

Charlize and Issac's Story

by mum, Julie Gravina

I always suspected something wasn't quite right with Charlize.

The nurses said she must be quieter than her twin brother Isaac, but when we got home on day four from hospital we learned there was more going on.

In the early hours of the morning, I fed Isaac. I went to wake Charlize up, and she just wasn't waking up. She was cold, and her face was very grey. She wasn't moving. I woke up my husband, Paul, and told him something was really wrong, 'we have to take her to hospital'.

I had this sinking feeling she was dying - and that's what was happening.

We got her to the special care nursery, and they were trying their best to figure out what was going on with her. She was getting worse, not waking.

The worst thing was - Isaac was going down the same way. No one knew why.

The next day, newborn screening results came in. It confirmed they both had a severe metabolic disorder. The diagnosis was Propionic Acidemia.

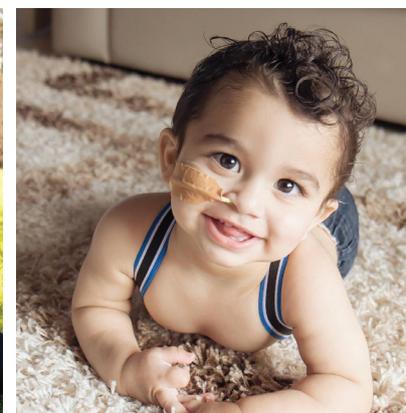
The doctor took us to a quiet room, and he explained the enormity of the diagnosis and what it meant.

It was hard hearing the diagnosis, because the outcomes were very poor. And the treatment was very difficult. It was explained the twins would need constant feeding day and night. They couldn't run out of energy; if they do, they break down muscle and release protein. Protein is like poison to their body. When they have too much protein, their body would, basically, end up with large amounts of dangerous toxins, which very quickly can give them brain damage.

Isaac had sudden brain damage at nine months old. We couldn't stop the progression of the disease. After Isaac received his brain damage, he was still the same little boy underneath - a lovely, lovely little boy, but he just lost his muscle tone and coordination.

A liver transplant was discussed. I was shocked the doctor said it was needed for both children. He said Charlize was also at risk of having sudden brain damage - just like Isaac.

It took around eight months for Isaac to receive his transplant, and it wasn't without its difficulties. Isaac had massive complications, but at six weeks post-transplant he was discharged from hospital.



After Isaac received his new liver he was on top of the world. He was learning at lightning speed and loving his new life. But he was only home for one month before he got septic shock and passed away very quickly.

It was very hard for our family, grieving the loss of Isaac but also having to concentrate on Charlize.

They said Charlize was like a ticking time bomb. It was a really hard decision, after we lost Isaac, to decide to transplant Charlize, it was only that her disorder was so severe.

I have no idea how we got through it. Four months after receiving her transplant, Charlize went into rejection in a pretty massive way. She experienced liver disease, she was jaundiced, itchy, bruises and lumps all over her. She'd be awake for only an hour a day. We're so grateful her next liver transplant came just in time, because on the morning of the transplant she went into liver failure.

It's been a year since Charlize received her second liver transplant. In the last five to six months, she's been in a lot of pain from multiple invasive investigative procedures as her body goes into mild rejection

For Charlize to have survived two liver transplants, to come out of it so happy and loving hospital - I think it's incredible.

Supporting research means so much to our family, because Charlize's life relies so heavily on receiving better treatments and cures.

We've given Charlize's liver cells to Professor Ian Alexander at Children's Medical Research Institute, and his team are working on gene therapy for Propionic Acidemia right now.

They are trying to get to the point, where for children born with Propionic Acidemia, gene therapy will either edit or replace the faulty gene. This will eliminate the need for a liver transplant and the children can go ahead with having a happy, normal live.

I think that is just so exciting. It's more than you could ever wish for.

