ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

ACT Sheet

Newborn Screening ACT Sheet

[Low to low normal Thyroxine (T4) and low to normal Thyroid Stimulating Hormone (TSH)]

Central Congenital Hypothyroidism

Differential Diagnosis: Pituitary or hypothalamic abnormality; transient congenital hypothyroidism; maternal hyperthyroidism; thyroid hormone binding globulin (TBG) deficiency; prematurity.

Condition Description: Central hypothyroidism is a rare disorder caused by pituitary or hypothalamic abnormalities. It is associated with low T4 levels in the setting of inappropriately low TSH levels. It can be isolated or can be associated with hormone deficiencies of the anterior pituitary gland (growth hormone deficiency, ACTH deficiency) or the posterior pituitary gland (diabetes insipidus). It can also occur with maternal hyperthyroidism during pregnancy.

You Should Take the Following IMMEDIATE Actions:

- Inform family of the newborn screening result.
- Ascertain clinical status (newborns can be lethargic and have feeding difficulties).
- Consult with pediatric endocrinologist the same day.
- Evaluate the newborn (jaundice, midline defects, cleft palate, phallic size). Assess for hypoglycemia, hypothermia.
- Initiate confirmatory testing and management in consultation with a pediatric endocrinologist.
- Provide family with basic information about congenital hypothyroidism.

- Report final diagnostic outcome to the state newborn screening program.

Diagnostic Evaluation: Thyroid Stimulating Hormone (TSH) low to normal with low or normal total T4/free T4, total T3 levels: can confirm the diagnosis. Thyroxine Binding Globulin (TBG) Testing : TBG deficiency should be considered if TSH levels are normal, T4 levels are low, and free T4 levels are normal. Growth hormone, prolactin, LH, FSH, and cortisol: should be performed if pituitary dysfunction is suspected.

Clinical Considerations: Congenital hypothyroidism is the most preventable cause of intellectual disability. Most newborns are asymptomatic. In some infants, congenital hypothyroidism is transient. Central congenital hypothyroidism can be isolated or associated with multiple pituitary hormone deficiencies. Infants require prompt therapy with appropriate doses of levothyroxine. If additional pituitary deficiencies are present, additional hormone replacement is indicated.

Additional Information:

How to Communicate Newborn Screening Results
Medline Plus
Pediatric Endocrine Society
Clinicaltrials.gov

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

Find a Genetics Clinic Directory
Genetic Testing Registry
Find a Pediatric Endocrinologist

 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957;#UH9MC30770; National Coordinating Center for the Regional Genetics Networks*)*

Local Resources *(Insert Local Website Links)*

State Resource Site (Insert Website Information)

Name	
URL	
Comments	

Local Resource Site (Insert Website Information)

Name	
URL	
Comments	

Appendix *(Resources with Full URL Addresses)*

Additional Information

How to Communicate Newborn Screening Results
- https://bit.ly/NBSResultsHRSA

Gene Reviews

Medline Plus
- https://medlineplus.gov/genetics/condition/congenital-hypothyroidism/

Clinicaltrials.gov
- https://clinicaltrials.gov/

Pediatric Endocrine Society
- https://pedsendo.org/clinical-resource/infant-with-suspected-congenital-hypothyroidism/

Referral (local, state, regional and national)

Find a Genetics Clinic Directory
- https://clinics.acmg.net

Genetic Testing Registry
- https://www.ncbi.nlm.nih.gov/gtr/

Find a Pediatric Endocrinologist
- https://pedsendo.org/patient-resources/find-a-pediatric-endocrinologist/

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