Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

SMN1 Absent

This screening result is likely a true diagnosis of spinal muscular atrophy (SMA); type unknown. Treatment is available and may be needed right away.

Next Steps

Within one business day, you should take the following recommended actions:

- **Consult** with a child neurologist who provides intrathecal drug therapy for SMA. Contact information for treating neurologists can be found on the resource list provided.
- **Contact** family to notify them of the newborn screening result and assess symptoms.
- **Evaluate** infant (hypotonia, areflexia, swallowing and feeding difficulties, tongue fasciculation); arrange immediate referral if symptomatic.
- **Arrange** referral to a child neurologist who treats SMA for a comprehensive evaluation.

If you have questions about the newborn screening result or your next steps, an on-call Newborn Screening Program genetic counselor is available at (651) 201-3548.

Review with Family

Discuss this result with the family as MDH has **not** notified them. Share your follow-up plan with them. Educate family about signs, symptoms, and when to contact you with concerns.

False Positives

Unlikely since both copies of the SMN1 gene were absent.

Differential Diagnosis

The absence of both copies of the SMN1 gene is associated with:

- Spinal muscular atrophy (SMA) — Incidence of 1 in 6,000

Clinical Summary

SMA is a neurodegenerative disease caused by mutations in the survivor motor neuron 1 (SMN1) gene. As a result of these mutations, motor nerve cells cannot function properly and eventually die.

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.

Drug therapy given intrathecally is available and has been shown to significantly modify the course of the disease with early treatment (especially pre-symptomatically) producing better outcomes. Since the drug has only been FDA-approved since 2016, long-term studies are not yet complete.