

Positive Result:

Blood Spot Screen Result Notification



SMN1 Absent

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby is missing both copies of the SMN1 gene.

What does this mean?

It is very likely that your baby has spinal muscular atrophy (SMA).

What happens next?

Your baby's doctor will refer you to a child neurologist who can treat SMA. The specialists will want to check on your child's health and send labs.

What is spinal muscular atrophy?

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 cannot 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. Newborn screening cannot tell the difference between these four types.

What health problems can it 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

Children with SMA can benefit from prompt and careful treatment.

What treatment options are available?

Although SMA cannot be cured, it can be treated. Possible treatments can include:

- Supportive therapies like respiratory therapy
- Drug therapy

Children who are treated with drug therapy before health problems begin do better and live longer. Since the drug has only been approved for use since 2016, long-term studies are not yet complete.

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

Resources

Genetics Home Reference:
<http://ghr.nlm.nih.gov>

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org