

Ava's Story



Ava is our precious gift from God. At exactly 38 weeks of being pregnant, I went in for a routine check-up. After doing the ultrasound, they noticed that the placenta was not looking so healthy anymore. The decision was made to send us to the hospital to start the delivery process. With each contraction Ava's heart rate would drop. Immediately it was determined that I would need a C-section. Thank God we made the decision when we did because the placenta was starting to separate from the uterine wall, which could have resulted in Ava being a stillborn.

Ava was healthy when she was born and nothing appeared to be wrong; little did we know that our lives would change so drastically. The night before we were going to be discharged, the Doctor took Ava to do an ultrasound scan of her head because they were concerned it was too small. When they brought her back to me, I noticed a large area of skin hanging on the back of her head. I immediately called the Doctor back, and they took her away from me without even an explanation. They came back and told us first thing in the morning she would be transferred to St. Peter's University Hospital but still with no explanation as to what was going on. Our hearts were so heavy, the thought of coming home with a happy healthy baby were a distant hope now.

Upon arriving at St. Peter's, Ava had numerous tests done in an effort to arrive at a diagnosis. The Doctor was fairly certain that she had Epidermolysis Bullosa (EB). She stayed in the hospital for 3 weeks. During

that time, both our family and the hospital were attempting to learn how to care for a child with this condition. Due to the rarity of the condition, no one could really shed light on how to help Ava, which was very unnerving, and resulted in more open wounds. Finally we were able to take our little girl home. We did not receive a diagnosis until 2 months after being discharged. The result was Junctional Epidermolysis Bullosa. JEB is caused by a genetic anomaly in the body's binding protein – a protein called Laminin. Laminin is what bonds together our organs, muscles, and skin. Ava was born without this protein and because of this her skin blisters very easily. Blistering can be caused by a number of different factors clothing, everyday handling, routine diaper changes, or simply being in one position too long. Each blister is equivalent to a 3rd degree burn, and she has to wear protective bandages over her arms, legs, hands, feet, and torso. The process can take up to 3 hours a day to complete. Currently there is no cure for this condition. Recently we found out that Ava has two gene mutations neither of them have ever been recorded. So we are really traveling in uncharted territory.

We are believing in God for a miracle. In spite of everything, Ava is such a happy baby. In eight months she has taught us to appreciate the precious gift that life truly is, to love our family and friends more than ever, and allowed us to see God guiding us and continually holding our family together.

