

### OUR BEST FOOT FORWARD

#### What to expect as South Carolina begins testing for Krabbe Diseases:

In May 2023, South Carolina joined the list of other states that currently screen for Krabbe Disease as part of their Newborn Screening (NBS) panel. The 10 other states screening for Krabbe Disease include NY, MO, KY, OH, TN, IL, IN, NJ, PA, and GA.

Krabbe Disease (KD), also known as globoid cell leukodystrophy, is a progressive neurodegenerative condition caused by the loss of myelin, the protective sheath around axons. The severity and age of onset varies widely. It is initially caused by a decrease in the level of an enzyme called galactocerebrosidase (GALC). Without enough GALC, individuals with Krabbe disease cannot break down psychosine, a naturally occurring toxin found in brain cells. Psychosine builds up and damages the brain's myelin. Although there is no cure, there is treatment available called hematopoietic (or blood) stem cell transplant (HSCT). KD has a prevalence of approximately 1 in 100,000. Two forms of KD have been described: infantile (IKD) and late onset. IKD is the most severe and comprises at least 85% of known cases. Patients typically present with symptoms including irritability, muscle weakness, feeding difficulties, fever of unknown origin, abnormal posturing and/or developmental delays within the first year of life.

#### What terms should you be familiar with?

- **Leukodystrophy** ("luke-o-dis-tro-fee"): a group of inherited genetic diseases that damage the myelin, or white matter, of the brain.
- **Psychosine** ("sigh-co-seen"): a toxic substance that builds up in individuals with Krabbe disease. Psychosine collects in the brain and spinal cord and damages myelin.
- **Myelin** ("my-len"): also known as white matter, forms a protective coating around nerves and helps them quickly carry information from one part of the brain and spinal cord to another.
- **Other names for Krabbe Disease** ("Crab-Ay"): Krabbe Leukodystrophy, Globoid Cell Leukodystrophy, GALC Deficiency, Krabbe is a type of Lysosomal Storage Disorder (LSD) and Leukodystrophy.

#### What will an outside acceptable limit screening result look like on the lab report?

First tier screening will be performed at the South Carolina (SC) Public Health Laboratory (PHL) using a quantitative test for  $\beta$ -galactocerebrosidase (**GALC**), the enzyme that is deficient in newborns affected with Krabbe disease.

The screening assessment will be based on the daily median of GALC results. The expected range will be "**>15% of the daily median**". Results will be reported as "**Within Acceptable Limits**" or "**Outside Acceptable Limits.**" Additionally, GALC screening results that are reported as "**Outside Acceptable Limits**" ( $\leq 15\%$  of the daily median) will include the calculated percentage of the daily median. Initial specimens reported as, "**Outside Acceptable Limits**" will be sent to a reference lab for 2<sup>nd</sup> tier **psychosine** testing.

#### What is the notification process for an abnormal result?

The SC Department of Health and Environmental Control's (DHEC) Maternal and Child Health (MCH) NBS staff will provide notification of **abnormal psychosine results** to the primary care provider and/or physician indicated on the NBS specimen collection form by phone, fax, and/or mail.

#### What are the next steps that should be taken?

When you receive an abnormal GALC result on the newborn screen, you should **WAIT** for the **psychosine** result prior to taking the next steps and providing clinical guidance.

If the psychosine is within acceptable limits, provide genetic education and counseling. No further blood spots are needed.

