

Parent Information

Newborn Screening Test Result for SPINAL MUSCULAR ATROPHY (SMA)

What is Newborn Screening?

After babies are born, five drops of blood are collected from the baby's heel to test for certain diseases. These conditions usually cannot be found simply by looking at or examining a baby.

What is Spinal Muscular Atrophy (SMA)?

SMA is a disorder that affects the nervous system. SMA damages the nerve cells that control the muscles. Because of this, a child with SMA may not move normally and will get weaker over time.

There are four types of SMA – I, II, III, and IV. The severity of health problems and when problems start vary. Your doctor or a doctor who specializes in SMA will explain this further to you.

What does a positive screening result mean?

Your baby is missing both copies of the survival motor neuron gene 1 (SMN1). The absence of SMN1 and the number of copies of the SMN2 gene helps determine what type of SMA your baby has. More testing may need to be done to confirm if your baby has this disorder.

What do I need to do now?

Your baby's doctor will tell you about the specific plan for your baby. If further testing is needed, it should be done without any delays even though your baby appears well. If your baby does have SMA, finding out quickly will allow treatment to be started promptly.

What problems can SMA cause?

SMA is a lifelong condition. If untreated, it can cause:

- Swallowing and feeding difficulties
- Breathing difficulties
- Muscle weakness
- Delayed motor milestones
- Loss of skills like sitting or crawling
- Shortened lifespan

How is SMA treated?

There are now multiple FDA approved treatments for SMA. Following further testing the pediatric neurologist will determine a specific plan of care that is most appropriate for your child.

Children with SMA should see their regular doctor and a doctor who specializes in SMA.

Resources for parents

Nebraska Newborn Screening Program
(402) 471-0374

www.babysfirsttest.org

www.curesma.org/

www.smafoundation.org

