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SCID Member Stories – Javeria

Severe Combined Immunodeficiency. SCID. The bubble boy" disease. It's fatal if not picked up and treated in the first year of life. It's difficult to diagnose and is usually not suspected unless a child is seriously /continuously ill, at which point it may be too late for treatment, and it was not on the newborn screening list in Australia (It has started recently for NSW on a trial basis, and that is a game changer).

It took the life of our first son, Zakariya, at 15 months of age in 2014. He went undiagnosed for 13 months and by then he had received his 1-year vaccinations; they were essentially fatal in his condition. He never made it to his transplant date, which was also delayed due to the unavailability of a matched donor, despite a worldwide regist1y search for weeks.

It isolated our second son, Ismaeel, from birth until he was 2 years old; we converted our home into a bubble, kept him cooped up inside and away from other people to protect him from the risk of virus or illness. We washed hands and sanitised and sterilised everything. All. Day. Long. We isolated ourselves so we wouldn't bring any germs home. Our daughter didn't go to school or see other kids for a year, and I didn't leave the house, except for hospital visits. Below is a photo of what we hope and pray will provide a cure: a stem cell transplant, which is risky with a list of potential complications.

Bottom left is dad donating his cells (we didn't spend time searching for a donor this time. A haplo 50% matched transplant was riskier, but we had no other option). It may not seem like much, but in the top right picture, these 40mls of pink liquid comprise millions of stem cells and vital T cells, harvested from peripheral blood. The harvesting took 6 hours, and the cells were then processed by 5 specialists in the lab for 8 hours. Many millions of cells were discarded to prevent certain complications. The processed cells were given as an infusion into Ismaeel's central venous line the following day (top right in photo). He was 3 months old.

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It has been a long, challenging journey. We waited over 3 years to know if it was successful in giving him a complete, reconstituted immune system. Although we technically came out of isolation after 2 years, it was a long process getting comfortable with going out into the world. We were still cautious as we weren't sure where his immune system stands, we still avoided crowded places and young children.

We have learnt now that his immune system has not fully reconstituted-which means as of last winter he started monthly immunoglobulin infusions (IVIg) to boost his immune defence. He has not been able to get his vaccinations as he did not respond to the trial batch of vaccines. So we rely on herd immunity and the IVIg. He thankfully had his central venous line removed a couple of years ago, so he needs a cannula for each infusion, which is still difficult at his age.

We are so grateful to see him grow and thrive and feel very blessed watching him approach his 4th birthday. So thankful for all the blood and plasma donors that make his IVIg infusions possible, and we'd also love to raise awareness of the need for bone marrow donors-particularly from non-Caucasian backgrounds as they are severely underrepresented on the bone marrow donor registries. These donors are amazing-often anonymous, selfless strangers giving the most incredible gift that keeps on giving.

Ismaeel, aged 4.

Lastly, the realisation of having newborn screening for SCID in Australia is incredible, and I know a lot of hard work has gone into this over many years. Both of my sons' newborn screening blood sample cards were used to validate the trial tests for SCID newborn screening. If they helped in any way to turn newborn screening for SCID to a reality and that, in turn helps another child/ren be diagnosed early and treated in time-then they are already part of a legacy that is bigger than us.

In Loving Memory - Zakariya