TEXAS Health and Human Services
Texas Department of State Health Services
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Interim Commissioner

Newborn Screening ACT Sheet
Congenital Hypothyroidism (CH)

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 AND CONDUCT AN IMMEDIATE CLINICAL INFANT ASSESSMENT:** Possible signs include jaundice, poor feeding, vomiting, constipation, excessive sleepiness, hypotonia, cool extremities, bradycardia, and macroglossia. Note: The absence of symptoms does **NOT** rule out disease
- ☐ **IMMEDIATE PHONE CONSULT AND REFERRAL TO A PEDIATRIC ENDOCRINOLOGIST:** See attached list
- ☐ **ORDER and COMPLETE THYROID LABS: TSH, Free T4, Total T4 and any other testing recommended by the specialist** within **ONE** business day. **DO NOT** wait for the result of repeat screen before obtaining serum labs.
- ☐ **INITIATE TREATMENT as per pediatric endocrinologist's guidance AS SOON AS POSSIBLE and ducate parents/caregivers that hormone replacement PREVENTS INTELLECTUAL DISABILITY and infant will need to take daily.**
- ☐ **Repeat the newborn screen at age 7 to 14 days old (if not already completed):** Thyroid labs are needed even if the second newborn screen is already submitted.
- ☐ **Fax Thyroid serum lab results to: (512) 206-3907**

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 faces, large fontanels, macroglossia, and umbilical hernia. Untreated congenital hypothyroidism results in developmental delay or intellectual disability and poor growth.

Additional Information:
American Academy of Pediatrics
http://pdoi.org/10.1542/peds.2022-060420

Disclaimer: This information is adapted from American College of Medical Genetics website ACT sheets.

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