on a bigger screen, or if all else fails, ask my husband or someone else to translate the information.

In my work supporting deafblind clients, I see that many of them can't access Braille because it was never encouraged earlier on in their lives, especially for people with Usher. For some people, communication is very specific, and all information must be

received this way, for example, hands-on sign. For those whose only communication is speech and using residual hearing, isolation is very real if their hearing declines, and they are left without the tools to access information in another format.

Accessing support

I require support at work to do my job effectively. This can mean that when support is cancelled or changes last minute, my work is adversely affected. I recently had an interpreter that cancelled last minute when I had work meetings to attend. Being unable to find a second interpreter at such short notice or an interpreter willing to solo work with me, I was forced to cancel these meetings.

Another support I had in place while my children were young was extra learning provision for my two children to help them with their communication, approved and provided by my previous local authority because my husband and I are both deaf. We felt it was important for our kids to not fall behind with their speech and learning development.

A memorable experience I have when out shopping when my young son was with me was a time when my support was off sick. While looking at the clothes, I turned and noticed that my son had vanished. I panicked and started to look for him, asking the assistant for help. The assistant was able to contact the other shop staff, who discovered him on the first floor, up from me, because he was fascinated by the escalators! I found him sitting by the escalators, still happy and talking to the staff there. I was very relieved when I saw him.

Having a communicator guide is an invaluable support mechanism. It means that I can meet my friends in different places which may not have good transport links such as a national trust site. It makes a huge difference to my life, being able to travel with confidence and without stress.

As I get older, my support needs change along with my changes in vision. I struggle to lipread my mother and brother because we do not see each other as often and my vision has deteriorated. Their faces have changed too. In the future, I may need to have a communicator guide to support me, or perhaps a speech to text to Braille device. I feel lucky to have my sister who also has Usher. This means she has some understanding and can communicate with me as well as having empathy with each other when we face discrimination, negative attitudes or need emotional support because of our Usher. I also feel lucky to have supportive friends I can reach out to.

Support at medical appointments

If I have a hospital appointment I would need to book a communicator guide, as well as informing the hospital to book a specific interpreter; although with a specialist deafblind interpreter, they can often guide also. This is organised by the hospital, through the interpreting agency that they have contracted to provide such services. Sometimes, they make things difficult by booking a different interpreter who does not meet my specific needs as a deafblind patient. This is time consuming as well as costly for the hospital and for me to travel to and from the hospital with a wasted trip.

On some instances, I would need to travel by car and would need to find a communicator guide to drive. If they cancel or are

otherwise unavailable at short notice, I have no way to make my appointment, and it is difficult for me to contact the hospital to inform them. I have previously faced poor attitudes of hospital staff when I needed to cancel an appointment because my communicator guide was unavailable and I could not travel to the hospital alone. The receptionist I spoke to was inconsiderate and complained that they had booked an interpreter for the appointment, acting as though I was to blame. If I had been without anyone to support me in making the phone call, I would also have been unable to inform them that I wasn't able to make it.

Human support is critical

Having communicator guide support is such a positive thing. This is especially the case when I feel isolated or am going through challenging stages in my life and experiencing difficult emotions as I can talk to my communicator guide openly. This is especially important as my close friends do not live nearby and my family live far away from me.

I am not keen to give up work as I think it will increase the chances of becoming isolated and depressed. I like being able to work; working with people and interacting is vital. I feel that since the COVID-19 pandemic, people tend to forget what it is like to be lonely and to feel isolated. There was so much less support and people with Usher were hugely affected during this time, being more vulnerable.

In the last 2 years, my reading vision has changed, so I tend to rely more on other people for information as I have not fully achieved my Braille Grade 2 to fluency yet. This means that my needs at

work and home have changed and I now have interpreters to work with me face to face more regularly as a result.

Conclusion

Emma's lived (and living) experiences demonstrate the complexity of living with Usher syndrome: work-life success when supports are easy to obtain and flexible enough to adjust to ongoing progression of impairments. These mini narratives also detail the delays, inadequacies and denials of some supports, and there are negative impacts of these on coping and adjustment.

This book has been grounded in the lives and professions of its authors: Annmaree, Emily and Emma. But you have heard from others too throughout. We have chosen to bookend this journey through the complexities and challenges, downsides and upsides by demonstrating how diverse and capable we are. The final chapter gives 'snapshots' of Usher lives of different ages, depicting Usher syndrome as a true spectrum.

