IDFA Immune Deficiencies Foundation Australia

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SCID Member Stories – Isabelle Grant, QLD

Isabelle was 4 months old when she stopped being able to breath properly and started turning purple around her lips and in her hands and feet. Fortunately, she was already waiting in the Emergency Department of the Children's Hospital, where doctors were able to immediately assist her. Isabelle's GP had seen her earlier that morning, suspected something wasn't right with her symptoms - so delayed her 4-month vaccinations and sent her into Emergency for review.

The Hospital's Paediatrician 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 Paediatrician had recently read a report on Severe Combined Immune Deficiency (SCID), and combined with her experience, she knew about this fatal condition, 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 pressure room in the Intensive Care Unit, battling Pneumonia. Everyone was astounded that after 4 months exposure to the outside World, this Bubble Baby had ONLY caught Pneumonia and Rhinovirus. 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. They went into lockdown in their home, not going anywhere except the hospital, in order to make sure no illnesses were picked up and carried back to Isabelle.

Immunologists urgently ordered a 'bridging treatment' from Germany – a tailor made Enzyme replacement injection for ADA-SCID. This was the only way to stop the deoxyadenosine toxin from continuing to spread through Isabelle's body, and give her a chance to survive until the Stem Cell Transplant she desperately needed to save her life. Isabelle also started monthly IV Immunoglobulin infusions.

After finally leaving ICU, Isabelle remained in hospital for months (still in an isolated positive pressure room), undergoing chemotherapy and then her life-saving Stem Cell Transplant, with her Dad as the donor. Fortunately, the transplant was successful and Isabelle was able to return home and live with her family in time for the Easter celebrations.

As a result of the ADA-SCID, Isabelle was also diagnosed with irreversible severe Hearing Loss. She was fitted with baby hearing aids and started speech therapies, while her family learned Auslan (Australian Sign Language).

Today, Isabelle is about to turn 2 years old – a milestone that she wouldn't have reached without the Stem Cell Transplant. She receives Subcutaneous Immunoglobulin (SCIg) infusions every week at home, to help support her new immune system. The SCIg infusion takes about 10 minutes to do on her leg, and she is so brave every time. Her parents are grateful to the generosity of every person who donates plasma - making this treatment possible for Isabelle. Her dad also donates plasma every 2 weeks.