‘We had better odds of winning Lotto but instead, we won Quinn’

Madeline Cox | July 31, 2018

Quinn is unable to see, walk or speak, the result of a single gene carried by both his parents.

Sasha and Rhett Campbell couldn’t believe how perfect he was. After four years of trying, several miscarriages and rounds of IVF, the fact that Quinn was finally here felt like a miracle. "He came into the world in a textbook delivery, he was a beautiful little baby boy," Sasha told *Kidspot*.

“He did all the right things in the first year of his life like any other gorgeous baby.”

Then suddenly Quinn’s progress slowed, he never started to crawl, let alone walk. Talking was also out of the question, the one-year-old didn’t even react when his name was called. Sasha began obsessively googling her son’s symptoms, becoming more and more convinced that something wasn’t right. The 39-year-old didn’t know how her family would cope.

He was a perfect baby. Images: Supplied

Wait and see

Then one day she stumbled upon a quote that changed everything. “It said basically it’s very normal to ask why us but it doesn’t actually help, the best thing you can do is ask what you can do about it,” Sasha recalled.

Overnight, she and Rhett decided to take Quinn in for testing. They needed to know exactly what was wrong with their boy so they could try and fix it. Unfortunately, it wasn’t quite that simple, with the doctor telling the couple that “it could be something, it could be nothing, there’s no guarantee that anything is actually wrong.”

All the couple could do was wait and see if Quinn eventually met his developmental milestones. If not then he would go in for further testing.

“I remember just constantly questioning what should we do and why should we do it and what was the best plan of action,” the mum-of-three remembered.

It was only after his first birthday, they realised something could be wrong. Image: Supplied

Further testing

Eventually, it became obvious that Quinn wasn’t making progress and further tests were secluded. It was also around this time that their GP noticed Quinn bringing toys close to his face while waiting for an appointment. He suggested that in addition to the other tests, the one-and-a-half-year-old also get an eye test.

“It was at the eye appointment that the ophthalmologist noticed corneal clouding on his eyes which he said was an indicator of a metabolic disease,” Sasha said.

They thought they finally had an answer. But unfortunately, the family has quite a wait ahead of them. Metabolic diseases are so rare that only one laboratory in Australia is able to test for them so the backlog is significant.

“Every two weeks I would ring the doctor to see if the results were in and of course they weren’t so it was back to square one, one step forward, two steps back,” Sasha said.

Quinn, pictured here with his brother Laif, is unable to walk or talk. Image: Supplied

A rare condition

Finally, after six months of waiting, Sasha and Rhett were told that Quinn had Mucolipidosis type IV, a metabolic condition so rare that it’s estimated only 100 people worldwide suffer from it. Quinn inherited the disease as a result of a single gene that Sasha and Rhett didn’t realise they both had. At this stage, there is neither treatment nor cure for the disease.

“Both my husband and I carry this rare gene and we had better odds of winning lotto but instead we won Quinn,” she said.

Mucolipidosis type IV affects people at a cellular level, damaging the Lysosome, which is the part of the cell responsible for repair. Instead of recycling old products, Quinn's cells allow them to build up and become toxic.

“From the outside, that means that Quinn can’t talk, he can’t walk, he doesn’t crawl, he’s vision impaired but will be completely blind by the time he’s 12,” Sasha said.

“He needs help with every single task.”

The two siblings love their brother so much. Image: Supplied

“We all help”

The entire family pitches in to help Quinn. From an early age, Sasha and Rhett’s other kids (Laif, 10, and Harlow, 2 neither of whom have the condition) have cared for their brother; something that Sasha believes has helped to be more compassionate, caring towards others.

“Our family is a team and everyone knows they have their role and the kids look out for their brother,” Sasha said.

“We will ask him do you want something to eat and he will often giggle as a yes or just shake his head for a no.”

“Even though he goes through so much Quinn is such a happy young man.”

He's such a happy kid. Image: Supplied

Jeans for Genes Day

It was for that reason that the family wanted to get involved in Jeans for Genes Day, to show other families in similar situations that "there's so much to enjoy when you slow down and look at things from a different perspective."

Awareness is also another big concern; it's the key to finding a cure for Quinn, although the family know that they are lucky to have any answers at all.

"I often think about what would happen if I lived in a third world country, there would be absolutely nothing," Sasha said.

"I hope one day there will be answers for every single family that end up in this situation."

You can donate online at www.jeansforgenes.org.au. For more information or to register your support visit jeansforgenes.org.au.