

## NORTH CAROLINA NEWBORN SCREENING PROGRAM

# Spinal Muscular Atrophy Fact Sheet for Providers

Spinal muscular atrophy (SMA) is a genetic neuromuscular disorder caused by a mutation on the SMN1 gene which leads to irreversible loss of motor neurons in the brainstem and spinal cord. The loss of functioning motor neurons leads to progressive muscle weakness and impairment of respiratory and bulbar functions.

### What are the signs and symptoms of SMA?

There are different types of SMA based on age of onset and symptom severity. The most common form is SMA type 1 which has symptom onset within the first few weeks or months of life. Early symptoms of SMA include hypotonia, lack of motor development, and muscle weakness. Infants with the most severe form may also have tongue fasciculations which cause problems with sucking or swallowing. Some infants may have breathing difficulties. Later-onset SMA presents with a less severe course between 6 and 18 months of age (type 2), early childhood (type 3), or adults (type 4) and generally correlates with increasingly higher levels of motor function.

### Is SMA treatable?

Yes, there are different types of treatment available, including an FDA-approved intrathecal injection (nusinersen/Spinraza) and an intravenous gene therapy product (onasemnogene abeparvovec-xioi/Zolgensma). Treatment should be started as soon as possible since SMA is a progressive disease. Infants with SMA who are treated early tend to have better outcomes.

### What should I do when a patient has a positive newborn screen for SMA?

Given how quickly symptoms of SMA may develop, it's critical to contact the family as soon as possible and refer the baby to a specialist for confirmatory testing. Remember, the newborn screen is not a diagnostic test and although false positives are rare, they are possible. Please take the following actions:

- Contact the family within 24 hours to describe the newborn screening result, provide the SMA parent fact sheet, and determine the family's preferred specialty care center. The SMA fact sheet has links to educational resources for the family.
- Refer baby and family to the specialty care center to coordinate follow-up with pediatric neurologist. See contact information in resource list.
- The specialty care center will follow-up with the family within 72 hours to schedule an appointment.

### Where do I go for more information?



#### Genetics Home Reference:

<https://medlineplus.gov/genetics/condition/spinal-muscular-atrophy/>



#### Cure SMA:

[www.curesma.org/newborn-screening-for-sma/](http://www.curesma.org/newborn-screening-for-sma/)



#### ACMG ACT Sheet:

<https://www.acmg.net/PDFLibrary/SMA-ACT-Sheet.pdf>

**If you have questions**, please contact the Newborn Screening Follow-up Program at 919-218-6460 or the Children with Special Health Care Needs Help Line at 1-800-737-3028.



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