



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

This practice resource is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this practice resource is completely voluntary and does not necessarily assure a successful medical outcome. This practice resource 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. 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 practice resource. Clinicians also are advised to take notice of the date this practice resource was adopted, and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.



ACT Sheet

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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