American College of Medical Genetics ACT SHEET

Newborn Screening ACT Sheet [Elevated TSH (Primary TSH test)] Congenital Hypothyroidism

Differential Diagnosis: Primary congenital hypothyroidism (CH); transient CH.

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 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; if done, reduced total T4 and low or normal T3 resin uptake

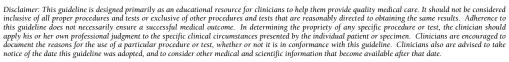
Clinical Considerations: Most neonates are asymptomatic, though a few can manifest some clinical features, such as prolonged jaundice, puffy facies, large fontanels, macroglossia and umbilical hernia. Untreated congenital hypothyroidism results in developmental delay and poor growth.

Additional Information:

<u>American Academy of Pediatrics</u> Genetics Home Reference

Referral (local, state, regional and national):

Testing
Clinical Services
Lawson Wilkins Pediatric Endocrine Society "Find A Doc"
Find Genetic Services





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LOCAL RESOURCE	S: Insert State newborn screening program web site links
State Resource s	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: Resoui	rces with Full URL Addresses
Additional Informati American Acade	
http://pediatrics.	aappublications.org/cgi/content/full/117/6/2290?maxtoshow=&HITS=10&hits=10&RESULTFORMAT
	nital+hypothyroidism&searchid=1&FIRSTINDEX=0&sortspec=relevance&resourcetype=HWCIT
Genetics Home l	Reference h.gov/condition=congenitalhypothyroidism
Defenuel (legal etate	usai sual and national).
Testing	, regional and national):
http://www.ncbi.	nlm.nih.gov/sites/GeneTests/lab/clinical disease id/20744?db=genetests&country=United%20Statezs
Clinical Services	lkins Pediatric Endocrine Society "Find a Doc"
http://lwpes	
Find Genetic Se	rvices

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.



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