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.

“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
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:


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

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.
**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.

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**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.
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.

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.

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):

<table>
<thead>
<tr>
<th>Hospital Name</th>
<th>Average C-R TAT</th>
<th>TAT Percent change</th>
</tr>
</thead>
<tbody>
<tr>
<td>MUSC Health Lancaster Medical Center</td>
<td>3.08</td>
<td>1.38</td>
</tr>
<tr>
<td>Carolina Pines Regional Medical Center</td>
<td>2.50</td>
<td>1.21</td>
</tr>
<tr>
<td>McLeod Health Clarendon</td>
<td>2.79</td>
<td>1.38</td>
</tr>
<tr>
<td>McLeod Health Loris</td>
<td>2.33</td>
<td>1.25</td>
</tr>
<tr>
<td>Prisma Health: Laurens County Memorial Hospital</td>
<td>2.04</td>
<td>1.13</td>
</tr>
</tbody>
</table>

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 Patwood  
  Spartanburg Medical Center  
  St. Francis Eastside
- **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
- **Conway Medical Center**

**Congenital Adrenal Hyperplasia**
- East Cooper Medical Center  
  McLeod Health Medical Center  
  Prisma Health Tuomey  
  Spartanburg 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  
  Newberry County Memorial Hospital  
  Piedmont Medical Center  
  Prisma Health Baptist  
  Prisma Health Greenville  
  Prisma Health Patwood  
  Prisma Health Richland  
  Prisma Health Tuomey  
  Regional Medical Center  
  Self Regional Healthcare  
  Spartanburg Medical Center  
  Summerville Medical Center
- **Piedmont Medical Center**
- **Prisma Health Baptist**
- **Prisma Health Greenville**
- **Prisma Health Patwood**
- **Prisma Health Richland**
- **Prisma Health Tuomey**
- **Regional Medical Center**
- **Self Regional Healthcare**
- **Spartanburg Medical Center**
- **Summerville Medical Center**

**Cystic Fibrosis**
- Lexington Medical Center  
  McLeod Health Medical Center  
  MUSC Shawn Jenkins Children's Hospital  
  Spartanburg Medical Center  
  Summerville Medical Center
- **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 Patwood  
  Prisma Health Richland  
  Prisma Health Tuomey  
  Regional Medical Center  
  Self Regional Healthcare  
  Spartanburg Medical Center  
  Summerville Medical Center
- **Piedmont Medical Center**
- **Prisma Health Baptist**
- **Prisma Health Greenville**
- **Prisma Health Patwood**
- **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
- **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 Patwood  
  Prisma Health Richland
- **Grand Strand Medical Center**
- **McLeod Health Medical Center**
- **MUSC Shawn Jenkins Children’s Hospital**
- **Prisma Health Baptist Parkridge**
- **Prisma Health Patwood**
- **Prisma Health Richland**

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**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

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**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.