



## **Newborn Screening ACT Sheet [Primary TSH test/Elevated TSH] Congenital Hypothyroidism**

### **Differential Diagnosis:**

- Primary congenital hypothyroidism (CH);
- Transient CH.

### **Condition Description:**

- Lack of adequate thyroid hormone production.

### **MEDICAL EMERGENCY: TAKE THE FOLLOWING IMMEDIATE ACTIONS:**

- Contact family IMMEDIATELY to inform them of the newborn screening test result.
- Consult pediatric endocrinologist; referral to endocrinologist if considered appropriate.
- Evaluate infant (see clinical considerations below).
- Initiate timely confirmatory/diagnostic testing as recommended by the specialist.
- Initiate treatment as recommended by consultant as soon as possible.
- Educate parents/caregivers that hormone replacement prevents mental retardation.
- Report findings to state newborn screening program.

*(See flowchart for information concerning the specific actions)*

### **You Should Take the Following Actions:**

#### **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; if done, reduced total T4 and low or normal T3 resin uptake.

#### **Clinical Considerations:**

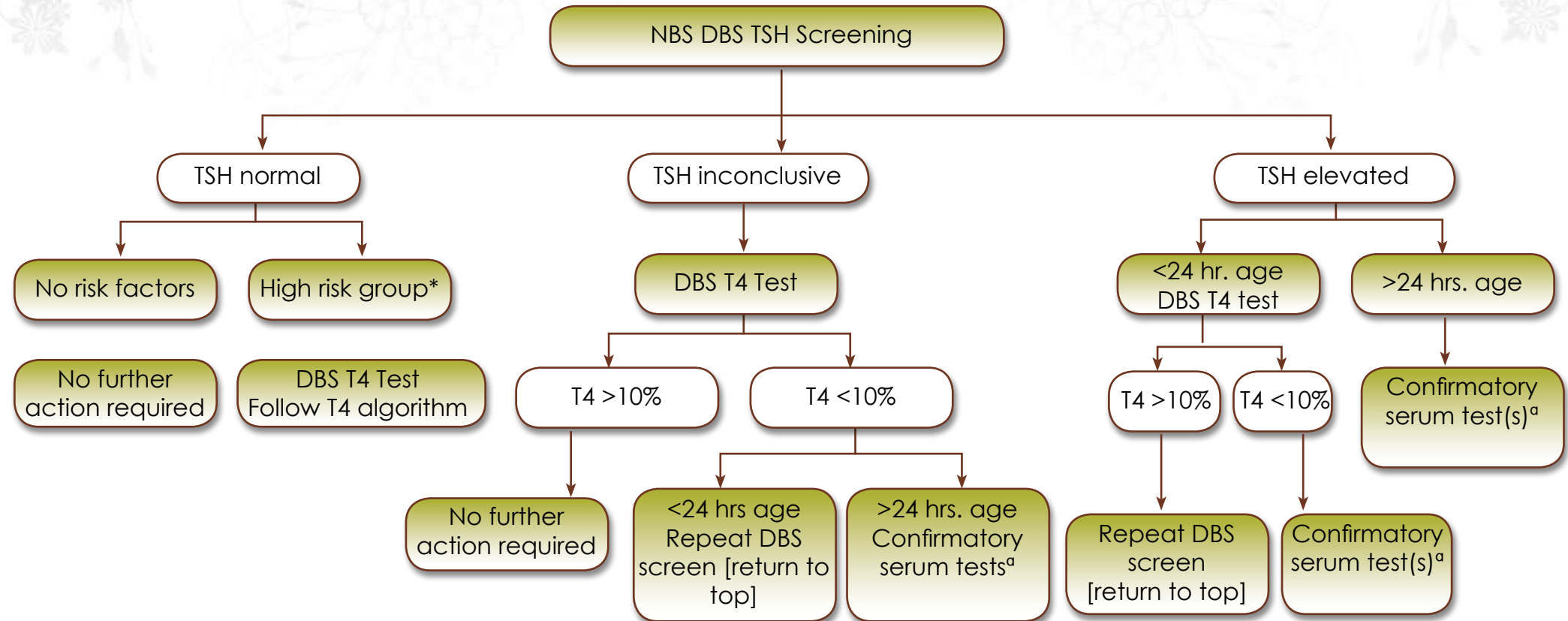
Most neonates are asymptomatic, though a few can manifest some clinical features, such as prolonged jaundice, puffy facies, large fontanelles, macroglossia and umbilical hernia. Untreated congenital hypothyroidism results in developmental delay or mental retardation and poor growth.

*Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality medical services. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen.*





**Congenital Hypothyroidism (TSH)**





Actions are shown in shaded boxes; results are in the unshaded boxes.

Abbreviations/Key

NBS = Newborn Screening

DBS = Dried blood spot

TSH = Thyroid stimulating hormone

T4 = Thyroxine or total thyroxine

\* High risk group

<1500 gm

NICU admission

Same sex twin

Transfusion

CHD/other severe congenital anomaly

Drugs: dopamine, steroids, iodine

<sup>a</sup> Confirmatory Serum Tests

Free T4 [or]

4 and T3 resin uptake (T3RU)

TSH

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