Usher Syndrome Family Stories

A beautiful insight into families and their experiences with Usher syndrome

USHER KIDS AUSTRALIA

www.usherkidsaustralia.com

About Usher Syndrome

Usher syndrome is a rare genetic condition characterised by congenital hearing loss or deafness, progressive vision loss, and in some cases vestibular dysfunction. The diagnosis may come just as a family is adjusting to life with a child with hearing loss. The age of diagnosis varies, but with improvements in genetic testing, this diagnosis is getting earlier; with some children still in infancy.

One thing remains consistent, however, and that is the utter shock and devastation families often feel when receiving the news that their child may lose their vision.

About UsherKids Australia

UsherKids Australia is a not-for-profit charitable organisation that was established in 2016 as a parent led support group to enhance the lives of children diagnosed with Usher syndrome and their families in Australia.

UsherKids Australia's mission is to ensure children diagnosed with Usher syndrome and their families have access to an informed, committed and caring community of clinicians, service providers, educators, researchers and peer support networks to allow them to thrive in their everyday endeavours.

The Schmid Family

MACKAY, QUEENSLAND

Julianne and Peter have three boys: Will (7), Hugo (5) and Callan (2). Both Will and Hugo were diagnosed with deafness through the newborn hearing screening program, but it took until Hugo was two years old, for the family to learn the boys have Usher syndrome Type 2C.

Julianne and Peter found the diagnostic process to be a really challenging time, as the medical professionals they saw had little information about the syndrome, leaving Julianne and Peter needing to do a lot of independent research themselves.

"We were teaching our specialists as much as they were teaching us."

However, both Julianne and Peter agree that a lot of positives came from the diagnosis. Once they knew the boys had Usher syndrome they were able to look for specific supports and found UsherKids Australia. It also meant they could commence specific therapies for Will and Hugo. This has allowed them to begin preparing for vision loss whilst the boys still have their sight.

"They’re typical kids, they just don’t have typical hearing and one day they won’t have typical sight."

Will is now at school and Hugo will start school next year. They have found that day cares and schools that have experience with kids who have disabilities have been fantastic at meeting Will and Hugo’s needs.

It has also exposed the boys to kids with other disabilities who are achieving fantastic things. Julianne and Peter are now focusing on providing Will and Hugo age-appropriate information about Usher syndrome as they get older.

"They can do just about anything they want to do. There shouldn’t be too many restrictions for them. There's always a way around something, that if they want to do something, I'm sure that there's a way of doing it."

The Carter Family

WELLINGTON, NEW ZEALAND

Oscar was diagnosed with hearing loss as a newborn and received cochlear implants at ten months old. Growing up, his mother (Tina) described him as being a strong, independent child, who was often clumsy and falling over. At nine years old, Oscar began to have difficulties seeing at night. After tests on his eyesight and some googling at home, Tina suggested to a specialist that Oscar may have Usher syndrome. When Oscar was eleven years old, genetic testing confirmed that he has Usher syndrome Type 1.

"I don't think we would have gotten any further if we didn't suggest it."

Oscar didn't necessarily find the diagnosis helpful because it was just a name for what he was going through. But for Tina and Dad, Daniel, receiving the diagnosis was challenging, and they wish they could have contacted another family who had gone through the same thing. At the same time, the diagnosis was useful to bring everything Oscar was experiencing together so they could get him well supported.

“So while you may not be able to participate in certain areas, you can have a full life, and potentially even fuller, because you can devote more of your attention to them.”

Oscar is now fifteen and enjoys playing the piano and making short films. He hopes to go to University to study Chemistry and Psychology. He encourages parents to not be too hard on their kids and to not expect them to be good at everything.

Oscar wants others to know that it's not that he can’t do things, just that it may take him a little longer than others. He has found that even small things that provide him with a bit more independence have made a large difference, like a vibrating alarm clock so he can wake up when he wants to.

Tina’s main message is to stay positive, reach out to other parents for support and focus on the amazing things your child can do, rather than the things they can’t.

The Alderman Family

MELBOURNE, VICTORIA

Shelley and Will found out that their son, Noah, had moderate to severe hearing loss through the universal newborn screening program. They describe the first year or so of Noah's life as being quite full on, as they learned to navigate the medical system, attended different appointments and tried to find answers. When Noah was two, and with Shelley pregnant with their second child, the family was told Noah has Usher Syndrome, Type 2A.

