KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: ELEVATED TSH

CONDITION: CONGENITAL HYPOTHYDROIDISM (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 fontanels, 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