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

ACT Sheet

Newborn Screening ACT Sheet

[Elevated Thyroid Stimulating Hormone (TSH) with or without low Thyroxine (T4) level]

Primary Congenital Hypothyroidism

Differential Diagnosis: Thyroid dysgenesis (agenesis, ectopic, or hypoplastic thyroid); maternal antithyroid medications; maternal antithyroid antibodies; iodine deficiency, excess iodine exposure; transient hypothyroidism.

Condition Description: Congenital hypothyroidism (CH) is a deficiency of thyroid hormone present at birth. It may be primary or central. In the United States, primary hypothyroidism is the most common form and is most often due to abnormal development of the thyroid gland. It is associated with elevated levels of TSH and normal or low levels of circulating thyroid hormones. The majority of newborns are asymptomatic. Therapy is essential to allow for normal growth and intellectual development.

You Should Take the Following IMMEDIATE Actions:

- Inform family of the newborn screening result.
- Ascertain clinical status (newborns are usually asymptomatic but can be lethargic and have feeding difficulties).
- Consult with pediatric endocrinologist the same day.
- Evaluate the newborn (newborns are generally asymptomatic; assess for jaundice, macroglossia, or umbilical hernia). Assess for hypoglycemia, hypothermia; perform a clinical cardiac examination given the increased risk of cardiac anomalies.
- Initiate confirmatory testing and management under the direction of a pediatric endocrinologist.
- Provide family with basic information about hypothyroidism.

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

Diagnostic Evaluation: Thyroid Stimulating Hormone (TSH) elevation in the setting of low or normal circulating thyroid hormone levels (total T4/free T4, total T3): can confirm the diagnosis. Radionuclide scans and/or thyroid ultrasound: may identify thyroid agenesis, or hypoplastic or ectopic thyroid tissue. Anti-TSH receptor antibodies: may antagonize the action of TSH and cause hypothyroidism.

Clinical Considerations: Congenital hypothyroidism is the most preventable cause of intellectual disability. Most newborns are asymptomatic. Infants require prompt therapy with appropriate doses of levothyroxine. In some infants, congenital hypothyroidism is transient.

Additional Information:

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

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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