Tom's Battle Against SCID

This is Tom's story, written by Leah his mum as they learn of his devastating diagnosis of life threatening Severe Combined Immune Deficiency (SCID), the most severe form of Primary Immunodeficiency.

Three weeks was all it took to go from "he needs Panadol" to "he is fighting for his life". So how did it happen?

Thomas first got sick at around 10 months old. He had been sick before and since starting day-care it was becoming more common. A cold turned into ear infections, then came the conjunctivitis and nappy rash that just wouldn't clear from the constant diarrhea. This is all normal for a baby though, right?

He got "better", but he didn't return to his normal happy self. Doctors assured me he was healthy, so I put his fussiness down to teething. He continued to become worse day by day, slowly enough that it was hard to remember if he was better or worse than before. His hands suddenly struck me as odd looking, but maybe they were always chubby like that?

Our first visit to Emergency had the doctors worried, and a blood test soon showed he had rotavirus and RSV. They were both viral so unfortunately had to wait it out. After a week had passed he still wasn't well, worse in fact, so we made our second visit to Emergency. The results were the same, but the combination of Panadol and Nurofen had him laughing and flirting with nurses, so we were sent home.

Each day he still wasn't well and was spiking fevers of 40°C. He was on Panadol and Nurofen around the clock. Another doctor visit resulted in a third Emergency visit, this time at a different hospital. He was given more pain relief and had a brief moment of happiness. No tests, no admission, that was good enough.

The paediatrician explained to me that being a new mum is stressful and it's easy to make things bigger in my head than they really are. I was so embarrassed that I had wasted everyone's time and dragged my baby to yet another hospital. I started to think I was the problem.

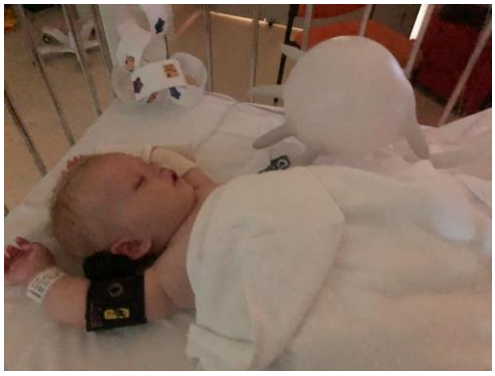
Another visit to the doctor and we were sent for an urgent paediatrician review. The paediatrician was confident that the problem was cow's milk protein intolerance. Tom was put onto formula to be reviewed in 2 weeks. So now I had an unhappy baby who would cry whenever he had a bottle, usually the one thing that would calm him.

By this stage Thomas had stopped crawling weeks earlier and was no longer able to sit unassisted. He didn't roll around in his cot while sleeping and screamed uncontrollably during nappy and clothes changes. He could no longer eat solid food or feed himself, instead dropping the food and giving up. My days consisted of sitting holding him. I couldn't walk around holding him - the movement caused him too much distress. It was clear he was in agony.

The final straw came when I sat him on the couch, propped up in the corner. There was no risk of him rolling off the couch. He couldn't move. He couldn't even hold his own head up, and it fell backwards onto a cushion I positioned under him. I had to leave him, so I could cry, and to call the paediatrician. This wasn't right. The paediatrician was on leave, and his covering doctor had little to offer.

Morgan got home that evening and took him for a bath to try and get a smile, but Thomas just sat staring into the distance. At that moment we both realised we were no longer watching our baby fight an illness - we were watching him lose. His eyes had gone distant and dark.

Terrified, we drove over an hour to the Holy Spirit Hospital to try and get someone to listen to us. It wasn't long before he had doctors and nurses swarming around him, getting the crash cart ready for the worst. After several cannulas were inserted and emergency IV was started he was admitted to Prince Charles Hospital next door.



The head of paediatrics was concerned and over the next few days they ran every test they could think of. We were told they still didn't know what was wrong but that it was rare and serious. After 4 days at Prince Charles, Thomas was transferred by ambulance to Lady Cilento Children's Hospital. His condition when he arrived was so poor that alarms were sounding, and 15-20 medical staff were running to stabilize him.

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It took 2 more weeks, a blood transfusion, 13 blood infusions, 3 procedures under general anaesthetic in theatre, MRIs, x-rays, a DNA test and countless blood tests to diagnose him with Severe Combined Immune Deficiency (SCID). There are less than 10 babies diagnosed in Australia each year. He had numerous infections in his body, his kidneys were swollen to the point of losing proteins which was causing fluid to leak into his body and make him swell. The immune cells that he did have (most likely maternal blood cells transferred during pregnancy) kept him alive but were also attacking his joints and giving him arthritis.

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Over the past couple of months Tom's doctors have worked to clear all the illnesses in Tom's body while he is kept in a confined special isolation room in the hospital. Visitors are controlled to limit the chance of infection. Physical contact is limited as the consequences of infections are very serious. Thankfully Tom has improved significantly.

Tom's parents are currently meeting with transplants doctors in hope of Tom receiving a bone marrow transplant before Christmas that will cure him from SCID.

National Newborn Screening for SCID would prevent the severe sickness, trauma and death experienced by kids like Tom prior to a SCID diagnosis and result in less organ and tissue damage from infection. Early detection and diagnosis through screening would enable earlier bone marrow transplantation curing this devastating rare disease in these precious babies.

The Immune Deficiencies Foundation (IDFA) is the peak patient body supporting those affected by primary and secondary immune deficiency disorders in Australia. IDFA strongly support the introduction of national newborn screening for SCID (www.idfa.org.au).