



Dustyn's Story

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Dustyn is the 3rd child to Katie and her husband, and though an unexpected pregnancy, they were thrilled. Her pregnancy was complicated towards the end, but she remained focused on doing all that was needed to ensure that Dustyn made a safe and healthy entrance to the world.

An ultrasound late in her pregnancy revealed that Dustyn and a very large cyst on his basil ganglia, which could be a sign of Down Syndrome. These cysts are known to go down, and upon obtaining a second opinion four weeks later, they received the great news that Dustyn's had gone down. At this time, they thought they had cleared their largest hurdle. Little did they know, the journey had just begun.

Finally, August 2nd, 2010 arrived. Katie had been plagued by feelings of anxiety and concern over Dustyn's health, and her and her husband were both excited and nervous for Dustyn to finally arrive. During labor and delivery, Dustyn's heart rate was erratic, which led to greater anxiety for Katie. A feeling of relief washed over her when she finally heard him cry and was able to hold him for the first time.

Katie almost immediately had concerns over Dustyn's health. He refused to eat, and though the doctors kept saying it was normal, it did not feel normal to Katie. She stayed positive, and fed him as much as she could around the clock in attempt to get him to eat, and eat enough.

She then noticed a blue tint around his eyes and mouth and immediately called the nurse. He was having breathing issues that the doctors called "dusky blue spells", but they offered no explanation as to why he was having them. As quickly as they began, they stopped happening and the cause was never determined. He also began eating, but became jaundiced, which required that he stay in the hospital a few more days under the lights. Finally, five days after he was born, Dustyn was ready to go home with his family until news arrived that would dramatically change their lives.



The doctor came into the room, and informed Katie and her husband that he was sorry to bring them the news, but Dustyn had tested positive on his newborn screening test for Glutaric Aciduria Type 1, also known as GA-1. Katie and her husband were in a state of disbelief, which so many questions swirling in their minds.

Dustyn was still discharged, but an appointment was made with a genetic doctor immediately. The doctor explained to them that GA-1 is a genetic disorder, caused by gene mutations that prevent him from properly breaking down Lysine and Tryptophan, two amino acids found in protein. All the information Katie was receiving was overwhelming, but she felt confident and dedicated to doing anything and everything Dustyn required.

The first four months went very well for Dustyn. He was put on a special diet, and had weekly blood draws and checks. Dustyn reached his milestones as expected, smiling and laughing and Katie's anxiety was finally easing as she enjoyed the new addition to her family.

Doctors discovered that Dustyn had an inguinal hernia. A very simple operation, but due to the GA-1 and the fasting

required before anesthesia, Dustyn would have to come in the day before the surgery to receive lipids and a dextrose I.V. to help him stay stable for both the anesthesia and the surgery.

While waiting for Dustyn during the surgery, Katie began to realize that it was taking far longer than expected. She became anxious and nervous, and when the surgeon finally emerged and took her to a side room to talk, she was terrified. Due to his underlying condition, his heart beat became far too low, and the surgeon had feared they were going to lose him. A quick acting nurse started a sodium IV along with the others he was already receiving, and he bounced back, his heart rate normal again. They required Dustyn to stay an extra day for observation.

Since the surgery, Dustyn has been in and out of the hospital for illness precaution. The local hospital was not experienced enough with children, resulting in Dustyn having to be transferred by ambulance to a children's hospital. The children's hospital was a significant commute, so Katie and her husband decided to move to be closer to the children's hospital for Dustyn.

This decision proved immensely helpful for them. They have a wonderful metabolic doctor, and the children's hospital is exceptional. Since their move, Dustyn has had a few issues with illness and some feeding issues, but nothing too major.

Despite things going well, Katie and her husband began to notice little things happening to Dustyn. It became obvious that Dustyn was not meeting his milestones as expected, for example he was unable to sit unassisted until he was 11 months old. He would say his first words, only for them never to be heard again. He finally began crawling at age one year, and now, at 17 months, he has taken four wobbly first steps. He has been unable to progress from baby foods to thicker foods and now experiences uncontrollable movements (tremors) which make it hard for him when he attempts to hold a spoon and feed himself.



Baffled by these issues, despite him seemingly doing so well on blood tests, Katie and her husband sat down with his metabolic doctor to discuss the cause of these issues. She explained that even with controlled diet and appropriate care when sick, a percentage of children still have effects from the disease. It's unknown why this happens, but she indicated that Dustyn's low tone would result in gross motor skills being delayed. His speech is delayed, and he also may have sleep apnea caused by a floppy airway. His issues with food are being caused by a sensory disorder. The tremors have caused concern there may be some damage in his brain, and he is being monitored closely for this issue.

Despite all of the challenges Dustyn has faced, he is an incredibly happy-go-lucky little boy. He is so strong, that there are times Katie feels he's stronger than her and her husband! He is sweet, caring and comforts Katie through his beautiful smile, delightful giggles and his adorable baby talk. He loves to snuggle and hug, and he adores his big brothers.

After all the stress, fear and anxiety, Katie is grateful and feels that Dustyn is here for a reason. He has taught his family that life is to be cherished. He has taught them to appreciate the little things in life. He has taught them that you never give up; he never stops trying. Their family has a new definition of the word love, and have become so much closer.

Katie is an active and supportive member of BBAF, offering advice, a kind word, or anything else another BBAF mom might need to hear. It is her strong belief that Newborn Screening gave Dustyn a chance at life and she is thrilled for the opportunity to share his story with the world, not only for awareness, but to remind us all to never lose hope. The smiles and giggles make it all worth it in the end.