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New Jersey to Begin Newborn Screening for Spinal Muscular Atrophy (SMA) on January 31, 2022

What is spinal muscular atrophy (SMA)?

SMA is a genetic neuromuscular condition that causes a loss of the motor neurons, resulting in progressive muscle weakness, atrophy, and poor muscle tone. Approximately one in 10,000 babies is born with SMA, including about 10 babies born in New Jersey each year. Babies with SMA appear healthy at birth, but due to severe, progressive muscle weakness, affected infants are unable to support their heads or sit unassisted and have difficulty breathing and swallowing.

When will SMA be added to the newborn screening panel?

Starting with all specimens tested on January 31, 2022, the New Jersey Newborn Screening (NBS) Program will screen all newborns for SMA.

What are the benefits of adding SMA to the newborn screening panel?

Newborn screening will allow infants to be diagnosed and treated as soon as possible. Without treatment, most children with SMA are unable to move independently, may be unable to breathe independently, and children with the most severe form may die before the age of 2. However, with early detection and treatment – there are 3 FDA approved treatments – survival rate is high.

How will the Newborn Screening Laboratory screen for SMA?

The NJ NBS Laboratory will screen for SMA by evaluating the SMN1 gene for deletion of Exon 7. This analysis has been researched extensively as a reliable and sensitive method. The deletion of exon 7 of the SMN1 gene is the cause of 95% of cases of SMA.

What will happen if an infant screens positive for SMA?

Infants with abnormal results will be referred for follow-up with a geneticist for evaluation and diagnostic testing.

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