

OUR BEST FOOT FORWARD

The South Carolina Newborn Screening (NBS) Program began screening for Pompe disease on February 22, 2021. Pompe disease is a Lysosomal Storage Disorder (LSD) caused by a defect in the acid alpha-glucosidase (GAA) gene. This defect results in glycogen accumulation, primarily in cardiac and skeletal muscle. There are three forms of Pompe disease: classic infantile onset, non-classic infantile onset, and late onset. Each form differs in disease severity and age of onset. The incidence of this disorder varies among different ethnic groups and affects about 1 in 40,000 in the United States.

Since the program began screening, there have been 6 infants identified with a form of Pompe disease. One of the infants identified by the SC NBS program is Nora. Here is her story from her mom's perspective:

NORA'S STORY:

"Our beautiful daughter, Nora, was born on August 28th, 2021, just 6 months after the state of South Carolina added Pompe to the newborn screening panel. I didn't know much about newborn screening, and I certainly had never heard of Pompe Disease, but that all changed when Nora was just 5 days old. We received a call that Nora's newborn screening results had been flagged as abnormal for Pompe Disease. A few days later, confirmation testing verified what newborn screening had indicated: Nora had Infantile Onset Pompe Disease. Words cannot begin to describe the shock, anxiety, and devastation we felt when we received those results. We were first-time parents who felt like our world was crumbling around us.

"Newborn Screening gave us our best possible outcome and for that we are forever thankful." – Nora's Mom



However, we would soon learn that receiving those results was not the end of our world, but instead the start of our daughter's opportunity to thrive. We are grateful for newborn screening in so many ways, for so many reasons. Because of this screening: we didn't have to watch our daughter's health decline and waste time trying to figure out the cause. Nora received an accurate and swift diagnosis. She began life-saving treatment at just 2-weeks-old and these treatments have allowed Nora to grow as strong and healthy as any normal child. Because of newborn screening, our daughter is thriving.



"Receiving those results was not the end of our world, but instead the start of our daughter's opportunity to thrive" – Nora's Mom

Looking at Nora, you would never know that she has a rare disease. She is a happy 8-month-old who has recently learned to crawl and is always on the move. As I watch my daughter meet her milestones and her personality blossom, it is always in the back of my mind what our reality could have been, had Pompe not been added to the newborn screening panel just 6 months before our daughter was born. We don't know the 'could haves' and 'maybes' of that situation, but we do know that newborn screening gave us our best possible outcome and for that, we are forever thankful!"

References:

acmg.net/PDFLibrary/Pompe

<https://medlineplus.gov/genetics/condition/pompe-disease/#frequency>

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

- **Mary Thomas** joined the SC NBS lab team in December of 2021 as a Laboratory Technologist II. She is a certified Medical Laboratory Technician with 12 years of laboratory experience.
- **Nicole James** joined the SC NBS lab team in February 2022 as a Laboratory Technologist II. She is a certified Medical Laboratory Technician with 3 years of laboratory experience.
- **Christine Harrelson, MS**, joined the SC NBS Follow up Program in March 2022 as a NBS Coordinator. She is a certified Medical Laboratory Scientist with 5 years of experience.
- **Marcelina Jackson** joined the SC NBS Follow up Program in April 2022 as a Metabolic Nutritionist. She is a Registered Dietitian with 12 years of experience.
- **Melissa Reynolds** joined the NBS Lab team in April 2022 as a Laboratory Technologist II. She has a Bachelor of Science degree in Biology with 1 year of laboratory experience.
- **Shelby Gifford** joined the NBS Lab team in May 2022 as a Laboratory Technologist II. She has a Bachelor of Science in Biochemistry with 1 year of laboratory experience.
- **Ashley Stewart** joined the NBS QI team in May 2022 as a Laboratory Technologist III. She has a Bachelor of Science in Biology with 1 year of laboratory experience.
- **Shatoya Wright** joined the NBS follow up Program in July 2022. She has an MBA with an undergraduate degree in Biology and 7 years of experience.

ON THE RUN

The SC DHEC Public Health Laboratory (PHL) and Maternal & Child Health Newborn Screening Program are working towards implementing screening for four additional disorders. Screening for Spinal Muscular Atrophy (SMA), X-Linked Adrenoleukodystrophy (X-ALD) and Argininemia are expected to begin later this year. Screening for Krabbe Disease is projected for 2023.

