



Newborn Screening  
Saves Babies  
One Foot  
at a Time

## Lilly

THIS IS A PERSONAL STORY SHARED BY A FAMILY WHOSE CHILD HAS  
MEDIUM-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY (MCADD)

We lost a child, Thomas, to MCADD in March of 2006. Although he was comprehensively screened at birth, we did not receive the results of NBS in time to save him. He died a few hours after leaving the hospital and we received word two days after his death that he had screened positive for MCADD.

My husband and I decided that we should have another child, not to replace Thomas or even take away a portion of the pain. We thought it would bring more happiness into our lives and give our 6 year old added joy of getting to experience being a big brother.



Although we chose not to proceed with any prenatal testing, we took all other necessary precautions with our baby. On November 27, 2007, Lilly was born. Because of our history with MCADD and because of very caring health professionals, Lilly was placed in NICU while we awaited test results. She was on IV carnitine, glucose and was given formula every 2 hours. On Friday, November 30, NICU doctors notified us that Lilly too had tested positive for MCADD. My original gut feeling was that this baby would not have the defect. It was quite a blow to our family; however, we realized this was treatable and knowledge is powerful.

Lilly is doing well now. She is 8 months old and is meeting all of her milestones. She has had more than her share of hospital stays in her short little life. When she was 12 days old, she was not taking enough formula, fighting her bottles, taking quite a bit of time to complete a feeding, etc. After about 8 hours of this routine, I called genetic specialist and he told me to head to the local ER and meet my regular pediatrician there and put the emergency protocol he had written for us into place. IT WORKED. We were very pleased at how well the doctors, staff, and ICU handled the protocol even though most of them had never heard of MCADD. The reason Lilly could not take her bottles correctly was because of trapped gas. Yes, trapped gas (it even showed up on the chest x-ray) prevents babies from getting enough in-take and can put them in trouble fairly quickly if you don't get them the help they need. At 3 ½ months, Lilly went back to the hospital.

This time for a twelve day stay because of poor feedings. Doctors did not find anything that would prevent her from eating. We put her on a different schedule, added cereal to her diet, and were discharged after she adapted to those changes. Two weeks later, Lilly refused to eat at one of her scheduled feedings, so we went back to our local hospital and stayed four days. Lilly was transferred to our state's only Children's Hospital. It was determined there, after extensive procedures, that she had some ulcerated areas in the stomach and was

experiencing some problems due to acid reflux. Lilly was discharged with an NG tube and us having the knowledge on how to put the tube in and how to use it. In May, Lilly had to go back to the local hospital and stay a week because of an upper respiratory infection. That's four hospitalizations in a six month period.

Lilly now has a NG feeding tube so that she can get her needed calories each day and we can feed her through the tube when she just can take all of her bottle. We know there will be more hospitalizations but we are happy to have the knowledge needed to take care of her. We are very happy that many health care professionals have received the proper education to provide medical attention to Lilly. We know that is why Lilly was sent to us with MCADD so she could make a difference for someone else.

Written and submitted August 3, 2008 by James & Melanie Shannon:

Parents of Lilly (MCADD), born November 27, 2007

Thomas (MCADD 3/14/06-3/16/06)

Tucker (Unaffected – 6 years old)