

Congenital Hypothyroidism

MEDICAL FACT SHEET POSITIVE NEWBORN SCREEN

Action required

Contact pediatric endocrinologist today.
See infant today.

Issues to discuss with endocrinologist

- Laboratory evaluation of infant
 - § Should testing be performed by primary care or endocrinology clinic
 - § TSH/free T4

False Positives

- Common
- Screening result can be impacted by specimen collection less than 24 hours, maternal thyroid disease, and prematurity

Review with family

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

After discussion with pediatric endocrinologist, contact family to coordinate clinic visit, lab work, and referral to endocrinology clinic, if necessary. Expect infant to be stable when family is contacted and at clinic visit.

Prompt follow-up and intervention are critical.

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 anomalies and Down Syndrome are at increased risk for hypothyroidism.

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

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 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

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.org

OMIM: www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM

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

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