Our story begins with Nolan. At age four he had the typical charm and energy of a young boy discovering life. He noticed small things and loved details. He had one older sister and two younger. We had no special concerns or premonitions about his health. In June 2017, following a one-month battle in PICU, Nolan succumbed to severe complications of chicken pox. A tsunami of grief and questions tore at our hearts.

As time passed, we found peace with the fact that some things would never be understood on this side of heaven. Our family grew as we welcomed a new little son and a fourth daughter. Five and one-half years after Nolan's death, Jemima began her life under NICU observation. Her oxygen saturation eventually stabilized and we were quite ready to settle in at home. Two days later, our relief was shaken by a call from Newborn Screening. Jemima was suspected of having a serious, genetic immune disorder. We were urged to seek immediate care and keep her away from crowds. We realized that we had most likely been given the missing puzzle piece concerning our oldest son.

With MSUD in our extended families, we knew of The Clinic for Special Children and their work with genetic disorders. We called the Clinic that day and couldn't have wished for a better response. Morning found us in an exam room at the Clinic with two of the team. Dr. Strauss spoke easily but thoughtfully. The Clinic would do what they could to keep our baby safe.

Jemima was diagnosed with a rare *RAG2* gene mutation. This can cause low T-cell and B-cell counts, leaving them especially vulnerable to viral infections, which in some cases can be fatal. The Clinic was able to go back and confirm *RAG2* was the cause of Nolan's illness. While this put a piece in the puzzle of Nolan's death, it wasn't reassuring.

We kept Jemima in isolation at home while we learned what we could about her condition. Testing for her siblings revealed that five-year-old Owen also had the same genetic change. A question hung foremost and heavy on our minds. Would these children be okay?

As the Clinic's first *RAG2* family, the path ahead was hazy, at best. Very little data was available worldwide for this unique gene misspelling. However, we had confidence in what the Clinic was doing for us. Both children's immune systems were under evaluation through extensive blood work. The Clinic kept us informed as they reached out to other experts in the field of healthcare and immunology.

In collaboration with the Children's Hospital of Philadelphia, we were presented with a recommendation for Bone Marrow Transplant. Owen and Jemima's incompetent immune systems could be replaced through a careful process using donor stem cells. Treatment carried certain risk and would be a huge commitment. It included months of isolation during the patient's recovery. 'I had fainted, unless I had believed to see the goodness of the Lord in the land of the living. Psalms 27:13'

Jemima's immune system was weaker than her brother's. In June 2023, at six months old she underwent a transplant at Children's Hospital of Philadelphia. At this point, we had not yet reached a decision for Owen. He appeared to be the picture of health. We knew firsthand though, how quickly that could change. After Jemima was through the most crucial phase of treatment, we were more ready to focus on the concerns for Owen. The doctors would support us either way, but they had concluded that his immune system could not be trusted to serve him well. It was time for a leap in faith.

Both children have come through transplants and are doing well. What a blessing! The Clinic has followed their progress through regular communication with CHOP. We are grateful that the Clinic for Special Children is willing to partner with the specialists for follow-up care as the opportunity presents.

We deeply appreciate the Clinic's compassionate support during an intense time for our family. We are aware that there was a significant amount of time, research, and dedication poured into our situation. Thank you for helping to make the Clinic possible!

The Martin Family