

# 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.