In the year after receiving the diagnosis, Shelley and Will used social media and UsherKids Australia to find others with Usher syndrome who are thriving and doing well in life. They both found this to be very beneficial to them, finding comfort in seeing others who have lived through what they are currently going through.

"So now that we know, it is much easier for me to navigate. There's less questions, there's less questioning."

Shelley and Will are now looking towards Noah's future. They think it will be important for Noah to understand what the future may hold for him and they plan to discuss Usher syndrome with him when he is able to understand. They hope to involve him in the Deaf community, so Noah can be around other kids who may face similar experiences to him.

“We want Noah to be proud of who he is, and that he’s doing an amazing job… but he does have challenges.”

Shelley and Will’s team recommended against going down the rabbit hole of googling everything to do with Usher syndrome, and they encourage other parents to do the same and stick to official websites.

They also suggest that parents build a team of professionals that they trust and can rely on, as their support is incredibly important. For friends and family of those with Usher syndrome, Shelley and Will recommend being a listening ear that parents can go to without judgment.

“Our eyes have been opened to all the things people with disabilities can achieve and the world of opportunities that are still available.”

The Feller Family

MELBOURNE, VICTORIA

Hollie and Dan found out that Harry, their third child, was profoundly deaf at birth through the newborn screening program. As Harry grew, he did not meet his gross motor milestones on time. After seeing Emily (co-founder of UsherKids Australia) and her son Louis in the paper, Dan began to suspect that Harry may also have Usher syndrome. After a number of genetic tests and a lot of waiting, Harry was diagnosed with Usher syndrome Type 1F at two and a half years old.

The diagnosis was a massive shock for both Hollie and Dan, but they were somewhat comforted by having connected with Emily and knowing there was another parent who had a similar experience. Hollie highly recommends reaching out to other parents, not only for emotional support, but so they can share helpful tips. She also found Facebook pages to be helpful some of the time.

> **"Every day you will wake up and the first thing you'll think of is Usher syndrome. Every night you go to bed the last thing you think of will be Usher syndrome. One day you'll wake up and it won't be the first thing you think of, and it won't be the last thing you think of. You just need to make that happen as quickly as you can, so you don't miss out on enjoying their childhood with them."**

Hollie and Dan chose not to share Harry's diagnosis with him and his older sisters for a number of years. They felt they needed to fully understand the diagnosis, what it meant for Harry and have a more positive outlook for him, before letting his siblings know. This approach really worked for their family and the kids accepted the diagnosis well when they were told.

“He never thinks that he can’t do something because he’s got Usher syndrome. He doesn’t see it as something that limits him.”

Hollie and Dan found early intervention services to be a huge help in improving Harry’s vestibular functioning. Physiotherapy, occupational therapy and gymnastics have all been beneficial. Hollie recommends searching for professionals who you trust and who work well with your kids. Having a network of other parents who can provide recommendations can be hugely beneficial in this process.

Hollie wishes others, particularly teachers, understood the impact dual sensory loss and balance dysfunction has on Harry day to day. Even though he may be doing extremely well, it does not mean that he doesn’t face additional challenges or isn’t trying hard to do well. She encourages parents to never second guess themselves when advocating for their children or feel that they are overemphasising the difficulties the kids may face. Hollie wants parents to remember that they are advocating for the hardest days the kids have, not the average or good days. She also believes it is important to organise time alone with other siblings.

“The more you can talk about it with people, the more your network expands and that opens up opportunities for you - as someone always knows someone who has tried something that might work for you .”

The Cox Family

SYDNEY, NEW SOUTH WALES

Vicki received confirmation that her daughter, India, had profound hearing loss when she was fifteen months old, the day before the birth of their second daughter Samira. At three weeks of age, Samira was also found to have profound hearing loss. It wasn't until Samira was ten years old and struggling to see on a low light walk through the Daintree Rainforest that they sought further testing for her eyesight. An ophthalmologist confirmed that the sisters have Usher syndrome that year.

Vicki began going to different conferences throughout Australia and internationally to learn more about the syndrome and meet other families. She believes she has received the most useful information from groups like UsherKids Australia and informal support networks such as Facebook pages.

