

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn
Screening Program



Elevated Tyrosine with Normal 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.
- **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

Screening result can be impacted by a neonate with an immature liver or liver disease; found more commonly in premature infants and those with jaundice.

Differential Diagnosis

Elevated tyrosine with normal succinylacetone is primarily associated with:

- Transient tyrosinemia of the newborn — common
- Tyrosinemia type 2 (TYR II) — Incidence of 1 in 250,000
- Tyrosinemia type 3 (TYR III) — only a few cases reported

Clinical Summary

Tyrosinemia occurs when the body is unable to break down the amino acid, tyrosine leading to a buildup in the blood.

The most likely reason for this screening result is transient tyrosinemia of the newborn. Up to 10 percent of newborns have transient tyrosinemia, likely due to vitamin C deficiency or immature liver enzymes from premature birth. It is benign and resolves without sequelae.

There are three types of tyrosinemia (I, II, and III) disorders. Unlike tyrosinemia types 2 and 3, tyrosinemia type 1 has elevated succinylacetone, which is pathognomonic for that type.

Tyrosinemia type 2 affects the skin and eyes. If untreated affected infants develop photophobia, eye redness, skin lesions, and poor coordination. Tyrosinemia type 3 is the least common type and symptoms are highly variable and not well known. Symptoms may include poor coordination and epilepsy. Treatment for both types may include a protein restricted diet and medication. If treated early, children can have healthy growth and development.