In April 2022, the South Carolina Newborn Screening Program and Public Health Laboratory began a partnership with the Bureau of Vital Statistics, located within DHEC's Bureau of Community Health Services. The objective of this partnership is to ensure that each SC newborn has received the required newborn screen by matching newborn vital records with newborn blood-spot screening records. This new process can allow newborns who missed screening to be tracked and provided with appropriate screening tests, diagnosis, and treatment/management in time to prevent catastrophic health outcomes if screened conditions exist. It can also provide validation of critical demographic information in both records as a quality check.

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?

1. Complete the Religious Objection Form: DHEC 1804, Newborn Screening Program, Parental Statement of Religious Objection:

****Can be found here:** <https://scdhec.gov/sites/default/files/media/document/Newborn-Screening-Manual-Appendix-20220428.pdf>

2. Include the parent's demographic information to assist the DHEC NBS program with parent notification of the refusal.

3. Fax the refusal form to DHEC NBS program at (F) 803-898-0337, email to newbornscreening@dhec.sc.gov or mail to: **DHEC Newborn Screening Program 2100 Bull St. Columbia, SC 29201**

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 harm, 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: _____

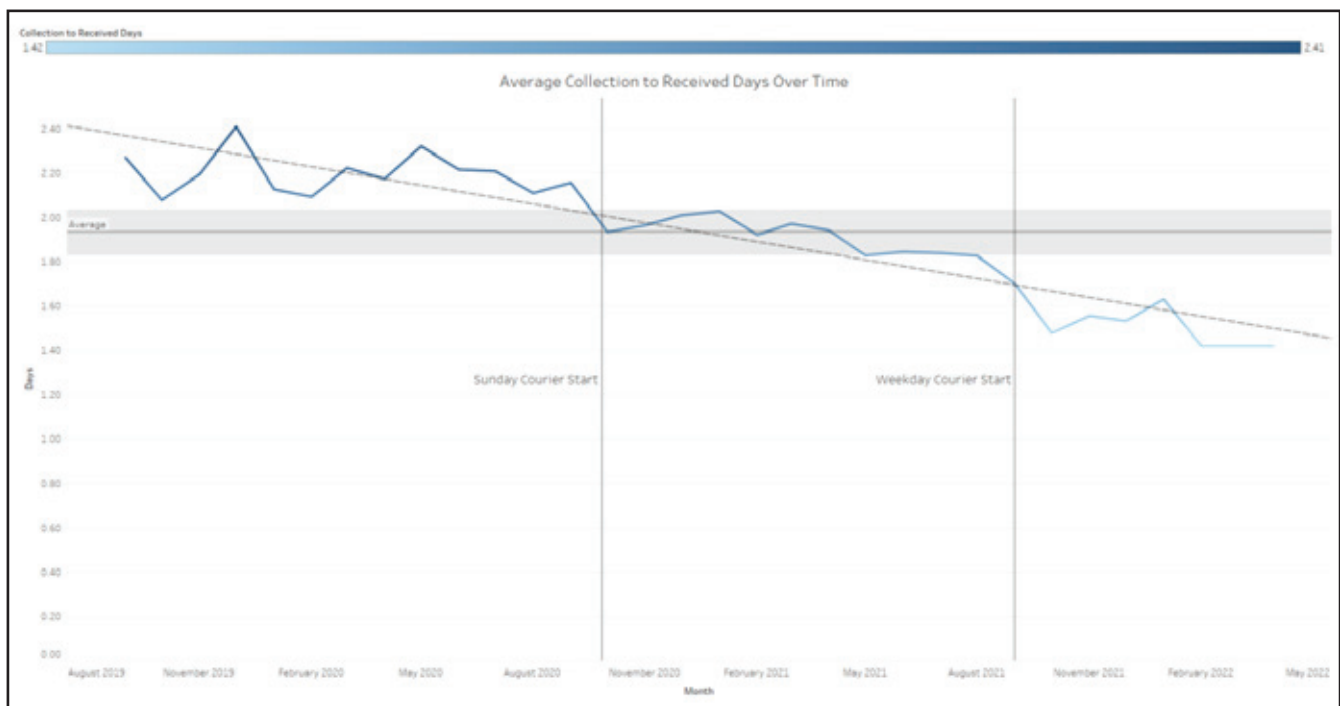
NOTE TO PROVIDERS: This form is only necessary if the parent or legal guardian refuses testing for inborn metabolic errors and hemoglobinopathies.

Courier QI Update

The SC NBS Laboratory implemented a Sunday courier service on September 27, 2020, and implemented a weekday courier service on September 16, 2021. Both services run in the evenings and pick up NBS specimens after 9:00 pm and deliver specimens to the Public Health Laboratory before 7:00 am the following day. Currently, 36 of 38 SC birthing hospitals are utilizing the Sunday courier service while 37 of 38 SC birthing hospitals are utilizing the weekday courier service.

