Immune Deficiencies Foundation Australia

PO Box 742, Wollongong, NSW 2520
1800 100 198
info@idfa.org.au
ABN: 99 117 585 976

SCID Member Stories – Georgia Hilliar, QLD

Our baby, Georgia Hilliar, was born in Brisbane with undetected SCID – Omens syndrome, this year on the 12th January. Our journey to diagnosis was horrific. We nearly lost our newborn daughter on multiple occasions. Georgia had apneas that ended her in PICU at Queensland Children's Hospital. We endured weeks of heartache. In and out of hospital not knowing what was wrong. Georgia had no ability to fight off infections, so they turned life threatening.

Our newborn experienced severe infections that turned septic, a horrendous all over body inflammation and internal inflammation and simply failed to thrive. We were days away from getting the 'live' 6 week vaccinations, which could have been fatal for her. Our baby was finally diagnosed with this rare genetic disorder, after eight long weeks. She was kept in a positive pressure room to keep her safe from infections, before undergoing a bone marrow transplant at Queensland Children's Hospital, which saved her life.

We were told without treatment, infants with undetected SCID usually die from infections within the first year of life. For Georgia and many other SCID babies, it was a long hard process to be diagnosed. It takes for the baby to be in a very serious condition, before further testing is completed, and they get the help they need. Queensland's newborn screening test failed Georgia. The purpose of this simple Newborn Bloodspot Screening is to help to identify rare but serious conditions, and early treatment.

We are asking for an urgent review. SCID meets this criteria of being both rare and very serious. SCID needs to be included in the newborn screening check. With this simple blood test, doctors can check for this rare genetic condition that can cause serious health problems and untreated, death. Early treatment of infants with SCID means babies, like Georgia, could avoid infections leading to a higher survival rate – less chance of severe complications during curative treatment.

Georgia's condition was missed. It was missed at the physical examination that happens within the first few minutes after birth. It was missed at the full and detailed assessment that happens prior to discharge from hospital. It was missed at the follow up assessment performed in the first week by her thorough General Practitioner outside the hospital. Again, it was missed at the 6–8 weeks assessment after birth.

If SCID was on the newborn screening check, this awful journey could have easily been prevented for Georgia and our family and many other Queensland families. This long process to diagnosis significantly affected our family. My husband, Beau was unable to work, as we needed to juggle being bedside in an isolated room with Georgia and take care of our two young boys, Orlando (four years old) and Huxley (two years old). Our boys were impacted by not being able to continue their education and attend day-care or kindergarten to reduce the risk of virus and infection in our home.

This was an extremely stressful situation, we just managed to stay afloat and support our baby. We aren't quite out of the woods yet, but we are on the road to recovery. Please avoid future heartache for families, extended hospital stays and cost for the healthcare system, and add SCID on newborn screening check. The USA, many European countries and NZ know the importance for including SCID in their newborn screening. It's time we do this too.

Pre-symptomatic identification and treatment would improve survival for all infants born with SCID. With an early bone marrow transplant, frequent follow-up and prompt treatment for infections, survival rates are very good. It's time to add SCID to the Newborn Bloodspot Screening program in Queensland.