



PHYSICIAN ACT SHEET for POSITIVE Newborn Screening Result for CONGENITAL PRIMARY HYPOTHYROIDISM

Disease Category: Endocrinopathy

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- **Contact family to inform them of the newborn screening result and schedule an immediate visit.**
- **Schedule a lab visit for the newborn within 24-48 hours for confirmatory testing.**
- **Consult pediatric endocrinologist.** Refer to an endocrinologist if considered appropriate.
- Report findings back to state newborn screening program.

Pediatric endocrinologists are available at Children's Hospital: Dr. Corley, Dr. Cabrera, and Dr. Des Mangles (402) 955-3871

Meaning of the Screening Result: Decreased thyroxine (T4) accompanied by increased thyroid stimulating hormone (TSH) suggests primary hypothyroidism.

Confirmation of Diagnosis: Diagnostic tests include serum Free T4 which will be reduced, and serum TSH which will be elevated. Be sure specimen is tested at a laboratory with established neonatal reference ranges. Serum T3 uptake may be recommended. The pediatric endocrinologist may also recommend radiographs.

Condition Description: The baby lacks adequate thyroid hormone production.

Clinical Expectations: If untreated, results in developmental delay/mental retardation and poor growth.

Additional Information:

- New England Metabolic Consortium – Emergency Protocols - www.childrenshospital.org/newenglandconsortium
- Gene Tests/Gene Clinics - www.genetests.org
- U.S. National Newborn Screening & Genetics Resource Center – www.genes-r-us.uthscsa.edu