



KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: ELEVATED TSH

CONDITION: CONGENITAL HYPOTHYROIDISM (CH)

DIFFERENTIAL DIAGNOSIS: Primary congenital hypothyroidism; secondary congenital hypothyroidism; transient CH; thyroxine binding globulin (TBG) deficiency.

METABOLIC DESCRIPTION: Lack of adequate thyroid hormone production.

ACTION TO BE TAKEN IMMEDIATELY:

- ◆ Contact family immediately to inform them of the newborn screening test results.
- ◆ 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 intellectual disabilities.
- ◆ Report findings to newborn screening program.

CONFIRMATION OF DIAGNOSIS: 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 EXPECTATIONS: Most neonates are asymptomatic, though a few can manifest some clinical features, such as prolonged jaundice, puffy facies, large fontanel, macroglossia and umbilical hernia. Untreated congenital hypothyroidism results in developmental delay or intellectual disabilities and poor growth.

REPORTING: Report diagnostic result to family and Kansas NBS program.

SPECIALISTS:

KU Medical Center	Kansas City KS	913-588-6326
Children’s Mercy	Wichita, KS	316-500-8900
Wichita Endocrinology	Wichita, KS	316-777-6404
Cotton-O’Neil Clinic	Topeka, KS	785-368-0460
Pediatric Endocrinology	Lawrence, KS	785-505-2551

DISCLAIMER: These standards and guidelines were adapted from the American College of Medical Genetics ACT sheets. They are designed primarily as an educational resource for physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonable directed to obtaining the same results. 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. It may be prudent, however, to document in the patient’s record the rationale for any significant deviation from these standards and guidelines