

### SCID Member Stories – Max, VIC



Our son, Max, was born in March 2017, full term and healthy. At three weeks he got his first cold and somehow he managed to fight it off. However, at six weeks he caught another cold and then croup at ten weeks. After many, many visits to the GP we were told "kids often get virus after virus". After the last cold, as she listened to his chest she said to take him to hospital if he gets worse.

At three and a half months Max had his first hospital admission for bronchiolitis, para flu 3 and rhinovirus. We stayed in there for a week, Max was on high-flow and then they sent us home. A week later I took him back to hospital.

We stayed for another week. He was back on oxygen with 'another' case of bronchiolitis and still para flu 3 and rhinovirus, narrowly escaping picking up whopping cough. They sent us home when he reached 92% oxygen. The RDNS nurses came to visit us at home and you could tell he wasn't right.

In July, on the third daily home visit, the nurse said he needs to go back. I took him to the WCH. He was mottled, so they rushed him to the resuscitation room and eventually admitted him for a short stay where he stayed for two weeks on oxygen. They kept trying to wean him off but couldn't. He still had para flu 3 and rhinovirus. We pleaded with the doctors not to send him home until we knew what was wrong. I remember asking if there was something wrong with his immune system. They didn't think so and told us that some babies just take longer to get better. Eventually they did test and what they found was terrifying and turned our lives upside down.

As they were training me up on how to use oxygen at home on August 11<sup>th</sup> 2017, we received the devastating news that our five month old baby Mighty Max had X linked Severe Combined Immune Deficiency (SCID) - "the most serious form of primary immunodeficiency...a rare disorder, thought to affect less than ten Australian children born each year (<https://www.allergy.org.au/patients/immune-deficiencies/severe-combined-immunodeficiency>)." Signs went up on his door about wearing a gown, mask and gloves to enter. Thankfully he had his own room each time because he was infectious with paraflu, protecting others! His big brother Blake was no longer allowed in and he wasn't allowed any visitors.

Once a positive pressure room became available, that became our 'bubble' for the next six weeks. Max had a GA where a PICC line was inserted for ease of medication delivery, blood tests and many, many transfusions of IVIG. On top of his paraflu 3, Max had PJP in his lungs. He required 24/7 oxygen and high flow to breathe. He was struggling to breathe. I had to stop breastfeeding so I didn't pass anything on to him. They gave Max the RSV vaccine to give him some protection. I remember a specialist telling me that it's an expensive vaccine.

We were told that Max needed a bone marrow transplant to survive and that he needed it as soon as possible. We could choose between Melbourne and Sydney to have it done. The next morning Chris (Dad), Blake (three year old brother) and I got tested to see if we were a match. Fortunately, his brave brother, Blake, was a perfect match! We chose to go to Melbourne for the transplant.

On 19<sup>th</sup> September a room became available at the Royal Children's Hospital Melbourne. Max and I were flown there by Medstar in a Royal Flying Doctors plane because of his oxygen requirements and because he couldn't risk going on a regular flight with people. After being in hospital in Adelaide for the past 3 months, we were off to Melbourne where we stayed for seven and a half months (3.5 of those in hospital, the rest daily outpatients visits).

Max was in hospital from September to January to have his bone marrow transplant. He spent a whole month of that in intensive care, because of his breathing complications and high flow oxygen requirements.

If Max had had the newborn screening test that included SCID, we would have been able to keep him isolated until he had his transplant. He would not have had the oxygen requirements. Often children who have bone marrow transplants leave hospital around day 30. Max left hospital on day 88 because of his breathing complications that he had prior to the transplant.

Genetic testing of Chris and I showed that I was unknowingly a carrier of SCID. Further testing on my mum showed that it started with me as a mutation.

Max has been diagnosed with Spastic Diplegia - Cerebral Palsy. An MRi scan shows that there was a bleed on his brain when he was very young, most likely when he was so sick and needed oxygen. Max is now four and a half years old. He can't stand independently. He requires a walker or sticks to walk. He is getting support from the NDIS which will be lifelong.

I know that adding SCID to the newborn screening test costs money. However NOT having it also costs money – a lot of money. Having SCID on the newborn screening test would save so much pain and heartache for babies and their families. It would give the baby a better chance of survival, not having complications before the tough process of a transplant.

We would have done things very differently to protect our precious baby if we had known he had no immunity to protect himself.