15
Snapshots of diversity and capabilities in the Usher community

Introduction

We have seen in earlier chapters the idea of a spectrum of residuals, resources and communication modes that impact life with Usher. Combine this with the degeneration of one or more senses, and we have not only complexity but also vast diversity in the population living with Usher syndrome. We want to end this lived experience-led and informed book, *About Us, by Us*, with vignettes from the communities. We bring now some mini portraits of people and families living with Usher syndrome to illustrate these points and showcase that with allies, inclusive attitudes, enabling employers, health and social resources and creative, adaptive technologies, we can harness capabilities and flourish.

Thank you to:

Bea
Gail

Jacqueline
Jane
Natalie
Peter
Stephen
Traynor
Wes

Bea

I'm Bea, I live in Australia and I'm 19 with USH2A. I was diagnosed at 6 weeks old with a moderate to severe hearing loss and fitted with my first hearing aids at 8 weeks. I had early intervention up until I started Kindergarten at 5, developing speech, language and learning how to read. I functioned at a high level during my school years, and to that I owe it to my parents and the support I received from Hearing Support teachers at school.

Around 14, my second cousin was diagnosed with USH2A. My mother knew we had the same hearing loss, and I was noticing changes in my vision. She took me to the optometrist, who diagnosed me with astigmatism. Mum brought up my cousin's diagnosis, and he took photos of my retinas and noticed mild pigmentation that normally he would rule out as insignificant, but given the medical history, he urged us to continue seeking out a definite answer.

The next year was filled with genetic testing as well as other eye tests and exams at the children's hospital. I was diagnosed with rod-cone dystrophy, and then at the end of the year, when I was 15, the genetic testing came in confirming Usher syndrome.

Year 11 and 12 at school were challenging, as my vision continued to change. I started to develop photophobia, and my night vision degenerated very quickly. Smaller print began to be very difficult to read, and I would often compare with my friends the reduction of my peripheral vision. It was a lot for a teenager to take on, but again, I had incredible support from my friends and family and from the hearing and vision teams at school. I learnt grade 3 Braille and finally got a cane just before I turned 18, which I now use in crowded and dim environments. I completed school with good grades, though that was a struggle due to the constant visual and hearing fatigue. I would often come home from school and completely crash out, unable to do homework or socialise much with my family because I was too tired. I would also become quite exhausted during my later classes, and there were times when I came close to falling asleep. But I passed and made it to university.

My parents have always encouraged me to explore different skills and experiences, so I've become quite proficient at music: piano lessons and I still play; percussion in community bands; singing lessons; vocal ensemble at university; and play the piano for church.

I've now moved out and am living on campus for university studying History, which was a huge dream for me. I'm also very excited to be a part of UsherKids Australia, which is an incredible support network for people with Usher.

Traynor

So I have Usher syndrome and I'm at uni now. I don't remember exactly when I first knew what this actually was but I had hearing

aids and everyone knew the blindness was gonna come get me. But it doesn't happen like you think it will.

I love sport, especially soccer, and I didn't think the not being very good at it was related to the Usher stuff until I got hit on the head by a ball I never saw coming. When I started talking rubbish, I got taken to hospital with concussion. The doctor in the emergency department said, 'Well, when you have very small visual fields, what does anyone expect?' Honest, but not very sympathetic. Zero for bedside manner.

So, after that I noticed more about seeing less, especially at night and also in bright light at the beach. School was good, I had to work hard to pick up all the information I didn't hear, but if you read the textbook and screenshot important pages you can build good notes.

I have good mates who don't mind driving me everywhere we are going and picking me up even when we are not doing anything together. Uber app is my good friend. I really wanted to learn to drive but my mother said NO. And meant it. She said you never ever want to be responsible for hurting anyone. She is right, but it doesn't mean I liked it.

I'm studying to be an engineer because I really like learning how things work. The 3D printer is good as I can print out models so I can feel all the working parts, or some of them anyway. Some people might say there's no future for a blind engineer, but I think I should do what I am interested in no matter what.

I use lots of different tech in my studies, and I have a math tutor as I find it hard to understand just from written descriptions and I'm training myself to 'see' the numbers, shapes, equations and so

on in my head. So, life is this seesaw between just enjoying now while keeping half an eye on the future.

