June 24, 2024

Dear Health Care Providers:

The Florida Department of Health, Newborn Screening (NBS) Program, is pleased to announce that beginning July 1, 2024, all newborns screened will be tested for Mucopolysaccharidosis Type II (MPS II).

It is important that each provider review the enclosed information sheet to become familiar with the muti-tiered process for screening and the procedures following identification of a positive MPS II screen. Newborns with presumptive positive results will be referred to the nearest contracted Genetic Specialty Center in Gainesville, Tampa, or Miami.

Follow-up staff from the NBS Program will direct these procedures and advise on actions needed. These procedures will only apply to MPS II; no changes will be made to the protocols currently practiced for other conditions on the screening panel.

If you have any questions, please contact the Florida NBS Program at 850-245-4201.

Thank you for your continued support of the Florida NBS Program.

Sincerely,

Joseph A. Ladapo, MD, PhD
State Surgeon General

Enclosure

Newborn Screening for Mucopolysaccharidosis Type II (MPS II) in Florida

Definition: A lysosomal storage disorder that affects many different parts of the body. MPS II, also known as Hunter syndrome, occurs mostly in males, although it has been reported in females in rare instances. The age of onset and severity of the disease is dependent on the form. Symptoms of the severe form of MPS II can begin between ages two and four and progresses faster than newborns identified with the attenuated (less severe) form of MPS II. Those identified with the attenuated form do not typically develop symptoms until late in childhood, or even adolescence.

Incidence: MPS II affects approximately 1 in 100,000 to 1 in 170,000 males.

Florida Method of Screening:

First Tier Screening: Screening for MPS II is completed by analysis of the iduronate sulfatase (I2S) enzyme. If analysis is normal, the sample is deemed within normal limits. If the I2S analysis is <15% of the daily median, second tier screening is performed.

Second Tier Screening: Samples will be sent for biochemical analysis to Revvity Omics. If the analysis is normal, the sample is deemed within normal limits. If out-of-range, third tier screening is performed.

Third Tier Screening: Sequencing of the I2S gene. Only infants identified with a pathogenic variant, likely pathogenic variant, or variant of uncertain significance will warrant a referral for confirmatory testing.

Referral to Genetic Specialty Center: Newborns with out-of-range results for MPS II are referred to one of three Genetic Specialty Centers for diagnostic evaluation following completion of third tier screening. If diagnosed with MPS II, patients will be referred to additional specialists, as appropriate, for clinical assessment and treatment.

For more information about MPS II, please visit the following sites:

Gene Reviews
https://www.ncbi.nlm.nih.gov/books/NBK1274/

Genetics Home Reference
https://medlineplus.gov/genetics/condition/mucopolysaccharidosis-type-ii/

Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/6675/mucopolysaccharidosis-type-2

Baby's First Test
https://babysfirsttest.org/newborn-screening/conditions/mucopolysaccharidosis-type-ii