

OUR BEST FOOT FORWARD

A Newborn Screening Success Story: A Mom's Perspective

When our son was a few weeks old we received a phone call from our pediatrician's office stating that his newborn screening came back abnormal. We were shocked since he was our third child and we have no known genetic abnormalities in our family. The pediatrician's office told us Jonah possibly has a mild form of VLCAD deficiency or is a carrier of VLCADD and they recommended that we contact Greenwood Genetic Center for further details. I remember writing down "VLCADD" on a piece of paper and having no clue what this meant. I called Greenwood Genetic Center to set up an appointment as soon as we could. Our genetic counselor was very helpful and reassuring in answering our initial questions.

During our first appointment we had several more questions; the genetic counselor and Dr. Champaigne were so graciously patient and informative. We decided to have our son's blood work done to recheck his biochemical findings. And we also had mine and my husband's DNA tested for VLCADD, a condition that renders the body unable to break down certain fats. We left our first appointment still not knowing for sure if our son had a mild form or was a carrier of VLCADD. Once the results were back, we found out that there was only one genetic mutation from one of the parents and his biochemical

markers came back lower than when he was first born. Our genetic counselor told us that the blood work is trending towards our son only being a carrier of VLCADD.

We're so thankful for the newborn screening and the wonderful team at Greenwood Genetic Center. VLCADD can be life threatening and we are grateful to learn more on how we can help our son. We're also very appreciative for the state of South Carolina being so invested and providing funding for families with metabolic disorders.

About VLCAD: Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency is a condition that prevents the body from converting certain fats to energy, particularly during periods without food (fasting). VLCADD affects 1 in 40,000 to 120,000 individuals.

Signs and symptoms:

- low blood sugar (hypoglycemia)
- lack of energy (lethargy)
- muscle weakness
- liver abnormalities
- life-threatening heart problems.

Children with VLCADD often need dietary supplements. Medium Chain Triglyceride (MCT) oil is a common supplement for individuals with VLCADD. This oil contains medium chain fatty acids, which are fats that the baby's body can break down. The patient's doctor might also prescribe L-carnitine supplements. L-carnitine is a substance that is naturally produced by the body, but the patient's body might not

make enough. Taking prescription L-carnitine supplements can help break down fats for energy and remove harmful substances in the body.

Sources:

<https://ghr.nlm.nih.gov/condition/very-long-chain-acyl-coa-dehydrogenase-deficiency>

<http://www.babysfirsttest.org/newborn-screening/conditions/very-long-chain-acyl-coa-dehydrogenase-deficiency>

ON THE RUN

Prior to sending a specimen to the DHEC lab please **RUN** away from these practices!

- Writing in Pencil
- Placing specimens in a plastic bag
- Photo copying specimens
- Using White Out

Please **RUN** toward these practices!

- Check expiration date of filter paper
- Fill in all the demographic areas
- Fill in primary care physician that will assume responsibility for infant after discharge
- Obtain a good blood spot

Are you in need of newborn screening filter paper forms?

- Contact Lab Supply at (803) 896-0913

NOTE: Please do not delay mailing your specimens to the DHEC lab. There is someone to receive specimens daily regardless of lab closings.

If your office or hospital doesn't collect the repeat newborn screen please schedule a collection at your local health department:

Low Country Region: (803) 759-3000

Pee Dee Region: (843) 673-6562

Midlands Region: (803) 635-6481

Upstate Region: Ask for the lab department.

- Anderson: (864) 260-5541
- Greenville: (864) 282-4100
- Spartanburg: (864) 596-2227



ON THE SPOT

The hospitals listed below had 0% unsatisfactory specimens for the 4th quarter:

- Georgetown Memorial Hospital
- Kershaw Health

CONTACT US. WE'RE HERE TO HELP!

DHEC Newborn Screening Follow Up: (803) 898-0593/(803) 898-1969

DHEC Lab: (803) 896-0891

Keep us on our toes. Please give us feedback on what you would like to see in our next Footnotes Edition. Email newbornscreening@dhec.sc.gov with your suggestions.