Jane

I am 30 years old and have Usher 2, but I haven't had genetic testing; there didn't seem to be a point in spending money to prove what we already know. I use restricted frame sign language (Auslan), but my parents tried really hard for a while to mainstream me. School was awful as I had no idea and got bullied, with kids chanting 'deafie, dumbie'. I have lots of signing friends who had the same issues. My parents realised I needed sign language to learn properly. I have a great group of friends, and it's a good relief to sign with them without having to struggle to lip-read and work out what is being said. Two of my friends who have Usher were brought up oral-only, but they are now in their 20s and don't speak, but sign instead. They say it's much easier. If I could tell you one thing that needs to be better, it would be doctors and hospitals. Communication needs to improve so much. I have had such bad experiences that I won't go now unless I really, really have to. But I have a craft business of my own, and I love seeing my friends, so life is good. Really.

Natalie

Usher diagnosis is complicated. We had the early knowledge of both our girls having hearing loss at 3 months of age; however, their genetic testing (2000s) did not include Usher. And we became comfortable with excellent progress in their speech and language, schooling and connections.

However, it was a major blow when our eldest was confirmed with retinitis pigmentosa at 14 years. My husband and I had sleepless nights for 5–6 months and then again once we had the genetics confirmed. I felt awful that I had not acted sooner about her comments that she had a little difficulty seeing and reading – in reality, the only difference it would have made was medication for her macular swelling. In hindsight, she was often tripping over the white baskets of laundry (on our white kitchen tiles), which we put down to having size 12 feet!

It's been really hard to understand what their vision is like. In connecting with other Usher kids and their families, we are all finding the words to share by learning from each other's experiences.

My personal experience has led me to work and advocate in the disability space. I think that the strength for our family is having knowledge of the modern approach to disability, their rights and the amazing supports which are available in their life right now, like the NDIS and technology.

We have learnt to accept that it is more than their hearing and vision – that energy levels and mental health are impacted. But this has created a new openness and different approach to life and school by considering what are our priorities are – well-being being the most important. We are on a different path and as their parents, we must be the strong support in the background.

Peter

I'm 43 and have Usher type 2. My two sisters also have this. We are a very mixed bag. I'm the youngest, and by the time I came along, the older two had been diagnosed. This meant my parents

found out early I was deafer than my sisters and started me learning sign language. But no one else learned it, so it was stressful at home. But sign language was good at school where I got sent to boarding school for deaf kids.

I'm not very good at English and I'm part signing, part dictating this. And I've never been able to get a job. 'You need to have the experience', they say. How can I get experience if no one gives me a job? When I went to Italy, I saw a bar run by deaf people and I wish we had something like that in England.

My sisters have more hearing than I do, so they seem to manage a bit better. One of them is in customer service on the telephone, sorting out people's online order forms that they have messed up. I keep going to the disability work office and they keep telling me to do more courses to get more skills to get a job. I have done five of these at the local college and still no job.

Gardening is how I keep busy. I like to touch the plants and I look after my parents' garden and grow vegetables. It's really important to know what is safe and what is not, and my father ripped out all the poisonous plants, just in case. There is a nice group near where I live and run by the sensory services, and I go to that when I have someone to help get me there.

My family is a bit messy as we don't have a way of talking to each other, as my sisters only started learning sign language this year and my parents never really learned. I have one sister who is hearing and she takes me to doctors' appointments. She has some sign language as she learnt some when I was little. I can tell her things.

Jacqueline

I am from England and I was born Deaf. I was diagnosed with Usher's in my early 20s. My motto for life is 'let's encourage friendship.' My story here is being translated by British Sign Language interpreter Monica.

My school was oral and I learned to lip-read. Out of school, I learned British Sign Language. This language was recognised by the UK government in 2003.

I started to need good light to see sign language and then I could only see signing in a smaller space. I began to miss bits and learned 'hands-on' signing. It was the same with walking. I walked alone. Then, for balance, I held on to another person. Next, I learned to use a cane. This gave me confidence and independence.

I love riding. I had dressage lessons with an interpreter, but now I cannot see the interpreter. I can still enjoy riding with others on a 'hack'. I love swimming, too. But the same happened, and I could no longer see to copy the people beside me in aquarobics. I need touch.

It is all about touch. But I also use my memory of shapes, colours, how things work.

Television was fine because I could make the subtitles bigger and bigger. Then the colour went. It was only black and white and I could not read the words. I need hands-on, but my husband does not always want to watch the same programmes. Instead, I crochet.

I like to plan my garden and learn about flowers and trees. Now I need help, hands-on. But I can recognise scents such as roses or herbs such as sage and rosemary.

I lead a full life. When I meet new people or go to new places, I need a lot of patience. Often, I reassure other people about how to try to communicate with me.

