



State of Louisiana

Louisiana Department of Health
Office of Public Health

LOUISIANA NEWBORN SCREENING PANEL UPDATE: NEW SCREENINGS TO BE ADDED TO THE NEWBORN SCREENING PANEL STARTING JANUARY 2022

In January 2022, the Louisiana Department of Health (LDH) will be adding screening for Spinal Muscular Atrophy (SMA), Mucopolysaccharidosis Type I (MPS I), and Glycogen Storage Diseases Type II (Pompe Disease) to the Louisiana Newborn Screening Panel.

What Are These Conditions?

Spinal Muscular Atrophy (SMA) is a disorder that progressively destroys motor skills and affects the ability to breath, swallow and talk. SMA occurs in 1 out of every 11,000 live births. Louisiana is expected to see SMA in 5 to 6 infants each year. The addition of SMA will have the largest effect on the infants detected and their families. Undiagnosed and untreated SMA may result in a life span of less than 1 year and has a traumatic effect on families. Early detection will allow affected infants receive early diagnosis and treatment, decrease the mortality rate and have a positive effect on the family structure.

Mucopolysaccharidosis Type I (MPS I) is a rare genetic disorder caused by a change in a single human gene. Studies of patients with symptoms suggest that about 1 out of every 100,000 people has MPS I. People with MPS I do not have enough of an enzyme that helps to break down certain waste products in cells. Babies with MPS I appear normal. There are 2 types of MPS I: the severe type and the attenuated type. Most children with MPS I have the severe type. In this type, MPS I can cause problems with the heart, airways, eyes and ears, muscles, bones, joints, and brain. These problems can worsen quickly and cause early death. Screening MPS I detects approximately 44 babies with the condition each year (about 1.1 out of every 100,000 children born). It can prevent up to 2 deaths before age 5 years due to the disease each year. The anticipated prevalence in Louisiana is about 1 case every 2 years.

Glycogen Storage Diseases Type II (Pompe Disease) is a rare disease caused by a change in a single gene. Studies of patients with symptoms suggest that between 1 and 2.5 out of every 100,000 people have Pompe Disease. The anticipated prevalence in Louisiana is about 1-2 case per year. People with Pompe Disease do not have enough of a certain enzyme that helps the body break down stored sugar. Babies with the disease appear normal. There are 2 types of Pompe Disease: infantile- and late-onset. The first type can cause muscle problems that begin in early infancy. Most children with Pompe Disease have the late-onset type. Problems from the disease can worsen quickly and cause death within the first year.

When will the new conditions be added to the Louisiana Newborn Screening Panel?

This addition is anticipated to begin in January 2022.

Will there be a fee increase?

There will be no increase in the newborn screening fee at this time.

Will more blood be needed to complete this test?

No additional blood is needed. Continue to collect 5 completely saturated or filled blood spots.

Where can I find more information on the new conditions?

Find more information at [Baby's First Test](#).

For questions or comments, please contact the Newborn Screening Program at 504-568-8254