

Positive Result:

Blood Spot Screen Result Notification



Elevated Thyroid Stimulating Hormone (TSH)

Next Steps

Today, you should take the following recommended actions:

- **Contact** family to notify them of the newborn screening result, assess symptoms, and arrange for collection of labs.
- **Evaluate** infant (jaundice and lethargy); arrange immediate referral if symptomatic.
- **Order** serum TSH and free T4 (thyroxine).
- **Fax** TSH and free T4 results to MDH program staff at (651) 215-6285.

If thyroid function studies are abnormal, a pediatric endocrinologist should be contacted immediately. Contact information for endocrinologists can be found on the resource list provided.

If thyroid function studies are normal (using age-appropriate cutoffs), no additional follow-up is required.

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.

False Positives

Screening result can be impacted by specimen collection before 24 hours, maternal thyroid disease, and prematurity.

Differential Diagnosis

Elevated TSH is primarily associated with:

- Primary congenital hypothyroidism — Incidence of 1 in 3,000

Other disorders to consider:

- Secondary hypothyroidism
- Transient hypothyroidism

Clinical Summary

Congenital hypothyroidism is an endocrine disorder that can result from abnormal development of the thyroid, ectopic thyroid, or the inability of the thyroid gland to produce adequate hormone levels. TSH levels are higher in newborns than in older children and adults; abnormal values can be significantly elevated.

Newborns are typically asymptomatic. Some infants may exhibit clinical features such as prolonged jaundice and sleepiness. If an infant is not screened and/or left untreated, symptoms begin to appear later in infancy and can include developmental delay, intellectual disability, and poor growth.

Affected children are likely to require thyroid hormone replacement therapy and monitoring by both primary care and specialty providers. If treated promptly, children with congenital hypothyroidism can be asymptomatic and are expected to develop normally.