"It's been good just to have a few families to connect with and have that support. It's been really valuable."

Now 21 and 19, India is studying science at university and Samira is studying screen and media at TAFE. Being able to advocate for themselves in higher education settings has been important for them to receive the support they need. Samira has found it beneficial to share her story with friends and classmates, so they can be accepting and know how they can help. She also enjoys competing at para-athletics events and being a part of the athletics club.

“We’ve always sort of had you know just set expectations like any other kids that study, do as well as they can at school, go on and pursue whatever they want to. So we always have that expectation that you’ll go out and do things.”

Vicki wishes others were more aware of how everyday things can be more challenging and tiring for India and Samira. Whilst friends and family have been supportive, it has been easy for them to forget the little things India and Samira need help with to get by. Vicki believes it is important to be open and honest with loved ones, so they understand the child’s needs and are able to help out.

She also wishes health professionals had more information on the syndrome, so they could provide better support for the families or link them up with appropriate supports.

“Just enjoy the kids, don’t get too caught up in all the therapy and what might happen, you’ve just got to get on with life, make the most of it.”

The Doak Family

PERTH, WESTERN AUSTRALIA

Bronwyn and Andrew's boys, Eamon and Kealan, were diagnosed as being profoundly deaf as infants. However, Bronwyn believed there was more going on, as Eamon was having difficulties with his balance. It wasn't until Bronwyn and Andrew underwent genetic testing themselves when Eamon was three and Kealan was two, and two letters later, that the boys were diagnosed with Usher syndrome Type 1D. Bronwyn said they received one letter informing them that there was no genetic cause to the boy's deafness, only to receive another two weeks later, confirming that the boys had Usher syndrome.

> **"That was just a horrible, horrible couple of months... I couldn't cope... I thought this is just too much for us."**

However, despite this extremely challenging way of receiving a diagnosis and a few unhelpful doctors, Bronwyn was able to bring together a team of professionals to support the boys. She found ophthalmologists with expertise in Usher syndrome, as well as psychologists and occupational therapists through speaking to other families of children with Usher syndrome. All these people came together to tell Eamon and Kealan about the syndrome and what they may experience as they grow up. The family has always been transparent with the boys about the syndrome and have found this strategy to really work for them and helped them prepare for vision loss.

“My boys are beautiful advocates for people with vision impairment. They will tell any person who wants to know why they use a cane.”

Open communication between the family has helped Eamon and Kealan become advocates for themselves as well as others with Usher syndrome.

In 2019, Eamon spoke in front of Parliament in Canberra about Usher syndrome and is a 2020 Telethon ambassador. Bronwyn encourages others to get their kids involved in things like sports and social activities early on. The family have also found it helpful to begin cane training whilst the boy’s vision is still good, so they can call on this skill easier down the track.

Bronwyn encourages parents to get in contact with other parents within the Usher community, as they can be fantastic sources of support and knowledge for managing the NDIS and finding supportive medical professionals.

The Shepard Family

MELBOURNE, VICTORIA

After bringing their second child, Louis, home from hospital, Emily and David knew something wasn't quite right. His loud, two-year-old sister was running around their home, making all the noise toddlers do, but Louis did not react to any of the sound. Emily decided to take Louis for a hearing test, where the audiologist confirmed that Louis was profoundly deaf. Despite feeling enormous shock and grief regarding the diagnosis, Emily's internet research and experiences with early intervention services led her to feeling positive about Louis' future.

"Don't spend so much time worrying about your baby that you miss out on all the good bits… it's easier said than done I know but I would say, just enjoy it. It's going to be okay."