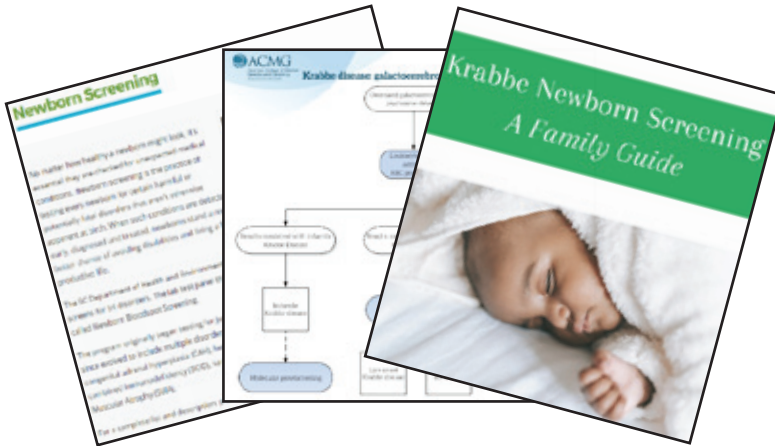
If the **psychosine** is abnormal, you should take the following **IMMEDIATE** actions:

- If **Psychosine** is **> 1 nmol/L**, consult with and refer to a pediatric metabolic specialist. A list of metabolic specialists will be provided with the notification documentation.
- If **Psychosine** is  **$\geq 10$  nmol/L**, along with a referral to a pediatric metabolic specialist, consult with and refer to a pediatric transplant specialist the **SAME DAY**.
- All abnormal psychosine results will be further reflexed for GALC molecular sequencing to confirm the diagnosis.
- Contact the family and inform them of the newborn screening results.

- Evaluate the newborn with particular attention to neurologic dysfunction.
- Provide the family with basic information about Krabbe disease.
- Report all clinical outcomes to the newborn screening program.

**Are there resources available? Yes! See links below:**

DHEC Newborn Screening [Website](#), ACMG Newborn Screening ACT [Sheets](#), and the Hunter's Hope [website](#).



Sources: (1) ACMG ACT Sheets and Algorithms [Internet]. Bethesda (MD): American College of Medical Genetics and Genomics; 2001-. Authors. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK55829> (2) Krabbe newborn screening - lysosomal storage disorders. Hunter's Hope. (2022, November 3). <https://www.huntershope.org/newborn-screening/krabbe-newborn-screening/>



**Would you like to be a part of our Newborn Screening team?**

We are growing and currently have job opportunities in the Newborn Screening Follow-up Program, First Sound (EHDI hearing screening) Program, and the Public Health Laboratory!

**Please check the DHEC careers [website](#) for all newborn screening job postings!!**

**WELCOME TO THE NEW STAFF MEMBERS ADDED TO THE SC NEWBORN SCREENING TEAM:**

- **Hailey Selander,BS:** laboratory Technologist II.

**ON THE RUN**

**What's NEW in 2023?**

We have 3 new disorders being added to the SC NBS Test panel in 2023:

**MAY 2023**

- Krabbe: [huntershope.org/newborn-screening/nbs-family-guide/](https://huntershope.org/newborn-screening/nbs-family-guide/)

**FALL 2023**

- Argininemia: [babysfirsttest.org/newborn-screening/conditions/argininemia](https://babysfirsttest.org/newborn-screening/conditions/argininemia)
- X-Linked Adrenoleukodystrophy (X-ALD): [babysfirsttest.org/newborn-screening/conditions/adrenoleukodystrophy](https://babysfirsttest.org/newborn-screening/conditions/adrenoleukodystrophy)

**Vital Records Matching Project:**

Did you know the DHEC Public Health Laboratory and Maternal and Child Health Bureau have partnered with the vital records department to locate infants without newborn screening specimens? This project began last year and has been a collaborative effort between these areas to match newborn screening demographics with birth certificate data. The findings for 2022 identified 306 newborns with birth certificates that were not matched to a newborn screening test or refusal form. Of the 306 newborns, 168 were born in South Carolina hospitals and 138 were home births. Babies born in hospitals were not matched often due to name and address changes, religious exemptions, out of state hospital transfers, and infant mortality. The 138 home births were babies born through midwifery groups, home birth centers, and unexpected home deliveries. Based upon these findings, the South Carolina Newborn Screening Team is targeting outreach efforts to ensure that a newborn screening specimen is obtained for every baby born in the state of South Carolina. It's the law!