Yes, sometimes I am stressed, angry or feel low. But I decide nothing has to stop. Find another way. Sign 'I can', not 'I cannot'. Enjoy life. This world is precious. Let us be hands-on and encourage friendship!

Stephen

Life with Usher's has been one of diminishing sensory inputs, requiring increased effort to achieve outcomes. Life begins with wonder. Everything is new and potentially exciting. Like most children, I try all sorts of crazy things. In 1961, I arrived at kindergarten only to find out I'm deaf. I do have some hearing, but in those days, they didn't split hairs over whether one was deaf or hearing-impaired. Hearing aids in those days were rather bulky and not at all cool. I was a naughty boy and didn't wear my hearing aids – a habit I maintained until my late 30s.

In 1972, I'm in the office of a retinal specialist. Another doctor conducting a physical examination on my brother had noticed anomalies in his retina. It became clear that, coupled with my known deafness, I had Usher's. The diagnosis was more relief than shock. I knew something was going on, and it had a name. A hard road to play, but knowledge is better than ignorance.

Understanding Usher's helped focus my efforts. I had to work harder, managing nonetheless to complete three university degrees, two when totally blind. My professional engineering career was in the electricity industry. Social and community

participation have been challenging, but self-knowledge helps. The past 30 years have brought increased involvement with the Usher's/Deafblind community. Learning sign language; learning Braille; learning to travel using a cane and dog guide; wearing those hearing aids I once refused to wear; enjoying community.

Sensory impairments compromise communication. Flexibility is essential when dealing with Usher's. In a professional capacity, always communicate directly with the individual. If they use sign language, you should use a suitably qualified professional interpreter. Verify you have been understood. If not understood, try a different approach, but always verify.

Gail

I know I have Usher 2A because I had genetic testing done about five years ago. It was part of the testing to see if I was suitable to take part in a trial to test the effectiveness of a drug in slowing retinitis pigmentosa. I didn't make it into the trial; my peripheral vision was too far gone.

In a nutshell, I have worn a hearing aid since I was about 6. Vision effects appeared in my 40s, but didn't start to have serious impacts until I was in my 60s. I have a PhD, have lived and worked in the US as a researcher and as an academic in Australia. I have co-authored two university textbooks (on climate change). I am currently president of a disability advocacy organisation. This isn't the typical Usher experience I know, but it's what I've had.

Wes

My wife has Usher syndrome (I am deafblind too, but my hearing loss is from the military – too many explosions – and my

vision loss is from vascular disease in both eyes) and I am her carer. We live in a retirement home. The facility is badly managed, and I am the one who makes sure my wife has everything she needs, helps find things and manages her tablets and appointments. We use walkie-talkies, which have a range of 3 kilometres, as they have big buttons and are easier for us to use than mobile phones. That way, she can call me if I am going to the shops and she realises we need more eggs or something.

We have both had different journeys with deafblindness. My wife has multiple medical conditions, and the blindness makes it hard for her to manage her medicines while the deafness makes it hard/impossible to understand the doctors and nurses. So, I have to be her eyes and ears, even though mine aren't very good.

It can be really hard sometimes to get doctors to tell you what is going on when she is in hospital, so I have learned to stand by the nurses' desk and wait for someone to come along who can explain things. I'm lucky I have good hearing with my Bluetooth hearing aids. I use a white cane, but I really love it when people say they didn't notice I am deafblind. Sometimes, they will tell me, 'But you don't look it', which is weird because I don't know that there is a special look or a funny walk that people with Usher or deafblindness have.

For my wife, it is different and more difficult. But we do fun things together – we like to have picnics (no blue cheese though, as we both hate the smell) and we like to dance and go for walks. I have a lot of friends from my working days that I keep in touch with, so

that keeps me going. We married later in life, and her kids didn't approve. Still don't. They are a problem.

They keep saying she needs to go into a nursing home because of her 'vulnerability' and all the 'risk' of having Usher syndrome and all the diabetes and heart problems. But I say she would be desperately unhappy and worse off in a home. We are happy the way we are right now. Together. All we really need to make things even better are some grandbabies!

Conclusion

A diagnosis of Usher syndrome, whether as an adult or infant or in between, is a complex and challenging time for individuals and families. And as vision and sometimes hearing loss progresses, there will be times of crisis and adjustment. With good support, understanding and a person-centred approach, we are capable and competent people who have much to contribute.

