The Unfinished Story of Usher

By Julianne Schmid

Our story starts

...like that of so many other families, but we are not sure how it is going to end.

My children are pretty much just like any other child their age. They go to school and day care, play sport, music and participate in everything that other kids their age do.

The most obvious difference is that they wear hearing aids and and are being prepared for life in the future with limited, or possibly no vision.

And we don't know when that will happen.

When our eldest child, William, was born in 2014, his newborn screening tests picked up an issue with his hearing. We had to drive almost five hours to Townsville, from our hometown of Mackay (QLD) for secondary testing.

When you first get the results and the nurse tells you, 'It will be all be fine, you've got no family history of hearing problems,' you try not to worry too much.

But our Townsville hearing test revealed our eldest child was hard of hearing. The audiologist said, 'I'm sorry he has congenital hearing loss.' Two years later (2016), our second child, Hugo, was born and had the same issues with his hearing.

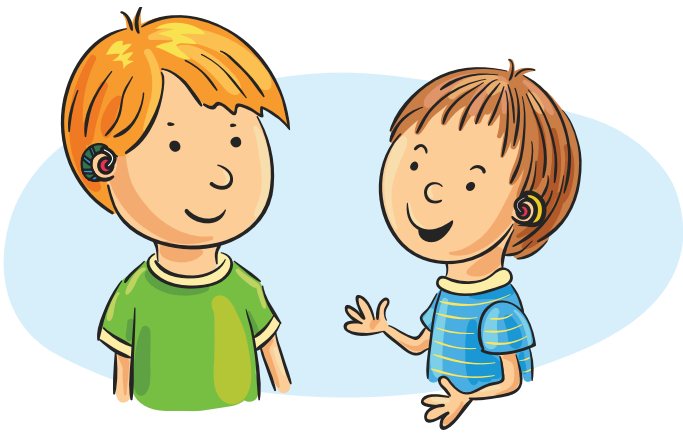
In 2018, when William was 4 and Hugo was 2, and I was nine weeks pregnant with our third son Callen — an appointment with a genetic counsellor revealed that our boys had Usher Syndrome Type 2C.

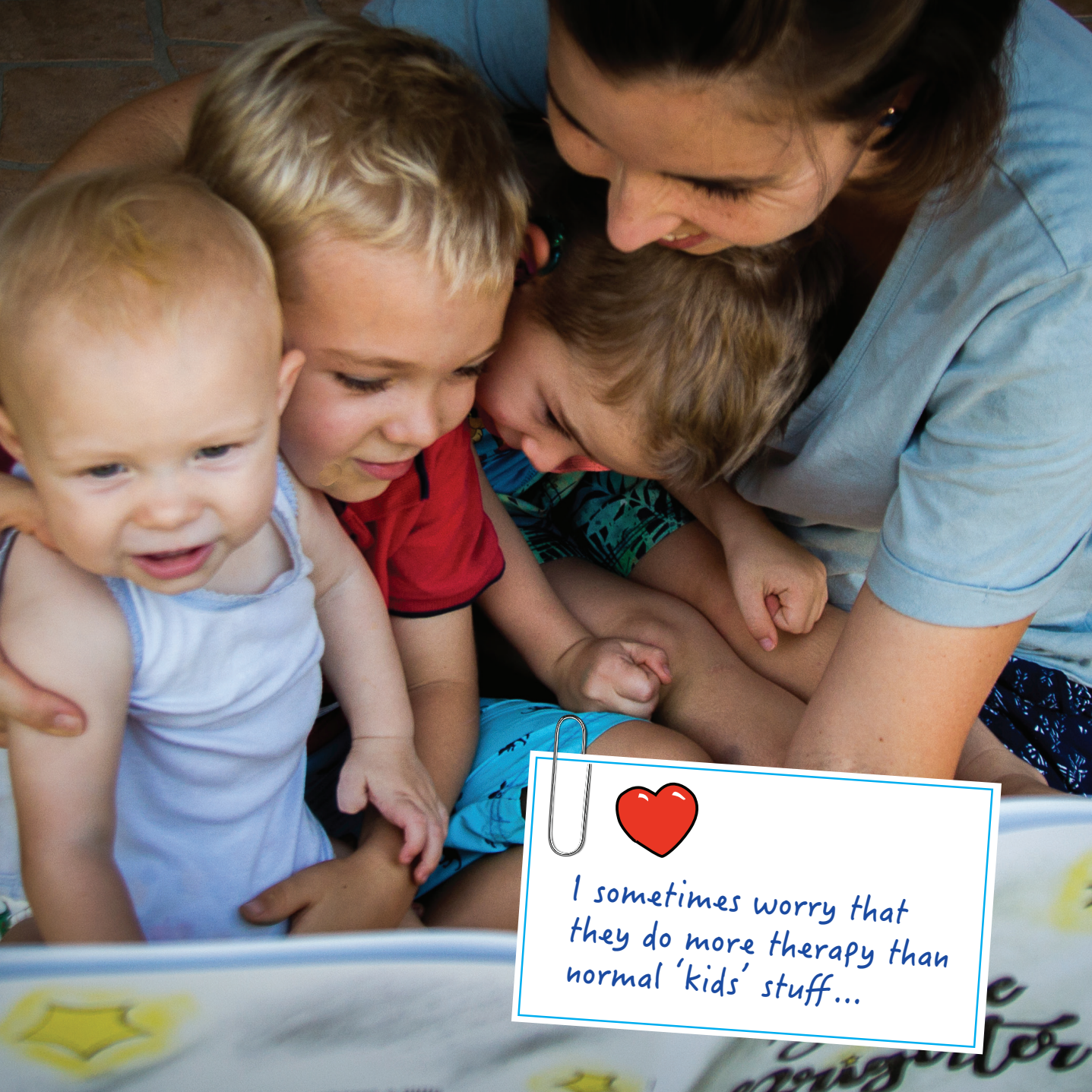
This meant our two eldest boys would eventually lose their sight in conjunction with their already compromised hearing. Amazingly, Callen does not have any signs or symptoms of Usher Syndrome.

