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 mental retardation.
- 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 fontanels, macroglossia and umbilical hernia. Untreated congenital hypothyroidism results in developmental delay or mental retardation 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

LOCAL RESOURCES: Insert State newborn screening program web site links

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

Name	
URL	
Comments	

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

Name	
URL	
Comments	

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

Clinical Services

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

Find Genetic Services
http://www.acmg.net/GIS/Disclaimer.aspx