

### SCID Member Stories – Jasper, WA



I gave birth to a perfect baby boy on the 28<sup>th</sup> of May, 2016. He was healthy, and chubby and all of the best things a baby can be. He slept well, he ate well and he was in the top 97<sup>th</sup> percentile for his height and weight at his first two appointments.

When he was around four months old he contracted conjunctivitis from his older sister. No big deal - we had treated this condition before and it usually cleared up fairly quickly. However, this time it did not. We started antibiotics finally after working to cleanse and use topical treatments. I started to get a nagging feeling that something wasn't quite right. However, I pushed my fears aside and hoped the treatment would start to work. As his eyes finally started to heal, he came down with some kind of virus. At first I thought it was just another cold, but then he started to cough and labour with his breathing. I took him to hospital for an assessment and told them about how it had started and how his breathing was laboured. I was told "It's just a virus, he has no temperature" and so I took him home.

A few days later he started coughing up clear fluid in a big bubble and then inhaling it. I took him back in to hospital and explained what was happening. He was weighed and I was shocked to hear that he had lost 500 grams. I was once again told to go home as it was "Just a virus and he had no temperature". I started to question myself and started to feel like maybe I was imagining things. A week later after watching his breathing continue to get worse and the instances of him sucking down these bubbles, I took him back to hospital once again under the rolling eyes of the on call doctor. This time though Jasper did cough up his big bubble in front of the doctor. He had also lost almost an entire kilogram and that is when they started paying attention.

He was sent to have an x-ray and had to have an intraosseous needle through his shin because he was so dehydrated. He required oxygen as his levels wouldn't hold and we were on the next Royal Flying Doctor Service flight out of Esperance to Kalgoorlie Hospital. He was around 5 months old.

We were at hospital for a week where we had fantastic care, although the doctors seemed stumped as to why he wasn't responding to antibiotics and why the infection continued to spread. He had at least five x-rays in that week. By the end of that week I had a sick feeling that he wasn't improving so I requested to be flown to Princess Margaret Hospital for specialist care. We spent another week in Princess Margaret Hospital on intravenous antibiotics that started to improve his oxygen flow, but even leaving the hospital there the final x-ray showed that it had spread through his lungs. He also contracted gastro while in hospital and continued to lose more weight. On discharge I made the decision not to head home until I was sure he was ok. I waited a few days and I was not happy with his breathing, so I took him back into the Princess Margaret Hospital emergency department where I was assured he was fine and sent home with oral antibiotics for a 10-day course.

We flew home and for a little while, Jasper seemed to be improving. He wasn't vomiting any more and he seemed to be sleeping well and eating well. However, he wasn't gaining weight. We got through Christmas and New Years and I was starting to see him decline again. I spoke to my husband on the 2<sup>nd</sup> of January and told him that I was taking him back into hospital and I wasn't coming out until I got some answers. Even he thought I was crazy at this stage. On the 2<sup>nd</sup> of January, 2017 we went back into hospital and we wouldn't come home again for seven months.

The horrors I watched my little boy go through in the following months broke my heart in more ways than I could have ever imagined. Being on the respiratory ward we were in line to be tested for cystic fibrosis. I could tell by the way the doctors and nurses were behaving that it is what they were expecting us to be diagnosed with. Luckily a new doctor came on ward to check him over and ordered every rare disease test he could, from

tuberculosis to immunodeficiency. This is how we got our diagnosis of Severe Combined Immune Deficiency or SCID.

All of a sudden I was seeing specialist after specialist. I was being asked if I had any support. I was being asked if I understood how serious the situation was and if I was prepared for stem cell transplant. Most of these meetings are a blur to me now. I remember bits and pieces but mainly I remember the worry on the faces of the immunology team, the haematology team and the oncology transplant team. These were the smartest people I had ever met and they were concerned.

The next three months were full of ICU visits, surgeries and medications. We were constantly waking up to the sound of the IV kinking or finishing, beeping loudly. I was far away from home and my husband and daughter. I couldn't see them due to the risk of another infection and Jasper already being so sick. I became reclusive and paranoid about germs and anyone trying to enter our room. I had to be though, as no one seemed to understand the concept of 'do not enter'.

When it was time for transplant and after Jasper had spent months being treated with IV antibiotics, antifungals, antivirals and total parenteral nutrition, I was told at best that he had a 50% chance of survival. Normally a transplant would never be considered for anyone who was as sick as Jasper, but it was literally our only option. Thanks to the brilliance of Dr Shanti, Fiona and all the other brilliant staff, a month later we were allowed out into outpatient care at Ronald McDonald House where we spent a further three months waiting to be healthy enough to head home.

There was so much more to it than that. It was complex and terrifying. There were moments that he was so sick that I begged god to take him if he wasn't going to let him live because he was in so much pain and obvious suffering that I was struggling to watch him go through it.

Thankfully he is a survivor and stronger than any person, adult or child that I know. But the physical scars are not the only ones that he carries. Because he was so unwell for so long, his development stopped in its tracks and he is about eight months behind socially and emotionally. He has trauma related issues whenever he has to have needles or go back into hospital. He becomes overwhelmed with fear, even when he is trying so hard to be brave. It's something we have put a great deal of work into moving forward from, but we still have a long way to go.

If Jasper had been diagnosed at birth, it would have allowed us to protect and isolate him from the very beginning and he would not have gotten as sick as he did. Treatment would have been much quicker and less intensive. He could have avoided months of treatment and instead have gotten through it possibly in as little time as a month with a lot less trauma attached. I wouldn't wish what we have gone through on anyone, and we are one of the lucky ones. I got to take my son home. Not everyone with this condition gets to do that.