## What are some things you can do to help:

1. Ensure parents are educated about newborn screening via the NBS brochure and website.
2. Ensure the information filled out on the newborn screening card matches the information filled out on the birth certificate.
3. If a parent/guardian religiously objects to the newborn blood spot screen, they must fill out and sign DHEC form 1804 and submit it to DHEC.

## Parental Refusal by Religious Objection:

Do you know what to do if a parent/legal guardian refuses to allow you to collect a newborn screen?

4. Complete the Religious Objection Form: DHEC 1804, Newborn Screening Program, Parental Statement of Religious Objection:  
**\*\*Form can be found here:** <https://scdhec.gov/sites/default/files/media/document/Newborn-Screening-Manual-Appendix-20220428.pdf>
5. Include the parent's demographic information to assist the DHEC NBS program with parent notification of the refusal.
6. Fax the refusal form to DHEC NBS program at (F) 803-898-0337, email to [newbornscreening@dhec.sc.gov](mailto:newbornscreening@dhec.sc.gov) or mail to: **DHEC Newborn Screening Program 2100 Bull St. Columbia, SC 29201**

## The 7-Spot Card

In April, SC NBS changed to a new 7-spot NBS collection card. By minimizing text on the cards and reorganizing the position of the pre-printed circles, the SC NBS Lab was able to add two additional pre-printed circles to the cards without compromising the size of the circles. An example of the new 7-spot card is pictured below. The NBS Lab has started sending these out with all new requests.

The addition of two new spots on the collection card will allow for additional blood, which is needed as the number of tests increases over time.

The new 7-spot card is also an effective tool in decreasing the number of unsatisfactory specimens received at the lab. The mission is to reduce the number of cards submitted without enough blood (quantity insufficient or QNS), which is the primary type of unsatisfactory specimen received. In 2022, 4.6% of newborn screening specimens submitted were unsatisfactory, with 36.5% of those being QNS. The 5-spot cards can continue to be used until they expire, or you run out, whichever comes first.

**APPENDIX A: Religious Objection Form: DHEC 1804, Newborn Screening Program, Parental Statement of Religious Objection**

I am the parent or legal guardian of \_\_\_\_\_, a child born \_\_\_\_\_ in South Carolina. I request that my child not be tested by blood spot screening in order to detect silent, deadly metabolic diseases and hemoglobinopathies. I certify that this refusal is based on religious grounds. Religious grounds are the only permitted reason for refusal under South Carolina law, Section 44-37-30 (C).

I understand that my child may suffer brain damage, other bodily harms, or death if a disease that can be detected by blood spot screening is not diagnosed. I understand that such harm can be lessened or prevented by early diagnosis and treatment. I understand that these diseases are usually silent and may be present in a child that looks healthy.

I understand that the blood spot screening test is the best way to detect these disorders early, and that testing is routinely done for every child. I understand that this testing is quick, easy and that the results are confidential. I understand that this testing has been the standard of care for all children born in South Carolina and the rest of the United States for many years.

I have been fully informed of, and fully understand, the possible devastating consequences to my child's health if blood spot screening is not done. I have been fully informed of, and fully understand the benefits of testing and blood specimen storage. I have been given the brochure produced by the South Carolina Department of Health and Environmental Control that describes the conditions for which testing is currently available and explains the benefits of testing and blood specimen storage.

I also understand that my child would have been tested for these conditions except for my objection. I have been given the opportunity to ask questions concerning this testing and these conditions, and all of my questions have been fully answered to my satisfaction.

I release and hold harmless the South Carolina Department of Health and Environmental Control, the hospital or other facility at which the birth occurred, the person(s) responsible for the collection of the blood spots, and any other person or entity relying on this objection, for any injury, illness and/or consequences, including the death of my child, which may result to my child as the result of my refusal of blood spot screening.

Parent: \_\_\_\_\_ Date: \_\_\_\_\_

Witness: \_\_\_\_\_

**NOTE TO PROVIDERS:** This form is only necessary if the parent or legal guardian refuses testing for inborn metabolic errors and hemoglobinopathies.

