Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

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:

- T-cell lymphopenias

Other disorders to consider:

- Severe combined immunodeficiency (SCID)

Clinical Summary

T-cell lymphopenias can be secondary, syndromic, or idiopathic.

T-cell lymphopenia has been identified secondary to some congenital anomalies. Examples include cardiac defects, gastrointestinal malformations, hydrops, and chylothorax.

Syndromic causes of T-cell lymphopenia include 22q11.2 deletion (a.k.a. DiGeorge) syndrome, trisomy 21 (a.k.a. Down syndrome), ataxia telangiectasia, cartilage-hair hypoplasia, and CHARGE syndrome.

There are some infants with T-cell lymphopenia where the cause of their lymphopenia cannot be determined even after diagnostic testing.

Whatever the cause of the T-cell lymphopenia, these children need medical oversight. Additionally, live vaccine avoidance and infection prophylaxis may be beneficial for these infants.

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