MEMORANDUM

DATE: May 26, 2026

TO: South Carolina Newborn Screening Community

FROM: Jenny Meredith, Ph.D., HCLD(ABB)
SC DPH Public Health Laboratory (PHL) Director

SUBJECT: South Carolina (SC) Newborn Screening (NBS) Program's new conditions, testing methodology and updated expected ranges

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Effective May 31st, 2026, the SC Newborn Screening Panel is expanding to include screening for two additional Lysosomal Storage Disorders (LSDs): Mucopolysaccharidosis Type II (MPS II) and Fabry Disease.

MPS II, also known as Hunter syndrome, is part of a family of diseases which are defined by the body's inability to break down and recycle certain long sugar molecules called mucopolysaccharides. The screening enzyme for MPS II is iduronate-2-sulfatase (I2S), which is decreased in patients at risk of having MPS II.

Fabry disease is a genetic condition that prevents the body from processing glycosphingolipids properly. The screening marker is alpha-galactosidase A (GLA or alpha-Gal A), which is decreased in patients at risk of having Fabry disease.

Both of these conditions are rare, with a prevalence of less than 40 cases per year in the U.S.

With the addition of these new conditions, the SC Newborn Screening Laboratory will now be screening for a total of five LSDs. The testing methodology for this screening is changing from Flow Injection Analysis Mass Spectrometry (FIA-MS) to Liquid Chromatography Mass Spectrometry (LC-MS). The current and new expected ranges for each of the LSDs are summarized in the table below.

Lysosomal Storage Disorder and Enzyme	Current Expected Range	New Expected Range
Pompe Disease: acid-α-glucosidase (GAA)	> 20% of the Median	**> 20% of the Median**
Mucopolysaccharidosis Type I (MPS I): α-L-iduronidase (IDUA)	> 10% of the Median	**> 10% of the Median**
Krabbe Disease: β-galactocerebrosidase (GALC)	> 15% of the Median	**> 10% of the Median**
Mucopolysaccharidosis Type II (MPS II): L-iduronate 2-sulfatase (I2S)	N/A	**> 20% of the Median**
Fabry Disease: α-galactosidase A (GLA)	N/A	**> 15% of the Median**

Specimens that fall outside the Expected Range for any LSD will be sent to the Greenwood Genetic Center (GGC) Laboratory for second-tier testing.

Additional updates include the following:
- All specimens collected before the newborn is 24 hours of age will be reported as inconclusive for Fabry and Krabbe disease. Another specimen will need to be collected after 24 hours of age.
- Fabry and Krabbe disease will be reported as inconclusive for all newborns with a birthweight of less than 2500 grams (low and very low birthweight), until the newborn is 28 days old. A specimen must be collected at or after 28 days for this population.

This change is part of the SC NBS Program's commitment to continuous improvement in providing quality healthcare which is the goal of the Public Health Laboratory. Please direct any questions to Beth Bair, M.S., Ph.D., Newborn Screening Section Director at bairea@dph.sc.gov; 803-896-0991.