*\*For refusal questions please see: Neonatal Screening for Inborn Metabolic Errors and Hemoglobinopathies Sections 44-37-30: section A of the South Carolina Code of Laws: add a link to the law [here](#)*

## ON THE SPOT

The birthing facilities listed below were the top 5 performers who achieved the lowest average percentage of unsatisfactory newborn screening specimens for 2022:



Hospital Name	2022 Unsatisfactory Percentage (%)
Prisma Health: Oconee Memorial Hospital	0.18
Newberry County Memorial Hospital	0.51
Beaufort Memorial Hospital	0.57
MUSC- Orangeburg	0.77
Saint Francis Eastside	1.23

The birthing facilities below were the most improved in 2022 for NBS specimen collection to received turn-around time (C-R TAT):

Hospital Name	Annual average for collection-to-received turn-around-time (TAT)*
Lexington Medical Center	0.75 day
Hilton Head Hospital	0.92 day
Carolina Pines Regional Medical Center	0.96 day
McLeod Health Dillon	0.96 day
MUSC - Orangeburg	1 day

## Newborn Screening Diagnosed Cases

To date, the following hospitals collected newborn screens *that led to these confirmed diagnosed cases in 2022:*

### Congenital Adrenal Hyperplasia

McLeod Regional Medical Center  
MUSC Shawn Jenkins Children's Hospital

### Congenital Hypothyroidism

AnMed Health  
Beaufort Memorial Hospital  
Colleton Medical Center  
East Cooper Medical Center  
Grand Strand Medical Center  
Lexington Medical Center  
McLeod Health Dillon  
McLeod Health Medical Center  
MUSC Health Lancaster Medical Center  
MUSC Shawn Jenkins  
Prisma Health Baptist  
Prisma Health Greenville  
Prisma Health Laurens  
Prisma Health Patewood  
Prisma Health Richland  
Prisma Health Tuomey Hospital  
Saint Francis Eastside  
Self Regional Healthcare  
Spartanburg Medical Center  
Summerville Medical Center

### Cystic Fibrosis

AnMed Health  
Lexington Medical Center  
McLeod Health Loris  
Prisma Health Baptist  
Prisma Health Greenville

Prisma Health Patewood  
Prisma Health Richland  
Prisma Health Tuomey Hospital  
Spartanburg Regional

### Hemoglobin Disorders

Aiken Regional Medical Center  
AnMed Health  
Bon Secours St. Francis Hospital: Xavier  
Carolina Pines Regional Med Center  
CHS: Pineville  
Coastal Carolina Hospital  
Colleton Medical Center  
East Cooper Medical  
Kershaw Health  
Lexington Medical Center  
McLeod Health  
McLeod Health Clarendon  
McLeod Health Dillon  
McLeod Health Medical Center  
MUSC Health Florence  
MUSC Shawn Jenkins  
Prisma Health Baptist  
Prisma Health Baptist Parkridge  
Prisma Health Greenville  
Prisma Health Laurens  
Prisma Health Richland  
Prisma Health Tuomey  
Piedmont Medical Center  
Regional Medical Center  
Self Regional Healthcare  
Spartanburg Regional Medical Center

