

## Newborn Screening ACT Sheet [Low T4 and/or elevated TSH (Primary T4 follow-up TSH test)] 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 to inform them of the newborn screening test result.
  - Consult pediatric endocrinologist; refer 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 developmental delay.
  - Report findings to 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 and poor growth.

### **Additional Information:**

[American Academy of Pediatrics  
Genetics Home Reference](#)

### **Referral (local, state, regional and national):**

[Testing](#)

Clinical Services

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

[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It 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. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site (insert state newborn screening program website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

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

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

Additional Information:

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):

Testing

[http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical\\_disease\\_id/20744?db=genetests&country=United%20States](http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/20744?db=genetests&country=United%20States)

Clinical Services

Lawson Wilkins Pediatric Endocrine Society "Find A Doc"

<http://lwpes.org/>

Find Genetic Services

<http://www.acmg.net/GIS/Disclaimer.aspx>

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It 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. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)