

### SCID Member Stories – Isabelle Grant, QLD



Isabelle was 4 months old when she suddenly stopped breathing. Fortunately, she was in the Emergency Department and was seen by her GP who delayed her vaccinations (including LIVE Rotavirus vaccine).

As a result of this, Isabelle spent 33 days in an isolated positive pressure room in Pediatric ICU battling Pneumocystis Jiroveci Pneumonia (PJP) and Rhinovirus. The PICU Nurses indicated that the cost for an ICU bed in a pressurised room was around \$8,000 per day (estimated total \$264,000).

Isabelle required a bronchoscopy and PICC insertion (surgery), was on 24/7 oxygen and a high-flow machine to breathe, along with NG and TPT feeding tubes. The cost for these surgeries, medicines, and X-rays, is, yet unknown. Within days of being admitted, Isabelle's Immunology team diagnosed ADA-SCID. They were astounded that after 4 months exposure, she had only caught PJP and Rhinovirus.

Before the Christmas shutdown period, Immunologists urgently ordered a 'bridging treatment' from Germany called "RevCovi" – a tailor made Enzyme injection for ADA-SCID. This was the only way to stop the deoxyadenosine toxin from spreading through Isabelle's system, so she had a chance to get the transplant she needed. Each RevCovi vial cost \$11,000 and Isabelle required 10 doses (estimated total \$110,000 + \$4,000 shipping).

Isabelle spent a further 33 days in hospital on the Ward (again, in an isolated positive pressure room), fighting to get well enough to be able to survive a Stem Cell Transplant. The chemotherapy and transplant process took a further 28 days in hospital. It also required a CVL to be inserted (surgery).

Brain wave testing revealed that the toxin had attacked both Isabelle's inner ears from birth to 4 months old – she was diagnosed with Permanent Severe Sensorineural Hearing Loss in both ears. She also required grommets (surgery).



She will require hearing aids and speech therapies for the rest of her life – at an unknown ongoing cost to the NDIS system and Hearing Australia. Additionally, Isabelle missed many of her 4–7-month development milestones while in hospital. She now requires ongoing NDIS funded therapies to assist with her development.

Notably, genetic testing on Isabelle's parents revealed they DO NOT carry the ADA-SCID recessive genes – the mother carries a deletion. The father did not detect the gene at all – so either spontaneous mutation by Isabelle or the Mosaic Theory in the father.

Regardless, 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 before getting a SCID baby to transplant. Particularly, with 'live' vaccinations on Qld's Immunisation Schedule.