What we need health and social care students and professionals to remember is:

- Be informed (read this book)
- Be good communicators
- Be person-centred and ASK individuals what they need and want
- Be supportive (but not overprotective)
- Be aware that families, siblings, partners and parents need care and support too
- Be up to date with assistive technology and apps and devices; this is a constantly evolving and changing area
- Be accessible; provide information in multiple formats

- Be attuned to the individual's grief journey; it may be long and complicated
- Be involved in research if you can, as the more knowledge we build about Usher syndrome, the better the evidence for better policies and practices will be.

Suggested learning activities

1. Get into groups of four or five, discuss and answer the following:
   (a) What happens in your country around diagnosis? Is there a newborn hearing screening in place? What are the pathways for babies identified with hearing loss?
   (b) Think about the medical, community and Usher perspective on diagnosis. Think about the differences between being diagnosed as a baby or child and as a young person or adult.
2. Using the chapter on health and well-being risks, brainstorm in small groups how you can minimise or eliminate some of these risks.
3. Design a webpage that informs people with low vision of the risk of benign visual hallucinations, also called Charles Bonnet syndrome.
4. Learn the deafblind manual alphabet, also called the tactile sign alphabet, for your region. Play games that help to reinforce this skill.

   The following links contain some simple signs, a Tactile Fingerspelling alphabet, a Print on Palm alphabet and a simple Braille table. This link below is from Deafblind Information Australia, and has a pdf available for download, one for the tactile alphabet and one with instructions for Print on Palm/Block alphabet.

https://www.deafblindinformation.org.au/living-with-deafblindness/deafblind-communication/deafblind-manual-alphabet/

https://www.deafblindinformation.org.au/wp-content/uploads/2025/06/DBIA-Deafblind-Alphabet-2024.pdf

https://www.deafblindinformation.org.au/wp-content/uploads/2023/05/DBIA-Block-alphabet-2023.pdf

Find someone who would also like to learn, and take turns spelling to one another, receiving with eyes closed. You can choose words from your surroundings, such as table, tree, floor, exit, and more, or choose a set of 16 letters and make up as many words as possible from only those letters.

Chinese whispers: Person one makes up a sentence and fingerspell it to person two, who must then fingerspell to person three and so on. The last person receiving the message has to say what they think it is.

Grandma's shopping list: Grandma goes shopping and she buys…. Person one fingerspells the first letter of the name of the item and person two has to fingerspell what they think the rest of the word is.

5. Make flashcards for common medical situations, including information in tactile and visual communication, for example, Braille, photos, drawings and smartphone flashcards that include auditory components.
6. Get an orientation and mobility specialist to discuss and demonstrate sighted guiding and environmental description.
7. Write an essay on how care and communication have impacted people living with disabilities.

8. Invite a person with Usher syndrome to come and share their lived experiences with the class; remember to ask what accessibility support they will need to do this.
9. Communication and accessibility technology is expanding all the time: choose a new device or app, explain how it works, give examples of how it can aid care and communication and give examples of its use in hospitals or medical care.
10. Have a mini tech expo in class: each person takes on the research of one app, device and so on, and then presents this to the class. This will showcase the great diversity out there.
11. Research the disabled peoples' organisations in your region and investigate what they can offer a person with Usher syndrome.
12. Write an essay on the legal protections and policies in your region that support better inclusion for people with disability generally and Usher syndrome.
13. Prepare a personal emergency plan for an older adult with Usher syndrome who is a tactile sign language user: how will you let them know there is an emergency? Who will come to see if they need help? What do they need to survive a few days without power?
14. The lecturer gathers a number of photos, graphics and graphs. The class has to write out descriptions of what they are seeing for the access of people with low vision.
15. In a safe space, practise negotiating the space with low or no vision with someone as safety observer. Borrow a cane or even a broomstick and see how using that can make moving around easier, navigating a place you know well versus a space you've never been to before.

16. Try doing simple tasks, such as folding clothes, playing checkers, buttering a slice of bread and eating a meal with a blindfold on but no speech.
17. Contact deafblind and Usher syndrome support organisations and volunteer.

