April 27, 2020

Dear Colleagues:

The Florida Department of Health, Newborn Screening Program (NBS) is pleased to announce that beginning in May 2020, all newborns screened will be tested for Spinal Muscular Atrophy (SMA).

It is important that each provider use the enclosed information sheet to become familiar with the protocol following identification of a positive result for SMA. Newborns with presumptive positive results will be referred to the nearest NBS Genetic Center by the NBS Program. These referral centers are at the University of Florida (Gainesville), University of Miami, and University of South Florida (Tampa).

Follow-up staff from the Newborn Screening Program will direct these procedures and advise of actions needed. These procedures will only apply to SMA; 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,

Robert D. Karch, MD, MPH, FAAP
Deputy Secretary for Children's Medical Services

Enclosure
RK/bw

Newborn Screening for Spinal Muscular Atrophy (SMA) in Florida

Definition: Spinal Muscular Atrophy (SMA) is a genetic disorder that affects the control of muscle movement due to the loss of specialized nerve cells called motor neurons. The progressive loss of motor neurons causes weakness and muscle atrophy. SMA affects muscles used for activities such as crawling, walking, sitting up, and controlling head movements. Severe cases of SMA affect the muscles used for breathing and swallowing, leading to early death.

Incidence and Prognosis: The incidence of SMA is approximately 1 in 11,000 newborns. Over half of these patients have the most severe form of SMA; these infants appear normal at birth but develop symptoms in the first few weeks to months of life. Without treatment, infants never achieve milestones, such as sitting and have a life-expectancy of less than 2 years. There are approved medications for treatment of SMA. Early treatment, ideally before symptom onset, provides the best response to these medications.

Florida's Method of Screening:

First Tier Screening: Screening for SMA is accomplished by performing DNA analysis for deletion of exon 7 in the SMN1 gene. Most, but not all patients with SMA, have a (homozygous) deletion of this SMN1 exon 7 on both alleles of chromosome 5q.

Parents should not be told that a negative screen rules out SMA. This testing method does not identify other types of mutations in the SMN1 gene or other genetic types of SMA.

Referral to an NBS Genetic Center: Abnormal results for SMA will be referred to one of the contracted Genetic Referral Centers by the NBS Program for confirmatory testing and genetic counseling. Once a diagnosis of SMA has been made, infants will be referred to an NBS SMA Treatment Center for ongoing care and treatment management.

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

- Genetics Home Reference
  - https://ghr.nlm.nih.gov/condition/spinal-muscular-atrophy
- The National Center for Biotechnology Information
  - https://www.ncbi.nlm.nih.gov/books/NBK1352/
- The Muscular Dystrophy Association
  - https://www.mda.org/disease/spinal-muscular-atrophy

SMA STATE FACT SHEET

Florida

Est. individuals living with SMA: 705
Est. babies born with SMA annually: 20
Est. number of SMA carriers: 425,987

Estimates for incidence, prevalence, and carriers are based on 2018 birth and population data for the state of Florida.

ABOUT SMA AND CURE SMA

Spinal muscular atrophy (SMA) – the number one genetic cause of death for infants – robs people of physical strength by affecting the motor nerve cells in the spinal cord, taking away the ability to walk, eat, or breathe. The disease is caused by a mutation in the survival motor neuron gene 1 (SMN1). Without enough of the SMN protein, nerve cells cannot function properly and eventually die, leading to debilitating and often fatal muscle weakness.

Cure SMA is the largest network of families, clinicians, and research scientists working together to advance SMA research, support affected individuals/caregivers, and educate the public and professional communities about SMA.

FLORIDA CHAPTER INFORMATION

Cure SMA has 36 volunteer-led chapters across the United States.
To find and contact the Florida chapter, visit www.curesma.org/chapters

SMA CARE CENTER NETWORK

SMA Care Center Network is the centerpiece of our efforts to address the changing landscape of SMA. The goal of the SMA Care Center Network is to develop an evidence-based standard of care that will improve the lives of all those affected by SMA.

NEW HOPE FOR TREATING SMA

Thanks to the dedication of our community and the ingenuity of our researchers, we now have treatments that target the underlying genetics of SMA. Currently, there are two treatments for SMA approved by the U.S. Food and Drug Administration (FDA) – Spinraza and Zolgensma. Both are SMN-enhancing treatments.

But our work is not done. We know what needs to be done to develop and deliver effective therapies that target other systems, pathways, and processes affected by SMA. Our goal is a combination of therapeutic approaches that can be tailored to each individual's age, stage, and type of SMA. These breakthroughs will continue to change the course of SMA for everyone affected—from infants to adults—and eventually lead to a cure.

TYPES OF SMA

There are four primary types of SMA that are based on the age of onset and highest physical milestone achieved. Type 1 is the most severe and most common, affecting 60 percent of those with SMA and is typically diagnosed during an infant's first six months.

Type 1 SMA
Onset: Before 6 months
Milestones: No sitting

Type 2 SMA
Onset: 6 - 18 months
Milestones: Sitting, not walking

Type 3 SMA
Onset: Childhood after 12 months
Milestones: Walking

Type 4 SMA
Onset: After 30 years old
Milestones: Normal

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925 Busse Road, Elk Grove Village, IL 60007 | 800.886.1762 | Fax: 847.367.7623 | info@cureSMA.org | cureSMA.org