

### OUR BEST FOOT FORWARD

#### Updates in Screening for Classical Galactosemia and Biotinidase Deficiency

Effective January 5, 2018, new cut-off values for total galactose (GAO), galactose-1-phosphate uridylyltransferase (GALT) and biotinidase (BIO) were applied to specimens received on and after that date. As a result of the transition to the automated Genetic Screening Processor (GSP) instrumentation, new cut-offs have been in use for population screening for over three months.

Data accumulated over three months of screening has been evaluated to adjust the cut-offs to reduce the frequency of potentially false positive screening results. These new cut-offs are shown in the table below.

	9/20/2017	New Expected Range
<b>GAO</b>	>7.50 mg/dL	<b>12.0 mg/dL</b>
<b>GALT</b>	≥ 5.4 U/dL	<b>≥3.75 U/dL</b>
<b>BIO</b>	≥76.5 U/dL	<b>≥70.0 U/dL</b>

These adjusted cut-offs provide more accurate screening for classical galactosemia and biotinidase deficiency. Classical galactosemia is an inborn error of metabolism that effects the body's ability to breakdown the simple sugar galactose using the GALT enzyme. Galactose is the principle carbohydrate in breastmilk and infant formula. If left untreated, infants can experience lethargy, jaundice, liver damage and even sepsis.

In 2017, the SC Newborn Screening program identified 2 infants that were diagnosed with classical galactosemia. Biotinidase deficiency is an inherited disorder where the body cannot recycle the vitamin biotin. If left untreated, infants can experience seizures, hypotonia, alopecia and candidiasis. In 2017, the SC Newborn Screening program identified 4 infants what were diagnosed with biotinidase deficiency. Newborn screening is the first line of defense when identifying these serious conditions.

#### If you have any questions, please call:

Dr. Ona Adair  
Chemistry Division Director  
803-896-0991

Sandi Hall  
Newborn Screening Section Supervisor  
803-896-0891

#### Sources:

<https://ghr.nlm.nih.gov/condition/biotinidase-deficiency>  
<https://ghr.nlm.nih.gov/condition/galactosemia#>



#### Beyond the Bloodspot

After newborn screening results have been reported and the diagnosis of a metabolic disorder has been made, DHEC continues servicing patients. The Metabolic Formula Program provides metabolic formula to all South Carolina residents with inborn errors of metabolism. Metabolic formulas are very complex. They are made up of specific fats, carbohydrates and amino acids necessary for each metabolic disorder.

Many patients rely solely on these formulas through infancy and continue to require them into adulthood. For example, a patient with glutaric aciduria type 1 would require a formula free of the amino acid lysine and low in tryptophan. Metabolic formulas are critically necessary to these patients and DHEC will continue to provide them to residents throughout their lifespan and as funds remain available.

#### ON THE SPOT

The hospitals listed below had 0% unsatisfactory specimens for 2017:

- Hilton Head Hospital

Please try to attend a First Time Every Time hospital training workshop in your region.

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

**Upstate:** Feb 23rd at Greenville Hospital System

**Pee Dee:** May 23rd at McLeod Regional Medical Center

**Midlands:** August 17th at Palmetto Health Richland

**Lowcountry:** November 8th at MUSC



## ON THE RUN

Thanks to all the hospitals who are participating in the DHEC NBS Specimen Transport Protocol using FedEx services. There are currently 29 participating hospitals! We believe the specimen transport protocol is a major factor in the decreased specimen transit time from the hospital to the laboratory that is reflected in some monthly hospital reports. Once full participation is achieved, the specimen transport protocol may serve as a shining example of how successful quality improvement initiatives can be achieved with the commitment of the NBS program and our partners, like you.

**For those of you already enrolled,** FedEx will be contacting you in the near future about setting up automatic shipments. That's right - no more daily scheduling for pick-up!

**For those of you who have not enrolled in the program, there is still time.** The FedEx Priority Overnight service is available at no cost to you. All you have to do is enroll through the DHEC Public Health Laboratory. If you would like to enroll, **please contact Toshiro Washington** ([washints@dhec.sc.gov](mailto:washints@dhec.sc.gov), 803-896-0795), and he will gladly enroll your facility. Thank you all for your partnership in making the NBS program the best it can be for SC newborns!

### Lab Closings:

February 19th and May 28th

Please continue to send specimens regardless of closings.

## EDUCATIONAL INFORMATION:

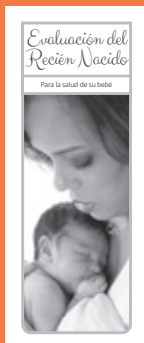
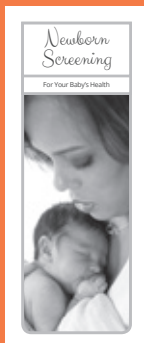
**Are you educating your parents about Newborn Screening?**

Visit [www.scdhec.gov/newbornmetabolicscreening](http://www.scdhec.gov/newbornmetabolicscreening) to find our newborn screening brochure and educational handouts for parents and providers.

**Need NBS brochures?**

- ML-000032 for English
- ML-025096 for Spanish

Please go to [www.scdhec.gov/Agency/EML](http://www.scdhec.gov/Agency/EML) to order the brochures.



## 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.