



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 office visit for the newborn within 24-48 hours for confirmatory testing.
- Consult pediatric endocrinologist. Referral to endocrinologist if considered appropriate.
- Report findings back to state newborn screening program.

Pediatric endocrinologists are available at:

- Children's Hospital: Dr. Cabrera and Dr. Des Mangles (402) 955-3871
- Nebraska Medical Center: Dr. Corley and Dr. Lutz (402) 559-9197

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

Metabolic Description: Lack of adequate thyroid hormone production.

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.

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