

**Mission:**

To protect, promote & improve the health of all people in Florida through integrated state, county & community efforts.



**Ron DeSantis**  
Governor

**Scott A. Rivkees, MD**  
State Surgeon General

**Vision:** To be the Healthiest State in the Nation

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February 4, 2020

Dear Health Care Providers:

The Florida Department of Health, Newborn Screening Program, is pleased to announce that beginning February 3, 2020, all newborns screened will be tested for Pompe Disease and Mucopolysaccharidosis Type I (MPS I).

It is important that each provider use the enclosed information sheet to become familiar with the protocol following identification of a positive result for Pompe Disease and MPS I. Newborns with presumptive positive results will be referred to the nearest Genetic Specialty Center. The referral centers are located in Gainesville, Tampa, and Miami.

Follow-up staff from the Newborn Screening Program will direct these procedures and advise of actions needed. These procedures will only apply to Pompe and MPS I; 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 Newborn Screening Program at 850-245-4201.

Thank you for your continued support of the Florida Newborn Screening Program.

Sincerely,

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Marcy R. Hajdukiewicz, MS  
Interim Director  
Division of Children's Medical Services

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Carina Blackmore, DVM, PhD, Dipl, ACVPM  
State Epidemiologist  
Director  
Division of Disease Control and Health Protection

Enclosure  
SR/bw

## Newborn Screening for Mucopolysaccharidosis Type I (MPS I) in Florida

**Definition:** A lysosomal storage disorder that affects many different parts of the body. MPS I is made up of a wide spectrum of severity and affected newborns are categorized as having the severe form or attenuated (less severe) form.

**Incidence:** The severe form of MPS I affects approximately 1 in 100,000 newborns. The attenuated form occurs in approximately 1 in 500,00 newborns.

### Florida Method of Screening:

**First Tier Screening:** Screening for MPS I is completed by analysis of the alpha-L-iduronidase (IDUA) enzyme. If analysis is normal, the sample is deemed within normal limits. If the IDUA analysis is <8% of the daily median, second tier screening is performed.

**Second Tier Screening:** Samples will be sent for biochemical analysis to PerkinElmer Genomics. If the analysis is normal, the sample is deemed within normal limits. If abnormal, third tier screening is performed.

**Third Tier Screening:** Sequencing of the IDUA gene. Abnormal results for first and second tier screening still warrant a referral regardless of whether an IDUA mutation is detected.

**Referral to Genetic Specialty Center:** Newborns with abnormal results for MPS I are referred to one of three Genetic Specialty Centers for diagnostic evaluation following completion of third tier screening. If diagnosed with MPS I, patients will be referred to additional specialists, as appropriate, for clinical assessments and need for treatment options.

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

Gene Reviews <https://www.ncbi.nlm.nih.gov/books/NBK1162/>

Genetics Home Reference <https://ghr.nlm.nih.gov/condition/mucopolysaccharidosis-type-i>

Genetic and Rare Diseases Information Center  
<https://rarediseases.info.nih.gov/diseases/10335/mucopolysaccharidosis-type-i>

Baby's First Test  
<https://www.babysfirsttest.org/newborn-screening/conditions/mucopolysaccharidosis-type-i>

## Newborn Screening for Pompe in Florida

Definition: A lysosomal storage disorder that affects many different parts of the body. There are two forms of Pompe Disease, Infantile Onset Pompe Disease and Late Onset Pompe Disease.

Incidence: Pompe affects approximately 1 in 40,000 newborns.

### Florida Method of Screening:

First Tier Screening: Screening for Pompe is completed by analysis of the acid alpha-L-glucosidase (GAA) enzyme. If analysis is normal, the sample is deemed within normal limits. If the GAA analysis is <15% of the daily median, second tier screening is performed.

Second Tier Screening: Samples will be sent for biochemical analysis to PerkinElmer Genomics. If the analysis is normal, the sample is deemed within normal limits. If abnormal, third tier screening is performed.

Third Tier Screening: Sequencing of the GAA gene. Abnormal results for first and second tier screening still warrant a referral regardless of whether a GAA mutation is detected.

Referral to Genetic Specialty Center: Newborns with abnormal results for Pompe are referred to one of three Genetic Specialty Centers for diagnostic evaluation following completion of third tier screening. If diagnosed with Pompe, patients will be referred to additional specialists, as appropriate, for clinical assessments and need for treatment options.

For more information about Pompe, please visit the following sites:

Gene Reviews <https://www.ncbi.nlm.nih.gov/books/NBK470558/>

Genetics Home Reference <https://ghr.nlm.nih.gov/condition/pompe-disease>

Genetic and Rare Diseases Information Center  
<https://rarediseases.org/rare-diseases/pompe-disease/>

Baby's First Test  
<https://www.babysfirsttest.org/newborn-screening/conditions/pompe>