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PHILIP D. MURPHY
Governor

SHEILA Y. OLIVER

Lt. Governor

SHEREEF M. ELNAHAL, MD, MBA
Commissioner

## New Jersey to Begin Newborn Screening for 6 Lysosomal Storage Disorders (LSDs) on Monday, July 8, 2019

## What are Lysosomal Storage Disorders (LSDs)?

Lysosomal Storage Disorders are a class of over 50 inherited metabolic disorders characterized by the accumulation of substances such as proteins, carbohydrates, and old cell parts inside cells. This happens because the body lacks one of the enzymes required to break down this material. As these materials accumulate they become toxic, damaging the cells and organs in the body.

## Which LSDs will be screened for in New Jersey?

New Jersey will screen for 6 LSDs: 1. Krabbe Disease, 2. Pompe Disease, 3. Gaucher Disease, 4. Fabry Disease, 5. Niemann-Pick Disease Types A and B, and 6. Mucopolysaccharidosis Type 1 (MPS1).

When will these 6 LSDs be added to the newborn screening panel?

Starting with all specimens received on Monday, July 8, 2019, the New Jersey Newborn Screening (NBS) Program will screen all newborns for these 6 LSDs. For the first 2 weeks of screening, results that are within acceptable limits will not be included on the laboratory report. All abnormal results will be reported to the appropriate care provider. Beginning on Monday, July 22, 2019, all LSD results will be included on the laboratory report.

What are the benefits of adding these 6 LSDs to the newborn screening panel? Newborn screening will allow infants to be diagnosed and treated sooner. Treatment can greatly enhance the quality of life of individuals with these disorders.

How will the Newborn Screening Laboratory screen for the 6 LSDs?
Using the dried blood spots currently collected from all newborns, the NJ NBS
Laboratory will evaluate the activity of the 6 enzymes responsible for these diseases.
Enzyme activity will be measured using tandem mass spectrometry.

What will happen if an infant screens positive for one of the 6 LSDs? Infants identified as having significantly decreased enzyme activity will be referred to a pediatric metabolic geneticist for evaluation.

Please notify the NBS Laboratory (609-530-8371) if you have any concerns or questions regarding the new test results you receive.