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Resources

Australian Resources:

Organisations:

Deafblind Information Australia
https://www.deafblindinformation.org.au/
Deafblind Australia
https://www.deafblind.org.au/
Deafblind WA
https://dbwa.org.au/
Deafblind Victoria
https://deafblindvictoria.com/
UsherKids Australia
https://usherkidsaustralia.com/
Able Australia
https://ableaustralia.org.au/
NextSense
https://www.nextsense.org.au/
National Disability Service
https://nds.org.au/
Vision Australia
https://visionaustralia.org/
Guide Dogs Australia
https://guidedogs.com.au/

Achilles Australia

https://www.achillesaustralia.org.au/

Charles Bonnet Syndrome Foundation

https://www.charlesbonnetsyndrome.org/

Certifications:

Dual Sensory Impairment for Health Professionals Module: http://dsiproject.org/course/dual-sensory-impairment-for-health-professionals/

The Certificate in Professional Studies (Deafblind Studies) course: https://www.bcu.ac.uk/courses/professional-studies-deafblind-studies-cert-dip-2025-26

Recognised Practising Deafblind Interpreter, Monash: https://www.monash.edu/news/monash-in-australia-first-accredited-training-for-deafblind-interpreters

Other:

Collaborating 4 Inclusion Person-Centred Emergency Preparedness

https://collaborating4inclusion.org/pcep/

Deafblind Block Alphabet/Print-on-Palm

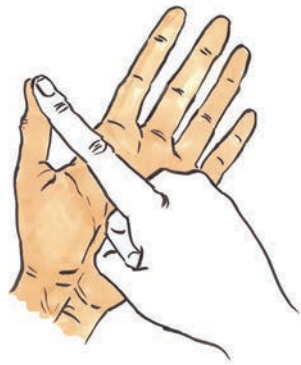
https://www.deafblindinformation.org.au/living-with-deafblindness/deafblind-communication/block-alphabet/

Deafblind Manual Alphabet

https://www.deafblindinformation.org.au/living-with-deafblindness/deafblind-communication/deafblind-manual-alphabet/

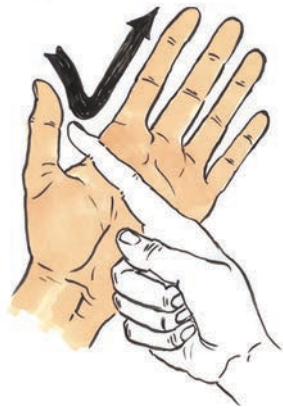
Deafblind manual alphabet (Australian)

