



Newborn Screening ACT Sheet

[Primary T4-follow-up TSH test/ Low T4 and/or elevated TSH]

Congenital Hypothyroidism

Differential Diagnosis: Primary and secondary congenital hypothyroidism (CH), transient CH, thyroxine binding globulin (TBG) deficiency.

Condition Description: Lack of adequate thyroid hormone production.

You Should Take the Following Actions:

- Contact family **IMMEDIATELY** to inform them of the newborn screening test result.
- Consult pediatric endocrinologist; referral to endocrinologist if considered appropriate.
- Evaluate infant (see clinical considerations below).
- Initiate timely confirmatory/diagnostic testing as recommended by the specialist.
- Initiate treatment as recommended by consultant as soon as possible.
- Educate parents/caregivers that hormone replacement prevents mental retardation.
- Report findings to State newborn screening program.

Diagnostic Evaluation: Diagnostic tests should include serum **free T4** and **thyroid stimulating hormone (TSH)**; consultant may also recommend **total T4 and T3 resin uptake**. Test results include **reduced free T4 and elevated TSH** in primary hypothyroidism. **TSH is reduced or inappropriately normal** in secondary (hypopituitary) hypothyroidism. **Low total T4 and elevated T3 resin uptake** are consistent with TBG deficiency.

Clinical Considerations: Most neonates are asymptomatic, though a few can manifest some clinical features, such as prolonged jaundice, puffy facies, large fontanel, macroglossia and umbilical hernia. Untreated congenital hypothyroidism results in developmental delay or mental retardation and poor growth.

Additional Information:

(Click on the name to take you to the website. Complete URLs are listed in the Appendix)

[New England Newborn Screening Program](#)

[American Academy of Pediatrics](#)

[Genetics Home Reference](#)

Referral (local, state, regional and national):

[Lawson Wilkins Pediatric Endocrine Society “Find A Doc”](#)

Contact local/regional University-affiliated medical center for referrals to Pediatric Endocrinologists.

Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines.

State Resource Site *(insert state newborn screening program website information)*

Name
URL
Comments

Local Resource Sites *(insert local and regional newborn screening website information)*

Name
URL
Comments

Name
URL
Comments

Name
URL
Comments

APPENDIX: Resources with Full URL Addresses

Additional Information:

New England Newborn Screening Program

<http://www.umassmed.edu/nbs/screenings/disorders/hypothyroidism.cfm>

American Academy of Pediatrics <http://pediatrics.aappublications.org/cgi/content/abstract/91/6/1203>

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

Referral (local, state, regional and national):

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