



**State of CT**  
**Genetics Newborn Screening Program**  
**Health Professional Fact Sheet**

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## **CONGENITAL HYPOTHYROIDISM (CH)**

Congenital hypothyroidism (CH) results from inadequate production of the thyroid hormone, which may be due to a number of causes: agenesis, ectopic thyroid gland, or inherited disorders of thyroid hormone biosynthesis. Patients who are not identified and treated promptly may have mental retardation, growth failure, and other neurologic abnormalities, as well as clinical symptoms such as prolonged neonatal jaundice, constipation, and lethargy. Screening in Connecticut was instituted in 1976.

### **GENETIC BASIS**

- Most cases are sporadic (thyroid agenesis/ectopic lingual)
- The remaining cases are disorders of thyroid hormone biosynthesis which are usually inherited autosomal recessive conditions.

**CT PREVALENCE** 1:4,070

### **CLINICAL FEATURES OF UNTREATED DISEASE**

#### **Severity**

More than 95% of infants with sporadic hypothyroidism show such minimal signs at birth that the diagnosis is missed.

#### **Symptomatic diagnosis**

A symptomatic diagnosis is difficult because clinical signs of congenital hypothyroidism such as jaundice, constipation, lethargy, and feeding problems are non-specific and are found in fewer than 30% of neonates with CH. When clinical symptoms do appear, it is usually not until the infant is 3 months of age or older. Infants with early clinical findings seem to have a higher incidence of developmental disabilities.

### **CLINICAL OUTCOME WITH SCREENING AND TREATMENT**

#### **Variability**

Early and adequate treatment and close, careful monitoring is important and needs to be delivered to prevent permanent retardation of intellectual function and/or skeletal growth. In those children that have been identified and treated early and adequately and who continue to receive adequate care, their neurological development is comparable to their peers.

#### **Interventions**

Pediatric endocrinologic consultation is advisable at one of the Endocrine Regional Treatment Centers. Oral levothyroxine should be given at a dosage to produce a T4 concentration in the upper normal range to normalize TSH levels. Growth and development must be monitored at frequent intervals, including measurement of thyroid hormone levels to prevent both under and over treatment and their associated morbidities.

**WARNING: REPLACEMENT LEVOTHYROXINE SHOULD NOT BE ADMINISTERED WITH SOY  
BASED FORMULA. SOY BINDS THE LEVOTHYROXINE AND PREVENTS ABSORPTION.**

### **SCREENING FOR CH**

#### **Laboratory tests**

Test for TSH on filter paper blood specimen utilizing fluorometric immunoassay.

## **ABNORMAL TEST RESULTS**

### **TSH levels of > 99.9**

- The primary care provider is notified by the DPH, Genetic NBS Tracking Unit nurse consultants.
- A referral is made by the DPH, Genetic NBS Tracking Unit nurse consultant to one of the Endocrinology Regional Treatment Centers at Connecticut Children's Medical Center or Yale.
- The primary care provider will be advised by the DPH, Genetic NBS Tracking Unit nurse consultant to contact the Endocrinology Regional Treatment Center to make a prompt referral and arrange for confirmation testing and evaluation.

### **TSH levels of 40 - 99.9**

DPH, Genetic NBS Tracking Unit nurse consultants notify the primary care provider's office of results and the need to obtain a second specimen.



Keeping Connecticut Healthy

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