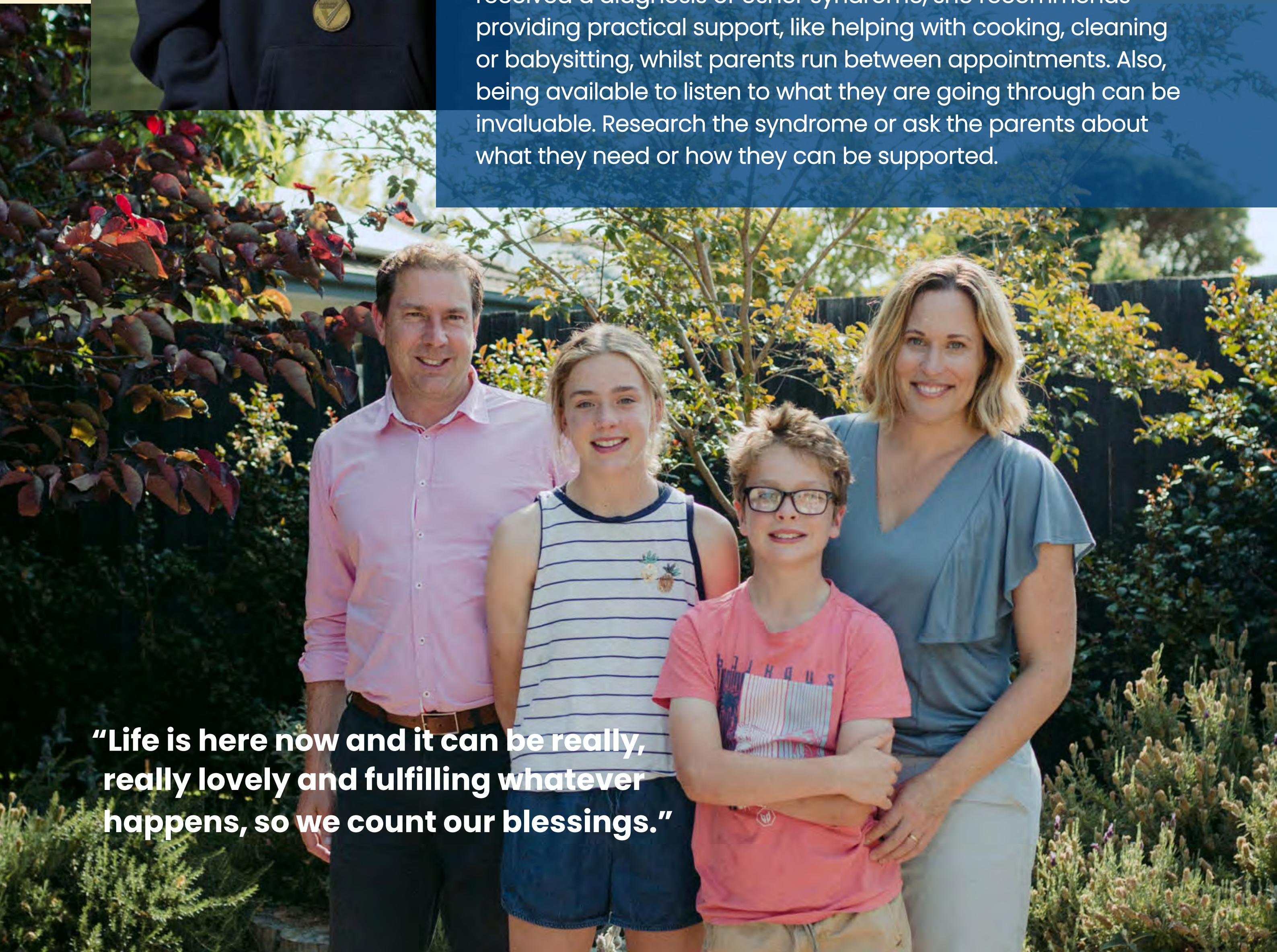
However, Louis was not meeting his gross motor milestones, and Emily again felt that there was something more going on. After suggesting a diagnosis of Usher syndrome to her ophthalmologist, only to be dismissed, the family went through genetic testing which confirmed that Louis has Usher syndrome Type 1.

Emily and David left the appointment desperate for support or another parent to speak to but found nothing. This feeling of isolation led to Emily co-founding UsherKids Australia to help other parents feel connected and supported.

“It might not be exactly what you envisioned for your child but all of the wonderful things about kids can still happen. They will still have the capacity to go to university and get married and travel the world.”

Now, eight years on, Louis is playing football, cricket and likes rock climbing. Emily says he does everything other ten-year-olds do, just with some clumsiness. He can advocate for his needs at school, something Emily recommends other parents teach their children to do. They speak openly about his diagnosis and there are no secrets.

