

Positive Result: Blood Spot Screen Result Notification



Elevated Succinylacetone

Next Steps

This week, you should take the following recommended actions:

- **Consult** with a metabolic specialist. Contact information for the metabolic specialists can be found on the resource list provided.
- **Contact** family to notify them of the newborn screening result and assess symptoms (diarrhea, vomiting, lethargy).
- **Evaluate** infant (failure to thrive, jaundice, tendency to bleed); arrange immediate referral if symptomatic
- **Arrange** referral to a metabolic specialist for further diagnostic work-up.

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 need for infant to avoid protein. Discuss signs, symptoms, and need for urgent treatment if infant becomes symptomatic.

False Positives

Unlikely. Elevated succinylacetone is pathognomonic for tyrosinemia type 1.

Differential Diagnosis

Elevated succinylacetone is primarily associated with:

- Tyrosinemia type 1 (TYR I) — Incidence of 1 in 100,000

Clinical Summary

Tyrosinemia is an amino acid disorder where the body is unable to break down the amino acid, tyrosine leading to a toxic buildup. There are three types of tyrosinemia, but type 1 is the only type where succinylacetone is elevated.

Tyrosinemia type 1 is the most severe form. Affected newborns present with diarrhea, vomiting, failure to thrive, lethargy, and a 'cabbage-like' odor. Additionally, affected newborns can develop hepatomegaly, jaundice, kidney problems, and liver failure. If left untreated, breathing problems, seizures, and coma sometimes leading to death can occur.

Treatment requires a lifelong protein restricted diet. Medication and supplements may be prescribed. If treated early, organ damage and neonatal death can be prevented.