

# Nebraska Newborn Screening Program

## Physician ACT sheet for POSITIVE SPINAL MUSCULAR ATROPHY (SMA) RESULT

### You Should Do The Following:

- Consult with the following pediatric neurologist to determine follow up:  
Dr. Rathore at 402-955-5372
- Notify baby's family of newborn screening results. Assess status of newborn and provide follow up information as discussed with Pediatric Neurologist.
- Arrange for follow up as specified by the pediatric neurologist.
- Arrange referral to pediatric neurologist who treats SMA for a comprehensive evaluation.

### SCREENING TEST RESULTS

The newborn screen for this infant revealed an abnormal result for Spinal Muscular Atrophy (SMA). The Survival Motor Neuron Gene (SMN1) gene was **NOT** detected.

Absence of the SMN1 gene causes spinal muscular atrophy (SMA). The SMN1 gene provides instructions for making the survival motor neuron protein. This protein is particularly important for the maintenance of motor neurons. The SMN2 genes cause production of some replacement protein for motor neuron survival. The copy number of SMN2 helps determine the type of SMA. **Referral to a pediatric neurologist for follow-up should be made immediately.**

If you have any questions about the newborn screening result or your next steps, please call the Nebraska Newborn Screening Program at (402) 471-0374.

### CONDITION INFORMATION

SMA is a neurodegenerative disease caused by mutations in the survival motor neuron 1 (SMN1) gene. As a result of these mutations, motor neurons cannot function properly and eventually die. It is the number one genetic cause of infant deaths.

There are four types of SMA – I, II, III, and IV – classified by age of onset and highest motor milestone reached. Type I is the most common and the most severe. SMA type I may be apparent at birth or within the first few months of life. Without treatment, these children are never able to sit without support and die within the first two years of life. Types II through IV are also significant and present with varying symptoms.

### TREATMENT

There are now several FDA approved treatments available. Please consult with the Pediatric Neurologist for more information.

### RESOURCES

Nebraska Newborn Screening Program  
(402) 471-0374

[www.babysfirsttest.org](http://www.babysfirsttest.org)

[www.curesma.org](http://www.curesma.org)

[www.smafoundation.org](http://www.smafoundation.org)

