

Minnesota Department of Health

Borderline Result: Congenital Hypothyroidism

MEDICAL FACT SHEET BORDERLINE NEWBORN SCREEN

Action Required

Order laboratory evaluation this week

Laboratory Evaluations Needed:

Repeat Newborn Screen

OR

Serum TSH (Thyroid Stimulating Hormone) and free T4 (Thyroxine)

Follow-up required if:

- Thyroid function studies are within normal limits, no additional follow-up required
- Thyroid function studies or repeat screen are abnormal, contact Pediatric Endocrinologist for guidance with evaluation and treatment

Borderline Results:

- Common
- TSH is elevated in the newborn period
- Screening result can be impacted by specimen collection less than 24 hours, stress, physiologic changes, maternal thyroid disease, and prematurity

Review with Family

Family has **not** been notified of result by MDH.

Most borderline results are normal on clinical testing. Expect infant to be stable when family is contacted and at clinic visit.

NICU Issues

Premature infants are likely to show delayed abnormalities on newborn screening. For infants < 1800g, all 3 specimens should be reviewed to exclude risk for thyroid disease. Babies with congenital abnormalities and Down Syndrome are at increased risk for hypothyroidism.

Newborn screens cannot be accurately interpreted if collected before 24 hours.

Clinical Summary

Children with borderline results found to be normal on subsequent testing require no special treatment.

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 thyroid hormones.

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

Affected children require life-long thyroid hormone replacement and monitoring by both primary care and specialty providers.

Incidence: ~ 1/3,000; affects all ethnic groups

Clinical Expectations

Prompt follow-up and interventions are critical if repeat testing is abnormal. If treated promptly, children with congenital hypothyroidism can be asymptomatic and are expected to develop normally. Affected children should be monitored for proper height and weight gain and developmental progression.

Resources

GeneTests: www.genetests.com

OMIM: www.ncbi.nlm.nih.gov/omim/

ACT Sheets: www.acmg.net/resources/policies/ACT/condition-analyte-links.htm

MN Newborn Screening Program:
www.health.state.mn.us/newbornscreening