

SCID Member Stories – Isabelle Grant, QLD



Isabelle was 4 months old when she suddenly turned blue and stopped breathing. Fortunately, she was already waiting in the Emergency Department of the Children's Hospital, where doctors were able to immediately assist her and save her life. Isabelle's GP had seen her earlier that morning, suspected something wasn't right with her symptoms - so delayed her vaccinations (including the live Rotavirus vaccine) and sent her into Emergency for review.

The Hospital's General Pediatric doctor saw Isabelle the night of her admission, took a look at her very low White Blood Cell count and immediately called the Immunology Specialist. Miraculously, the General Pediatric doctor's son had done a school project on Severe Combined Immune Deficiency (SCID) the year before – so this doctor knew what the symptom's of the potentially fatal condition were, and that Queensland DID NOT screen its Newborn babies for SCID. Lots of blood tests were then run and Isabelle was immediately isolated from every other patient in hospital.

After officially being diagnosed with ADA-SCID, Isabelle spent over a month (including her first Christmas) in an isolated positive pressure room in the Intensive Care Unit, battling Pneumocystis Jiroveci Pneumonia (PJP). Everyone was astounded that after 4 months exposure to the outside World, this Bubble Baby had ONLY caught PJP and Rhinovirus with no infections. Isabelle was on around-the-clock oxygen and a high-flow machine to keep her breathing, with feeding tubes and one-to-one nurse monitoring.

It was a devastating time for Isabelle's family, with a huge impact on their everyday life.

Before the Christmas shutdown period, Immunologists urgently ordered a 'bridging treatment' from Germany – a tailor made Enzyme injection for ADA-SCID. This was the only way to stop the deoxyadenosine toxin from conditioning to spread through Isabelle's system, so she had a chance to get to the Stem Cell Transplant she desperately needed to survive.



After finally leaving ICU, Isabelle spent another month in hospital on a ward (again, in an isolated positive pressure room), fighting to regain enough strength to be able to survive the Transplant. During her third month in hospital, Isabelle underwent chemotherapy and her life-saving transplant, with her Dad as the donor.

Brain wave testing also revealed that the ADA toxin had damaged both Isabelle's inner ears after her birth – she was diagnosed with Permanent Severe Hearing Loss in both ears. She will require hearing aids and speech therapies for the rest of her life – at an unknown ongoing cost. This hearing loss was entirely preventable if she had been screened at birth and the SCID detected. Additionally, Isabelle missed many of her 4–7-month old development milestones while she was in hospital critically unwell.

Notably, genetic testing on Isabelle's parents revealed they DO NOT carry the ADA-SCID recessive genes. So Isabelle's SCID could not be foreseen and could only be diagnosed by screening her at Birth.

Don't play a defensive game – it's more cost-efficient to screen at birth and avoid complications and permanent damage, before getting a Bubble Baby with SCID to transplant. Particularly, with 'live' vaccinations on Qld's Immunisation Schedule.