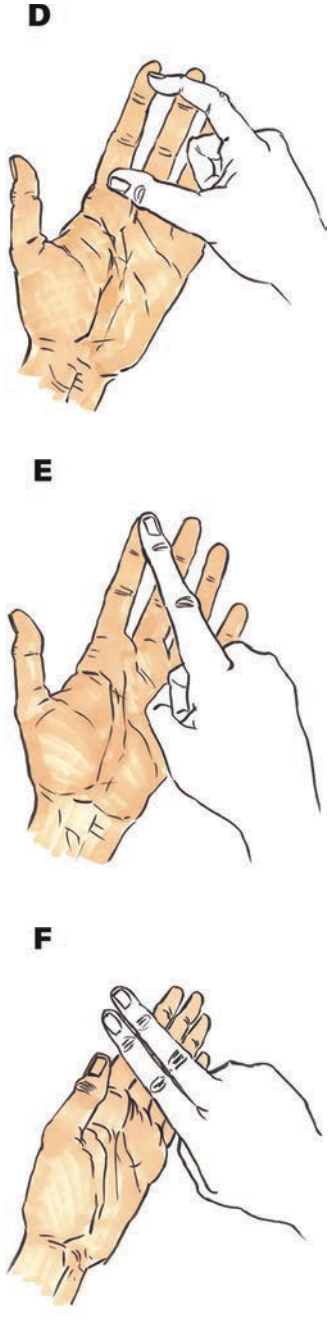
A



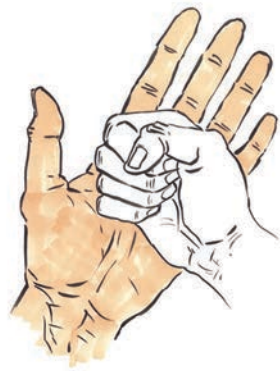
B

C



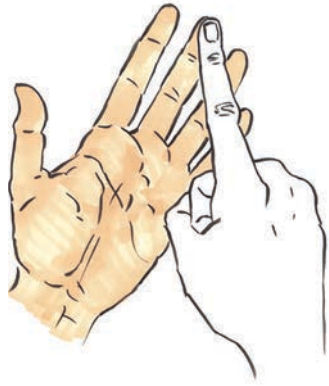

D
E
F

G



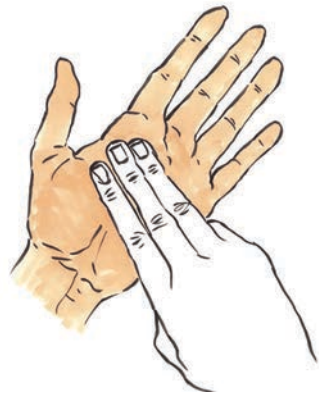
H

I

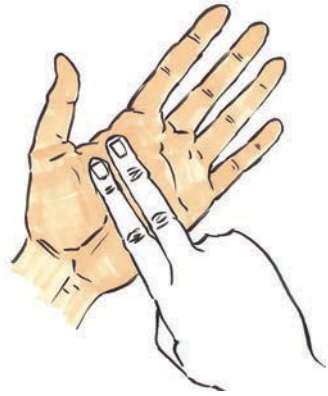



J
K
L

M



N



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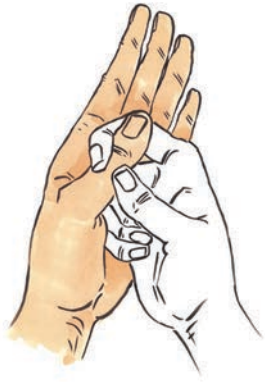

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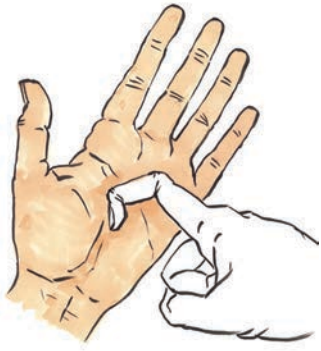

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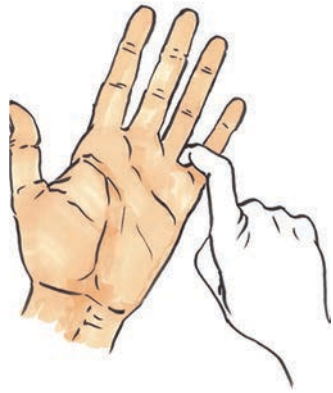

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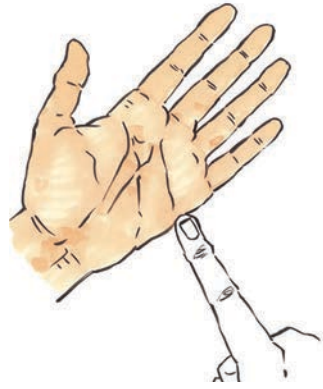

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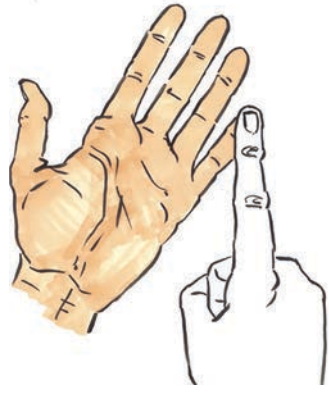

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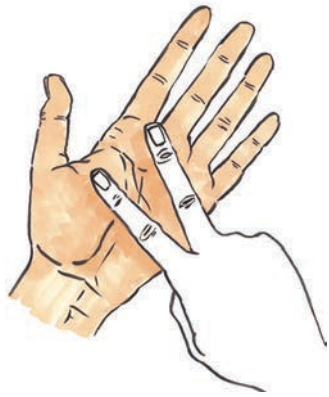

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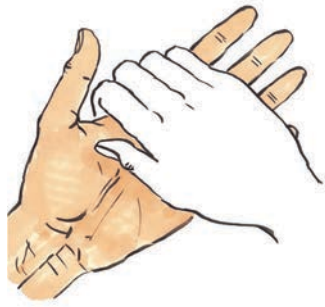

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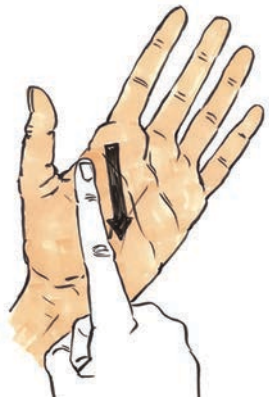

Z

United Kingdom Resources:

Organisations:

