TEXAS Health and Human Services | Texas Department of State Health Services

Newborn Screening ACT Sheet

Congenital Hypothyroidism

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 IMMEDIATELY** to inform them of the newborn screening test result.
- **Consult pediatric endocrinologist;** refer to endocrinologist if considered appropriate.
  (See attached list.)
- **Evaluate infant** (see clinical considerations below).
- **Initiate timely confirmatory/diagnostic testing** as recommended by the specialist.
- **Initial testing should include Serum TSH and Free T4.**
- **Repeat newborn screen on day of life 7-14** if second screen has not yet been done.
- **Initiate treatment as recommended by consultant as soon as possible.**
- **Educate parents/caregivers** that hormone replacement **prevents intellectual disability.**
- **Report findings** to 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. 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, poor feeding, poor tone, and umbilical hernia. Untreated congenital hypothyroidism results in developmental delay or intellectual disability and poor growth.

Additional Information:
Genetics Home Reference
Congenital hypothyroidism: MedlinePlus Genetics