In the year following the implementation of the Sunday courier service, 90% of SC birthing hospitals saw at least a 5% improvement in turn-around-time and hospitals, on average, saw a 12% decrease in turn-around-time compared to the previous year! Since the implementation of the weekday courier service, 93% of hospitals have seen at least a 5% decrease in turn-around-time and hospitals, on average, are seeing a 24% decrease in turn-around-time compared to last year!

The graph below shows the average collection to received days since August 2019 through May 2022. The two vertical lines show when the Sunday courier was implemented in September 2020 and when the new weekday courier was implemented in September 2021. As shown in the graph, there has been an obvious improvement in the number of days it takes for newborn screening specimens collected to be received by the SC PHL.



The SC NBS Laboratory Will Be Piloting the New 7 Spot Card

The SC NBS Laboratory will be piloting 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 circle. An example of the new 7-spot card is included below. The NBS Lab is hoping to pilot these new cards in the upcoming months.

The pilot will determine if the new 7-spot card is an effective tool in decreasing the number of unsatisfactory specimens received at the lab. The NBS program is particularly interested in specimens that are submitted without enough blood (quantity insufficient or QNS), which is the primary type of unsatisfactory specimen received. In 2021, 4.5% of initial specimens submitted were unsatisfactory. Of these specimens, 37.5% were QNS.

Part 3: 338 White CF
Part 4: 338 Black CF
Part 5: 338 Other CF
Part 6: 338 Unknown CF
Part 7: 338 No Race Specified CF
Part 8: 338 No Sex Specified CF
Part 9: 338 No Ethnicity Specified CF
Part 10: 338 No Race, Sex, or Ethnicity Specified CF
Part 11: 338 No Race, Sex, or Ethnicity Specified CF
Part 12: 338 No Race, Sex, or Ethnicity Specified CF
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Part 100: 338 No Race, Sex, or Ethnicity Specified CF

TAKE A LOOK AT A NEW RESOURCE TO ASSIST PEDIATRIC OFFICES WITH NBS PROCESSES:

Newborn Screening Success For Pediatric Offices



Provider responsibility:

- ◊ Per SCDHEC Regulation 61-80, Section E, the healthcare provider listed on the newborn screening (NBS) card is legally liable for handling the patient's NBS results.
- ◊ This includes conveying all abnormal, normal, and unsatisfactory results for patients who have not yet made appointments with the office.

Recollection tips:

- ◊ Recollect the newborn screen whenever results are abnormal, unsatisfactory, or inconclusive as soon as possible.
- ◊ Do NOT recollect a newborn screen for abnormal hemoglobin or CF DNA results.
- ◊ If the patient only had an abnormal TSH or 17-OHP result, collect a serum sample for lab confirmation. Send a copy of those results and any diagnostics to the program.

Specimen Shipping:

- ◊ NBS specimens should be shipped ASAP using the fastest method, like FedEx.
- ◊ Specimens must dry 3-4 hours before shipping. Do NOT batch ship; send specimens daily.

Updates:

- ◊ Normal lab reports will no longer be routinely faxed.
- ◊ e-Reports is a web portal to access all NBS results.
- ◊ To gain access, contact NBSLab@dhec.sc.gov.
- ◊ Note, NBS reports over six months old will not be in e-Reports.

Scan code to see the Newborn Screening Manual and protocol for patients discharged from the NICU.



Remember:

- ◊ Confirm every new patient had a newborn screening specimen collected.
- ◊ Review results before the two week appointment.
- ◊ Be sure to inform parents of all normal and abnormal results.
- ◊ Collect a subsequent specimen if needed.

Scan code below for tips on effective communication of results to parents and caretakers provided by HRSA.