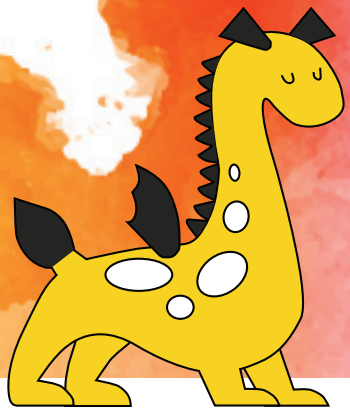
When our fears became our reality, we were angry and extremely sad. That was where our story as an Usher family really began.

In many ways, we had been preparing ourselves for the worst, and although we hoped for the best, we knew that there was nothing in our power that could change the outcome.

It's funny because the two older boys are very aware that the youngest doesn't wear a hearing aid and they say "I wish I had ears like Callen" but the little one puts on the hearing aids because he wants to be like his big brothers. I wait for the day that the boys draw pictures of themselves with hearing aids, then maybe I'll know it's okay.




I sometimes worry that they do more therapy than normal 'kids' stuff...



Will and Hugo love their toy dragon. They love books and are little bookworms. They go to school, play sport, music and participate in everything that other kids their age do.

Both boys also attend regular speech lessons and have therapy to help with balance and coordination in the form of physiotherapy and occupational therapy. They see an audiologist and ear, nose and throat specialist annually, and they now also see ophthalmologists biannually to monitor their vision. We have also incorporated orientation and mobility sessions a few times a year to help prepare them for a life with compromised vision.

I sometimes worry that they do more therapy than normal 'kids' stuff...

so I hope that the extra- curricular activities will assist the therapy, because being a kid is one of the most important jobs in the world.

Being a real water baby myself, the boys also love the water and so it is important to me that the boys learn water safety. They have regular swimming lessons, but they can't wear their hearing aids in the water and that really impacts them significantly. It's hard for them to pay attention and to learn what they need to know. Something that should be full of fun for them, can be really hard.

Their favourite activity is a combination of gymnastics, parkour and martial arts. It stimulates and challenges them. They are jumping on boxes, climbing up ropes and jumping into foam pits.

Right now, all we can do is explain their condition in a way that is appropriate for them, give them the tools and training to assist them when their vision is impacted, and meet them where they are at, which means letting them be kids and doing whatever it is they need to do to be who they are.

We know that if we don't talk about Usher Syndrome and other rare genetic disorders, no one is going to get the help and information they need to have a positive impact on their child's future.

When we learned about the research surrounding Usher Syndrome, we thought, 'wouldn't it be great if, in our kids' lifetime, there is a genetic cure?' We hope for a breakthrough; we've heard about clinical trials, and about the exciting progress being right on the edge of something big happening.

In many ways, research is moving faster than anyone is ready for. They can say what the genetic issue is, but they don't necessarily have the medicine to treat it yet.

This is why the work happening at Children's Medical Research Institute is so exciting. Their goal is to turn genetic knowledge into treatments and even cures and to do this as soon as possible.

We have high hopes that things will happen in our children's lifetime—maybe even before they lose their vision. If not, we are paving the way for future generations.

I would like to think that genetic research might one day shed some light on ways to either stop or slow down the progression of even more diseases that are genetically inherited. This is why research happening today is so important for the future.

When you get a diagnosis, you're not given a lot of hope. To be told, 'there is no cure', and to think that now there is a possibility, in our kids' lifetime, that some progress could be made, and research could stop the disease developing—that's really exciting.

This Christmas will be like any other, the kids will have a long list of things they are asking Santa for! My Christmas wish is much more simple: for my kids to have a "normal" childhood. In the meantime, we'll just keep working on doing all the things they want to do, while they can still do them. We don't know what future Christmases will be like for them.

I'd love my children to have the opportunity to live a life without a genetic disability. That would be our happy ending.

You can change the ending to our story...

Julianne


Will you help give William and Hugo the chance they deserve? Change the story for Usher Syndrome and other genetic diseases by donating today.
Donate today at
https://bit.ly/future-without-usher
CHILDREN'S MEDICAL RESEARCH INSTITUTE
Jeans for Genes®