



Angel Foundation

Rebekah's Story

Written By: Angel Mom, Laura

Pamela and her husband Ken are very young parents to two wonderful children, and are just like most families in many ways. The one thing that makes them different from other families in their small town is something that others are not aware of until the family shares about it. Their daughter, Rebekah, who is now five years old, was diagnosed around 3 weeks of age with a very rare Fatty-Acid Oxidation Disorder called Medium Chain Acyl-CoA Dehydrogenase Deficiency. (MCADD).



Children with MCADD have an enzyme that is needed for breaking down medium chain fatty acids that is either missing or not functioning properly. Rebekah's body cannot break down and store the fat needed for fasting for long periods of time or during illness.

Rebekah was born on November 19, 2005 after an uneventful pregnancy. She weighed 6 lbs 2.7 oz and seemed very healthy. After having some time to hold her, a nurse mentioned they would need to take her to the nursery to do a little test that was really "no big deal" and that it would just require a few heel pricks. Since Pamela's niece was born just prior to Rebekah, she was somewhat familiar with what she had heard referred to as the PKU test, so she didn't give it much thought.



After a few days in the hospital, the new family went home, and everything continued wonderfully. Rebekah took well to nursing, and was sleeping well too. The family had no worries and were just enjoying life and watching their beautiful little girl grow.

After several days at home they received a phone call from their pediatrician. He stated that Rebekah needed to come into his office the next day, and that it was a very important matter. He reminded them of the Newborn Screening test that Rebekah had received in the hospital, and stated she had screened positive for a rare disorder called MCADD. He said he was not at all familiar with the disorder, and the only things he knew was that Rebekah could not go for more than 2-4 hours without eating. They would need to learn about MCADD together. Pamela immediately burst into tears at the end of the conversation. Her daughter possibly had a disorder that would change her life and that of her family forever. She was understandably terrified and afraid to even let Rebekah sleep for fear that something could happen.

During the visit with the pediatrician, the family was able to get some more information about MCADD. They further discussed Rebekah not being able to fast for long periods of time, and were able to get an appointment with genetics at the local Children's hospital.

The geneticists provided even more answers and information to the family. Further tests were run and confirmed Rebekah's diagnosis. Rebekah was placed on carnitine and the family was given her treatment plan and emergency protocol. Pamela became a bit more comfortable and began to feel confident she would be able to provide Rebekah with the care she needed to live a full and happy life.

Rebekah has only been hospitalized twice for her MCADD for precautionary reasons while she had the flu or a virus. She is a very healthy child and very active. She loves to sing, dance and she plays sports. She recently started preschool and Pamela has found that to be one of the hardest things to deal with because she has to trust others with Rebekah's care.

Presently Rebekah is not allowed more than 30% fat intake in her daily diet, so her family has to carefully watch what she eats and make special arrangements for birthdays and special holidays where different foods are offered and available.

Rebekah has gotten used to taking her carnitine everyday, twice a day, peeling icing off cake at birthday parties, and only being allowed chicken nuggets if they have a special treat out at a fast food restaurant.

Her family is grateful for how well Rebekah has done, and that things have been relatively easy so far given her diagnosis. But just as any parents of a child with a medical condition, it is always in the back of their minds that an illness at any time could change all of their lives forever. Rebekah being ill, or around others who are ill, leaves the family on understandably on edge. They are constantly watching to ensure that she is eating, and eating foods with the appropriate amount of fat.

The family stays strong despite the fear and hardships and takes solace in knowing that no matter how Rebekah's story began, or how it changes in the future, her story will ultimately change another child's life. Comprehensive Newborn Screening saved Rebekah's life, as well of that of her entire family. Without it they would not be where they are today. It is the family's deepest hope that Rebekah's story can help another family find comfort in knowing that no matter how scary it seems, they are not alone. There are others out there who are sharing the same experiences. They are part of a very small family with big hearts who will always be there to support them and who fully understands the everyday stresses that come along with having a child with such a rare disorder.