Emily hopes that other families can find and seek support. She believes the first week or month after receiving the diagnosis is the hardest and that outside support, such as family counselling or contacting another family, can be hugely beneficial. For those trying to support a family who has just received a diagnosis of Usher syndrome, she recommends providing practical support, like helping with cooking, cleaning or babysitting, whilst parents run between appointments. Also, being available to listen to what they are going through can be invaluable. Research the syndrome or ask the parents about what they need or how they can be supported.



“Life is here now and it can be really, really lovely and fulfilling whatever happens, so we count our blessings.”

The Sandles Family

MELBOURNE, VICTORIA

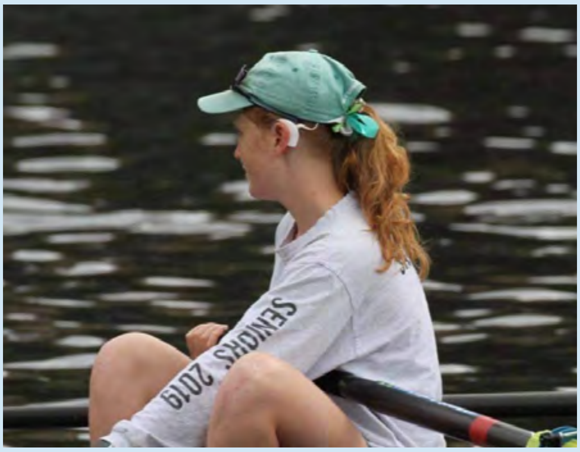
Maggie determinedly completed her secondary schooling in 2020, and is now eighteen in her first year living independently whilst studying at University. She continues to love sport and spending time with her friends outside of school. Maggie was diagnosed with deafness at eight months of age, after many months of her mother, Bronwyn, telling medical professionals that she believed Maggie was deaf. Maggie was then fitted with a cochlear implant at twelve months and, at six years old, she needed her first pair of glasses. However, it wasn't until an optometrist referred Maggie to a specialist at fourteen years old, that she was diagnosed with Usher syndrome. At this stage Bronwyn was a Teacher of the Deaf and, despite being aware of Usher Syndrome, never thought anyone she knew would be diagnosed with the syndrome.

Mum Bronwyn described both the diagnosis process and medical support to be inadequate and frustrating. From the Optometrist who blurted the diagnosed to Maggie without warning or support, to subsequent appointments with medical professionals, the family felt unsupported and uninformed. They were not told the impact this diagnosis may have on Maggie, with most of Bronwyn's information about Usher syndrome coming from the internet. The lack of integration between different medical professionals and services was also challenging to navigate.

"The kids need to have something that's really positive in their lives so they need to have some outlet. "

Maggie said it took quite a while to come to terms with her diagnosis, and that she is unsure if she is yet to fully accepted it. From a young age, Maggie has found participating in sports to provides her with many opportunities, including making social connections. Sport has been such an impactful power in her life on all levels, from playing Deaf Netball and Football to rowing at State level. She now has her sights on trialling for the Paralympics in paratriathlon one day. Bronwyn agrees, saying sport has been an unexpected positive and is the most organised of all activities aimed at children with deafness.

“They need to grab hold of something that they can excel in and direct them into a way that has social opportunities for them.”



Bronwyn has made a conscious effort to not treat Maggie any differently from her two siblings. They focussed on providing Maggie with many opportunities, from small things like travelling far and wide to increase language exposure and early life on the family farm with her extended family, to flying to Perth for a Deafblind conference which allowed Maggie to meet others with Usher syndrome. Maggie believes these experiences had a positive impact on her self-determination and assisted in the development on her speech and language when she was younger.

Bronwyn recommends that all parents do their own research and educate themselves on Usher syndrome and make connections through social media. She believes knowledge will greatly help parents be the best advocates for their children and ensure their kids get all the resources they need to succeed. Maggie encourages other kids and teens to be open and honest about what they need, saying ‘make it easier on yourself and ask for help’.

“Always do your own research as you have to become the expert if you’re going be the best advocate. ”

This resource has been produced by UsherKids Australia in the hope that it brings a sense of belonging and hope to newly diagnosed families. We thank the eight Usher families involved for their openness and willingness to share their personal stories.

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