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 fontanels, 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.

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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):

Lawson Wilkins Pediatric Endocrine Society "Find A Doc" http://lwpes.org/

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

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