Bright Deafblind BSL
https://brightdeafblind.com/

Deafblind UK

https://deafblind.org.uk/

Deafblind Scotland

https://www.dbscotland.org.uk/

Deafblind Enablement

https://deafblind-enablement.co.uk/

Usher Kids UK

https://www.usherkidsuk.org/

Cure Usher Syndrome

https://cureushersyndrome.com/

SignHealth

https://signhealth.org.uk/

Sense Scotland

https://www.sensescotland.org.uk/

NRCPD - National Registers of Communication Professionals working with Deaf and Deafblind People

https://www.nrcpd.org.uk/

Signature

https://www.signature.org.uk/

Esme's Umbrella - Charles Bonnet syndrome UK

www.charlesbonnetsyndrome.uk

Macular Society

www.macularsociety.org

Legislations:

The Equality Act 2010

https://www.legislation.gov.uk/ukpga/2010/15/contents

The Care Act 2014

https://www.legislation.gov.uk/ukpga/2014/23/contents

Department for Work and Pensions (DWP)

https://www.gov.uk/government/organisations/department-for-work-pensions

Access to Work (ATW)

https://www.gov.uk/access-to-work

Certifications:

Birmingham City University (Professional Studies (Deafblind Studies) - CPS / DPS)

https://www.bcu.ac.uk/courses/professional-studies-deafblind-studies-cert-dip-2025-26

Other:

Usher Syndrome Information Handout (UsherKids Australia) – This fact sheet from UsherKids Australia provides an overview of what Usher syndrome is and available supports.

https://usherkidsaustralia.com/wp-content/uploads/2024/07/Usher-Syndrome-Infographic.pdf

Tough Talks: Talking to Children about Sight Loss – Royal National Institute for Blind People (UK) – General recommendations for parents on how to discuss vision loss with children

https://habilitationviuk.org.uk/wp-content/uploads/2016/08/APDF-ENG170108_Tough-talks-rnib-abt-sight-loss.pdf

UsherKids Australia Eye Clinic Video – This video shows the various tests to assess visual function, retinal imaging and specialised vision electrophysiology assessments for patients with Usher syndrome. It describes ways parents can prepare their children for undergoing eye tests and may also be useful to show older children so they know what to expect when attending the clinic.

https://vimeo.com/684026390?share=copy

Hearing Tests Educational Video (UsherKids Australia) – A helpful video for children and families about what to expect when coming in for a hearing test.

https://vimeo.com/684025949?fl=pl&fe=sh

Hearing Tests for Cochlear Implants Educational Video (UsherKids Australia) – A helpful video for families to explain what to expect regarding appointments following cochlear implantation

https://vimeo.com/684025393?fl=pl&fe=sh

International resources:

Deafblind International Resources
https://www.deafblindinternational.org/resources/

Accessibility Guidelines for Sensory Loss, Deafblind Ontario

https://accessibilitycanada.ca/wp-content/uploads/2020/09/Accessibility-Guide-For-Sensory-Loss-DeafBlind-3rd-Edition.pdf

Usher Coalition US

https://www.usher-syndrome.org/

Russ Palmer and Riitta Lahtinen's website

https://www.russpalmer.com/

139 Haptic Signals, Danish DeafBlind Association

https://www.dovblinde.dk/viden-om/kommunikation/haptiske-signaler/

Recommended further reading

Assi, L., Shakarchi, A. F., Sheehan, O. C., Deal, J. A., Swenor, B. K., & Reed, N. S. (2020). Assessment of sensory impairment and healthcare satisfaction among Medicare beneficiaries. *JAMA Network Open, 3*(11), e2025522. https://doi.org/10.1001/jamanetworkopen.2020.25522

Avery, S. (2018). *Culture is inclusion*. Sydney, NSW: First Peoples Disability Network Australia.

Bilby, L. (2012). *The Auslan dictionary: For teachers, parents and professionals*. Kanoona, Australia: Bilby Publishing.

Blackett, B., Cox, L., Johnson, J., Kek-Pamenter, J., Lattouf, A., Miller, P., Valencia-Forrester, F., & Worsley, R. (2021). *Disability reporting handbook* [PDF]. Media Diversity Australia. https://www.mediadiversityaustralia.org/wp-content/uploads/2021/11/MDA-Disability-Reporting-Handbook_FINAL-V5_21112021.pdf

Eggins, S., Slade, D., & Geddes, F. (2016). *Effective communication in clinical handover: From research to practice*. Berlin, Germany: Walter de Gruyter GmbH & Co. KG.

Lahtinen, R., & Palmer, R. (2012). Environmental description. In E. Perego (Ed.), *Emerging topics in translation: Audio description* (pp. 105–114). Trieste: EUT Edizioni Università di Trieste.

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