Health Department contacts if your office needs help collecting NBS specimens:

- Pee Dee MCH Program Manager — (843) 623-9823
- Low Country MCH Program Manager — (843) 953-0274
- Midlands MCH Program Manager — (803) 745-5060
- Upstate MCH Program Manager — (864) 898-5984

ON THE SPOT

The birthing facilities listed below are the top 5 performers who achieved the lowest average percentage of unsatisfactory newborn screening specimens for 2021:

- Regional Medical Center: **0.35%**
- Beaufort Memorial Hospital: **0.94%**
- MUSC Health Kershaw Medical Center: **1.1%**
- Prisma Health Oconee Memorial Hospital: **1.34%**
- Aiken Regional Medical Center: **1.78%**



The birthing facilities below were the most improved in 2021 for NBS specimen collection to received turn-around time (C-R TAT)

Hospital Name	Average C-R TAT		TAT Percent change
	2020-2021	2021-2022	
MUSC Health Lancaster Medical Center	3.08	1.38	-55.2%
Carolina Pines Regional Medical Center	2.50	1.21	-51.6%
McLeod Health Clarendon	2.79	1.38	-50.5%
McLeod Health Loris	2.33	1.25	-46.4%
Prisma Health: Laurens County Memorial Hospital	2.04	1.13	-44.6%

Thank you for being champions for dried blood spot collection in your facilities!

Newborn Screening Diagnosed Cases

To date, the following hospitals collected newborn screens that had confirmed diagnosed cases in 2021:

Amino Acid Disorders

▪ PKU

Conway Medical Center
Prisma Health Patewood
Spartanburg Medical Center
St. Francis Eastside

Acylcarnitine Disorders

▪ Isovaleric Acidemia

Prisma Health Greenville

▪ MCADD

Lexington Medical Center
AnMed Health

Primary Systemic Carnitine Deficiency (CUD)

Self Regional Healthcare

3-MCC

Conway Medical Center

Congenital Adrenal Hyperplasia

East Cooper Medical Center
McLeod Health Medical Center
Prisma Health Tuomey
Spartanburg Medical Center

Congenital Hypothyroidism

AnMed Health
Colleton Medical Center
Conway Medical Center
East Cooper Medical Center
Grand Strand Medical Center
Lexington Medical Center
McLeod Health Dillon
McLeod Health Medical Center
MUSC Health Florence Medical Center
MUSC Health Lancaster
MUSC Shawn Jenkins Children's Hospital
Piedmont Medical Center
Prisma Health Baptist
Prisma Health Greenville
Prisma Health Patewood
Prisma Health Richland
Prisma Health Tuomey
Regional Medical Center
Roper St. Francis Berkeley
Self Regional Healthcare
Spartanburg Medical Center
St. Francis Eastside

Cystic Fibrosis

Lexington Medical Center
McLeod Health Medical Center
MUSC Shawn Jenkins Children's Hospital
Spartanburg Medical Center
Summerville Medical Center

Galactosemia

Prisma Health Baptist

Hemoglobin Disorders

Aiken Regional Medical Center
Beaufort Memorial Hospital
Bon Secours St. Francis Xavier Hospital
Carolina Pines Regional Medical Center
Coastal Carolina Hospital
East Cooper Medical Center
Grand Strand Medical Center
Lexington Medical Center
McLeod Health Dillon
MUSC Health Florence Medical Center
MUSC Health Lancaster
MUSC Shawn Jenkins Children's Hospital
Newberry County Memorial Hospital
Piedmont Medical Center
Prisma Health Baptist
Prisma Health Greenville
Prisma Health Patewood
Prisma Health Richland
Prisma Health Tuomey
Regional Medical Center
Self Regional Healthcare
Spartanburg Medical Center
Summerville Medical Center

Pompe Disease

McLeod Health Medical Center
McLeod Health Loris
Prisma Health Greenville

SCID/T-Cell Lymphopenia

Grand Strand Medical Center
McLeod Health Medical Center
MUSC Shawn Jenkins Children's Hospital
Prisma Health Baptist Parkridge
Prisma Health Patewood
Prisma Health Richland

NBS COLLECTION TRAINING OPPORTUNITIES:

Please join us virtually this Summer and Fall for "Dried Blood Spot Collection" trainings.

Save the Date based on your regional area. Scan the QR code below to register:



Midlands:

Tuesday, July 12, 2022, 12-1pm

Upstate:

Monday, August 15, 2022, 12-1pm

Pee Dee:

Tuesday, September 13, 2022, 12-1pm

Low Country:

Tuesday, October 11, 2022, 12-1pm

EDUCATIONAL INFORMATION:

Are you educating parents about Newborn Screening?

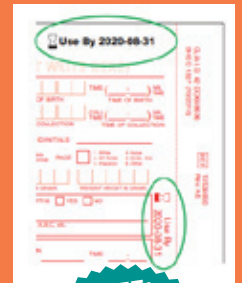
Visit our website at scdhec.gov/health-professionals/lab-certification-services/newborn-screening to find our *updated* newborn screening brochure and educational handouts for parents and providers.

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.

Are you in need of NBS collection forms?

You can now email 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!

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 with your suggestions.