

### OUR BEST FOOT FORWARD

#### September is National Newborn Screening Awareness Month!!!

Did you know that most states follow the Recommended Uniform Screening Panel (RUSP) when adding new conditions to their panel? South Carolina is currently screening for 31 of the 34 recommended conditions with plans to add 3 new conditions in the near future.

The committee that works to set national guidelines is called the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC). The committee meets regularly to discuss proposals from parent advocates, organizations and medical experts in order to keep newborn screening up to date. In addition, the secretary of the U.S. Department of Health and Human Services reviews the committee's recommendations. The committee and the Secretary work together to create the RUSP. The RUSP is a list of conditions, including 34 core conditions that the committee recommends every baby should be screened for. The RUSP recommendation is not a law or mandate, but it serves as a helpful guide for the states. Visit the committee's website at [www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html](http://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html) to learn more.

Source: [www.babysfirsttest.org/newborn-screening/the-recommended-uniform-screening-panel](http://www.babysfirsttest.org/newborn-screening/the-recommended-uniform-screening-panel)

#### ON THE SPOT

The hospitals listed below had 0% unsatisfactory specimens for the first half of 2018:

- Georgetown Memorial Hospital
- McLeod Health Cheraw
- McLeod Health Clarendon



#### NBS Collection Training Opportunity:

We have had great participation for 2018 newborn screening dried blood spot collection training!

**Upstate:** 24 participants

**Public Health Lab:** 22 participants

**Pee Dee:** 27 participants

There are **two** more opportunities in 2018 to attend a training workshop. We hope you are able to participate.

**Midlands:** August 17, 2018

**Lowcountry:** November 08, 2018

Please email [nbscollectiontraining@dhec.sc.gov](mailto:nbscollectiontraining@dhec.sc.gov) to register for an upcoming training.

#### Updates in Screening for Cystic Fibrosis:

##### Disorder Summary:

The SC newborn screening laboratory screens dried blood spots to identify newborns with an increased risk for Cystic Fibrosis (CF). Cystic Fibrosis is an inherited disorder affecting the pulmonary, digestive and reproductive systems. CF causes an excessive production of abnormally thick and sticky mucus. If left untreated, CF can lead to malnutrition, hypoxia and premature death. Due to advancements in newborn screening, public health laboratories are able to perform screening measuring immunoreactive trypsinogen (IRT), the primary screening marker for CF. When an abnormal or elevated IRT value is yielded, a repeat specimen is requested to allow for a second analysis of the IRT level. This two-step testing is referred to as the IRT/IRT protocol. These patients are referred to their nearest Cystic Fibrosis Foundation-accredited care center to pursue further diagnostic testing.

##### Updated Lab Method:

In an attempt to decrease the number of false positive CF screening results, the laboratory is bringing on a qualitative genotyping (DNA analysis) test to detect a panel of approximately 60 mutations and variants in the CF transmembrane conductance regulator (CFTR) gene. The panel represents the most frequently identified mutations found in CF patients. This will enable the laboratory to transition from an IRT/IRT protocol to an IRT/DNA protocol. In the IRT/DNA protocol, an initial specimen that yields an abnormally high IRT result will be reflexed for DNA analysis on the initial specimen, thereby reducing the need for repeat specimens for CF screening.

**Next Steps:** Validation of the IRT/DNA protocol in the laboratory has been completed. A recorded webinar will be held on August 15 to discuss the test and change in testing algorithm. Administrative requirements must be completed to add the new protocol into the NBS laboratory and follow-up workflows. More information for the newborn screening healthcare community and news of the start of the pilot study for preliminary screening for CF by the IRT/DNA protocol is forthcoming. We anticipate the pilot study to start the first week of September.



## ON THE RUN



### How do I fill out the demographic information on the filter paper when an infant is being adopted?

The baby's name field on the newborn screening filter paper form should reflect the infant's legal name at the time of birth or adopted name (if known). If the infant's name is unknown at the time of collection, enter the infant's name as mother's last name, first name "Adoption." (Example: "Smith, Adoption")

If the birth mother is not to be contacted, please enter the lawyer's first and last name or adoption agency's name in the mother's name fields as the legal guardian. In the street address field, enter the lawyer or adoption agency's address as the legal guardian contact. Also, enter the phone number of the lawyer or adoption agency in lieu of the birth mother's phone number.

When known, always enter the physician's demographics and infant's medical record number in entirety. This will assist the Newborn Screening Program in locating an infant if there is a need to alert the lawyer/legal guardian or adoption agency of an abnormal or potentially life-threatening result and ensure the infant gets the medical attention she or he needs.

### Sickle Cell Results for NCAA

September is Sickle Cell Awareness Month. If you are in need of a patient's sickle cell results for NCAA requirements please fax a request including the patient's name, date of birth and mother's name at the time of the patient's birth to 803-896-0889 or call 803-896-0795.

### Lab Closings for the remainder of 2018:

- **Monday, Sept. 3, 2018:** Labor Day
- **Tuesday, Dec. 25, 2018:** Christmas
- **Saturday, Nov. 10, 2018:** Veterans Day Observance
- **Wednesday, Dec. 26, 2018:** Christmas
- **Thursday, Nov. 22, 2018:** Thanksgiving
- **Tuesday, Jan. 1, 2019:** New Year's
- **Friday, Nov. 23, 2018:** Thanksgiving

## EDUCATIONAL INFORMATION:

Are you educating your parents about Newborn Screening? Visit our website at [www.scdhec.gov/newbornmetabolicscreening](http://www.scdhec.gov/newbornmetabolicscreening) to find our newborn screening brochure and educational handouts for parents and providers.



### Are you in need of NBS brochures?

Please go to [www.scdhec.gov/Agency/EML/place\\_order.aspx](http://www.scdhec.gov/Agency/EML/place_order.aspx). Using the ML number search, you can order the brochure in English (ML-000032) or Spanish (ML-025096).

### Meeting announcement: Updates to the DHEC regulation for newborn screening

There will be a stakeholder meeting on August 22, 2018 at 11:30 a.m. in the Mills Jarrett Building, Room N200 (2100 Bull St, Columbia, SC 29201) to discuss proposed updates to South Carolina Code of Regulation 61-80 for newborn screening. For more information, contact Dr. Brent Dixon at [dixonrb@dhec.sc.gov](mailto:dixonrb@dhec.sc.gov).

To view the South Carolina Code of Regulation 61-80 for newborn screening please visit <https://www.scstatehouse.gov/coderegs/Chapter%2061-80%20through%2061-92.pdf>.

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

### DHEC Newborn Screening Follow Up:

(803) 898-0593 or (803) 898-1969

**Public Health Laboratory:** (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.