MY CHILDREN HAVE AN INVISIBLE DISEASE

LIKE MOST PEOPLE, I WAS EXCITED TO HAVE MY FIRST CHILD. LIKE MOST PEOPLE, I ASSUMED I WOULD HAVE A HEALTHY BABY.

When I had Henry, he was very sleepy and didn't want to feed, but I thought I was an over-anxious first-time mum. Less than 48 hours after he was born, I remember this overwhelming, horrible feeling inside me that something wasn't right. It was 4 am, and I called the midwife to say he doesn't look right. They took him away and told me to get sleep, but an hour later they came back and said, **'You have a very sick baby.'**

I went into the special care nursery, and there was this doctor, and he was saying that this baby needed a lumbar puncture. The child was under a heat lamp, and it had all these tubes attached, and I thought, 'I'm glad that's not my baby, my baby's not that sick.'

But it was Henry, and he was that sick.

Nobody knew what was wrong. He got better, and we were sent home.

Then we got a call from the pediatrician saying his newborn screening tests showed something odd, but they couldn't put a name to it. Then we saw a metabolic specialist who said Henry had Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency, or LCHADD.

LCHADD is a rare genetic disease. The first explanation was that it is a rare condition that prevents the body from converting certain fats to energy, particularly during periods without food (fasting). Because of that, we were told he would be on a very strict diet for the rest of his life, and if he became ill he would have to be rushed to hospital.

What I didn't know at that point was the potential complications that could lead to him going blind, weakening of the heart, and permanent nerve damage.

I went through a period of grief for Henry. I had this beautiful baby that I adored, but I wanted to bundle him up and put him back inside me to protect him.

When I found out I was pregnant again, this time with Rosalie, we had the screening and found out she had LCHADD too. We had a one in four chance, so the numbers didn't work in our favour. I went through that period of grieving again. I was grieving the opportunity for my children to live a long and healthy life – that's the reality.

Henry and Rosalie won't eat food. They are on a special formula. Henry drinks it himself, but when Rosalie won't, I put it in a feeding tube. They see physiotherapists, speech therapists, occupational therapists and gastroenterologist – we're always in some sort of waiting room.

Henry has now been diagnosed with autism as well. He's very high functioning. Henry learns really fast and he's such a happy boy.

Rosalie is my sassy one. She's determined, and she hits the ground running. She's a great complement to Henry.

Despite the setbacks, Jess and her husband, Tim, count themselves blessed.

"We are so lucky to have two healthy children despite their diagnosis. If our babies had been born 30 years ago, they likely would not have survived past infancy. We are lucky to have access to a medical team that do so much for our children. We have online support and are able to speak to other families with children who have LCHADD and similar conditions - and we share in the happy times and the hard times."

Henry and Rosalie have such a rare disorder, that their future is unknown. Jess says she's been reassured her children should be able to lead happy lives if they continue to monitor them so closely.

"I pin my hopes on organisations like Children's Medical Research Institute (CMRI), that push research and advance knowledge in areas that may have a follow-on effect for things that relate to my children's future, even in the obscure rarity of their condition."

Gene therapy is a key area of research that will help children like Henry and Rosalie and many others who suffer from genetic conditions. Your support today to Dr Leszek Lisowski's gene therapy research will help accelerate delivery of its benefits.

Jessica Gowans