Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Urgent Positive Severe Combined Immunodeficiency (SCID)

Next Steps

This week, you should take the following recommended actions:

- **Consult** with a pediatric immunologist/ infectious disease specialist. Contact information for the specialists can be found on the resource list provided.
- **Contact** family to notify them of the newborn screening result and assess for signs of illness or infection; arrange immediate hospitalization if symptomatic.
- **Arrange** laboratory testing and referral as recommended by the immunologist/infectious disease specialist.

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 the follow-up plan with them. Educate family about signs, symptoms, and when to contact you with concerns. Until further evaluation is complete, the family should avoid: unnecessary public exposures, individuals who have recently received a live vaccine, and contact with ill people.

False Positives

Screening result can be impacted by transfusion, prematurity, illness, and newborns with certain congenital anomalies.

Differential Diagnosis

This result is primarily associated with:

- Severe combined immunodeficiency (SCID) — Incidence of 1 in 50,000 (more common in the Amish, Somali, and Navajo indian populations)

Other disorders to consider:

- Syndromic T-cell lymphopenias like 22q11.2 deletion (a.k.a. DiGeorge) syndrome and cartilage-hair hypoplasia
- Secondary T-cell lymphopenias due to chylothorax, hydrops, third spacing, etc

Clinical Summary

SCID is a group of rare, potentially fatal disorders with multiple causes and inheritance patterns. The most common form is X-SCID followed by adenosine deaminase (ADA) SCID. Individuals with SCID have a combined absence of T-lymphocytes and B-lymphocytes resulting in virtually no immune protection from bacteria and viruses.

Newborns with SCID are typically asymptomatic. In most cases, infections are the first indication. Affected individuals have persistent or recurrent infections in infancy and die early if effective intervention is not provided.

Treatment with immunoglobulin replacement therapy, enzyme replacement therapy, and antibiotics to prevent infection exists. However, hematopoietic stem cell transplantation is the most effective treatment. Transplants performed within the first few months of life or before infections occur are most successful.