

Mum's heartbreak over twins' diagnosis

It is every mother's worst nightmare and being a parent of twins, the situation Julie Gravina found herself in was even more heartbreaking.



Remembering the morning she discovered one of her twins “cold and grey” still brings tears to the eyes of mother Julie Gravina.

Her twins had been in a comatose state for the first few days of being born and even afterwards Ms Gravina said she'd always suspected that something wasn't quite right with Charlize as she was always quieter than her more boisterous brother. Then one morning Ms Gravina went to feed her baby boy Isaac, who was crying.

“By the time I got to Charlize to give her her feed, she was cold and grey and she wasn’t moving,” Ms Gravina said.

“That’s when I knew, I just thought straight away, I thought that she was dying.”

Sadly, the truth would be even more upsetting than she realised.

After taking Charlize to the special care nursery, Isaac also started getting sick and no one knew why.

Seven days after she gave birth, the twins’ newborn screening results came in and it confirmed both babies had a severe metabolic disorder called Propionic Acidemia, a condition that impacts less than one in 100,000 people.

Those with the condition are unable to process protein components properly and this can lead to high levels of ammonia and acids in the body.



A metabolic specialist strongly recommended that Ms Gravina and her husband Paul think about liver transplants for the twins.

“I was shocked that he said for both Isaac and Charlize,” Ms Gravina said.

Isaac was the first one to undergo the transplant at when he was one year old because “he would show us that he was fine but he was the one who would always get the sickest”.

Six weeks after the transplant Isaac was discharged from hospital but he was only home for one month until he got septic shock and died very quickly, just before he was due to turn two years old.

“It was really hard for our family, grieving the loss of Isaac but also putting that aside as well and having to concentrate on Charlize,” she said.

Within months, daughter Charlize had to have the same procedure which killed her brother.

“It was a really hard decision, after we lost Isaac, to decide to transplant Charlize. It was only that her disorder was so severe,” Ms Gravina said.

“What happened to Isaac was just a very unfortunate set of circumstances and Charlize will have her own story to tell. She’ll have a very different story.”

Charlize, now four, has had two liver transplants and Ms Gravina is hoping that more research will give her a future.



Ms Gravina will be sharing her story at the [Denim Dinner](#) on Saturday, June 22 at The Star in Sydney.

The [Jeans for Genes](#) event will raise money for the Children’s Medical Research Institute, which is committed to developing treatments and cures for children’s genetic diseases.

Charlize’s liver cells have been donated to the institute for research.

“I hope in Charlize’s lifetime that she will receive better treatments and possibly cures,” Ms Gravina said.

“They are trying to get to the point where, instead of children with Propionic Acidemia needing a liver transplant — they’re working on gene therapy.

“I think that is just so exciting, given the diagnosis that our children were given, and the outcomes, it’s more than you could ever wish for.”

Her hope is shared by father Chris Fenn whose son Ethan, 5, has Cystinosis, which is a condition that can impact everything from his kidneys to his eyes.

“No parent expects to hear their child has an incurable disease,” Mr Fenn said.

“It hit pretty hard.

“It really brings home what a cure is. Before that — a cure was just a word.

Now to think, through research, we are getting cures for things we used to call incurable — I can’t imagine anything more important to us. It’s only through research that we can change incurable to curable.”

Link to story: <https://www.news.com.au/lifestyle/health/health-problems/mums-heartbreak-over-twins-diagnosis/news-story/6b1e2f2eaebb1ede6d21ccc41b4af505>