



Newborn Screening
Saves Babies
One Foot
at a Time

Alli

THIS IS A PERSONAL STORY SHARED BY A FAMILY WHOSE CHILD HAS
GLUTARIC ACIDEMIA TYPE I (GA-I)

Our family has been very fortunate and I thank God every day. I have no idea where we would be today without Pediatrix (formerly known as Neo Gen Screening) and their newborn screening test. During the last year my stepmother was fighting the battle of leukemia and my husband and I decided to save our babies cord blood for future use for her or my husband or children later down the road. So we researched and decided to go with cord blood registry. We were very thankful that the newborn screening test was included in our cord blood kit. We had never heard of the new "tandem mass spectrometry" (MS/MS) testing.



My pregnancy and labor went very well. Alli came 2 days early with a big smile and is the perfect baby. I never thought about anything going wrong especially since our first child was healthy. Since the expanded newborn screening packet was free of charge we decided let's do it, why not? Northside Hospital would not do the test for us, they said they were only allowed to do the PKU test. So on our first visit to the pediatrician, one week old, they pricked Alli's heel and we mailed off her test. Not ever expecting to hear anything back. But within two days we had the results. She had tested positive for Glutaric Acidemia Type 1. Glutaric Acidemia Type 1 is an inherited disorder of amino acid metabolism. Which means our child cannot use the amino acids lysine and tryptophan in a normal way. A special diet is the only treatment. Our pediatrician had no knowledge of this genetic disorder so he referred us to Dr. Fernoff at Emory University. They immediately ordered a blood sample and a urine sample. Both test came back positive for Glutaric Acidemia Type 1. So next they took a skin sample to verify that the test results where not false positives. Dr. Singh (our nutritionist) started Alli on a special diet immediately; Alli was 3 1/2 weeks old. Leaving the doctors office in tears and not having a cure for your child is an unexplainable feeling. My husband and I started looking on the internet to research and to find out as much information as we could about Glutaric Acidemia Type 1, which I think made us even more worried about Alli's future.

Looking at Alli you would never know there was any type of problem, she acted and looked just like any other 31/2 week old baby. Not knowing how Alli's body is going to react under certain illnesses and not having the knowledge of how to help our Alli under this crisis is very frightening. After waiting about 3 months for the skin results, we had positive result for Glutaric Acidemia Type 1.

Alli is now 27 months old. She is so adorable. She smiles and laughs all the time. She weighs about 25 lbs. and about 31 inches tall. My husband and I are a lot more at ease. We have a speech therapist and physical therapist that help Alli and she is doing wonderful. We have talked to several doctors who have GA1 patients that were diagnosed by the expanded newborn screening test. They have excellent results and their patients are living a normal life. Our child's life depends on the special formulas and special protein-free foods available. We

have complete confidence with Dr. Singh at Emory University to help monitor Alli's diet. We have an emergency plan available if needed. Without the proper nutrition Alli would not have a normal life. We pray every night that Alli will have a normal life.

Thank you for taking the time to read about our daughter. If we can be of any assistance to you or someone you know please give us a call (770-449-8668) or email us at graysonup@comcast.net. If you have any information about GA1 please share with us. We are always interested in learning as much as we can. Thank you.

Sincerely,
Donna Upchurch
Updated August 4, 2004 by Donna Upchurch
Mother of Alli (GA1), born April 29, 2002