

Borderline Result:

Blood Spot Screen Result Notification



Mildly Elevated Thyroid Stimulating Hormone (TSH)

Next Steps

This week, 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.
- **Order** serum TSH and free T4 (thyroxine) — a repeat newborn screen is an acceptable alternative to collecting these clinical labs.
- **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.

Possible Explanations for Result

Screening result can be impacted by specimen collection before or around 24 hours of age, stress, physiologic changes, maternal thyroid disease, and prematurity.

Additionally, screening result may be due to congenital hypothyroidism (primary, secondary, or transient). Congenital hypothyroidism is more common in children with congenital anomalies and certain genetic syndromes (e.g., Down syndrome).

Clinical Summary

Most borderline results resolve on retesting.

In the event that thyroid function studies are abnormal, it is important to be aware of the clinical presentation. 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.

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.

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.

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Borderline Elevation of Thyroid Stimulating Hormone (TSH)

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has slightly high TSH levels.

What does this mean?

Borderline elevations of TSH are common and your baby is likely healthy. It is possible, however, that your child may have congenital hypothyroidism (CH). CH is a type of thyroid disease. A borderline result does not mean your baby has thyroid disease, but more testing is needed to know for sure.

What happens next?

Your baby's doctor will order more testing. Most likely there will be two labs drawn at your baby's clinic. These two labs are called TSH and free T4 (thyroxine). It is possible they may just order a second newborn screen. Most retesting comes back normal.

If the results from this additional testing are normal, no more testing or treatment will be needed.

If the results from this additional testing remain elevated, your baby's doctor will let you know what additional testing or treatment is needed. You should be referred to a pediatric endocrinologist who cares for children with thyroid disease.

What is congenital hypothyroidism (CH)?

CH is a condition present in a baby at birth that affects the body's thyroid gland, a small organ in the lower neck. The thyroid gland makes thyroid hormone. People with CH are not able to make enough thyroid hormone. Thyroid hormone is needed for healthy growth and development.

What health problems can it cause?

CH is a lifelong condition. If untreated, it can cause:

- Jaundice (yellow skin or whites of the eyes)
- Weak muscle tone
- Learning disabilities
- Delayed growth and weight gain

Children with CH can benefit from prompt and careful treatment.

What treatment options are available?

Although CH cannot be cured, it can be treated. The most common treatment for CH is a medication that replaces the thyroid hormone that isn't being made correctly. If treated before symptoms develop, children can have healthy growth and development.

Children with CH should see their regular doctor and a doctor who specializes in thyroid disease (an endocrinologist).

Resources

Genetics Home Reference:
<http://ghr.nlm.nih.gov>

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org