Summerville Medical Center  
Tidelands Waccamaw

### Lysosomal Storage Disorders

- **Pompe**  
Spartanburg Regional Medical Center  
Lexington Medical Center

- **MPS1**  
Prisma Health Richland

### Metabolic Disorders

- **Biotinidase Deficiency**  
Coastal Carolina

- **PKU**  
Prisma Health Patewood  
MUSC Shawn Jenkins Children's Hospital

- **MCAD**  
Lexington Medical Center

- **3-MCC**  
Prisma Health Richland

- **GA-1**  
Prisma Health Patewood

- **Galactosemia**  
Prisma Health Richland

### Spinal Muscular Atrophy (SMA)

Prisma Health Richland  
Spartanburg Regional Medical Center

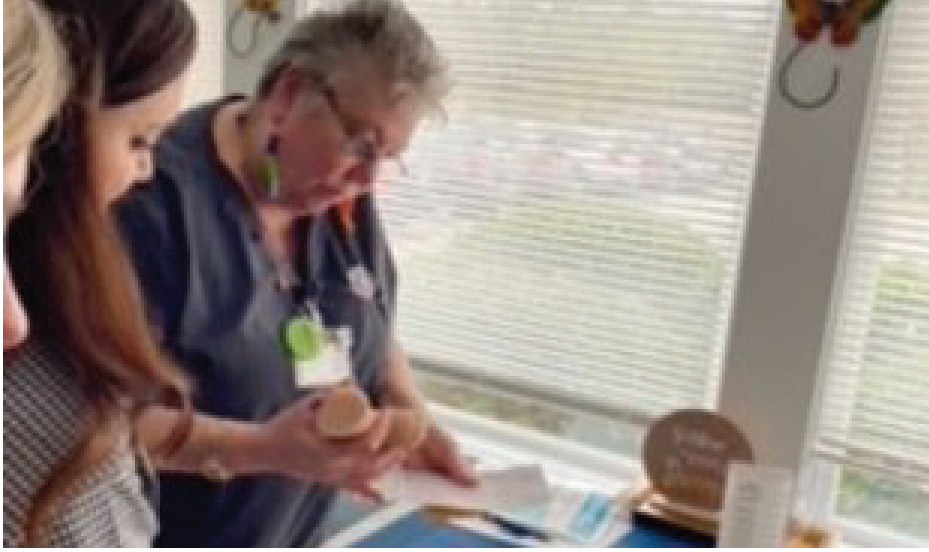
### SCID/T-Cell Lymphopenia

MUSC Health Lancaster



## FIRST TIME EVERY TIME!

Would you like for your staff to be trained on how to collect an adequate newborn screen? Please email [harrelcl@dhec.sc.gov](mailto:harrelcl@dhec.sc.gov) to be added to the 2023 NBS training schedule.



### Certificate of Achievement

This certificate acknowledges that

\_\_\_\_\_ has completed the

### DHEC Newborn Screening Training

Presented on the \_\_\_\_\_ Day of \_\_\_\_\_ in the Year \_\_\_\_\_



\_\_\_\_\_ Date \_\_\_\_\_

CR-01338 4/23

**WE ENJOY HELPING OUR PARTNERS PUT THEIR BEST FOOT FORWARD!**

**CONTACT US. WE'RE HERE TO HELP!**

**DHEC Newborn Screening Program:**

(803) 898-3192

**Newborn Screening 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](mailto:newbornscreening@dhec.sc.gov) with your suggestions.

## EDUCATIONAL INFORMATION:

**Are you educating parents about Newborn Screening?**

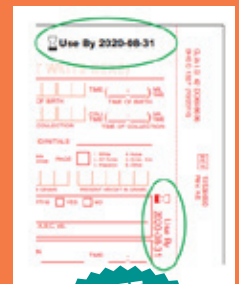
Visit our website at [scdhec.gov/health-professionals/lab-certification-services/newborn-screening](http://scdhec.gov/health-professionals/lab-certification-services/newborn-screening) to find our *updated* newborn screening brochure and educational handouts for parents and providers.

**Are you in need of NBS brochures (ML-000032, Eng. and ML-025096, Spa.)?**

Please go to: [scdhec.gov/about-dhec/educational-materials-library-product-order-site](http://scdhec.gov/about-dhec/educational-materials-library-product-order-site).

**Are you in need of NBS collection forms?**

You can now email [PHL-supply@dhec.sc.gov](mailto:PHL-supply@dhec.sc.gov) to receive collection forms for your office or facility or call 803-896-0913.



**NOTE:**  
The next batch of filter paper expires on 09-30-2024!