

The New Jersey Department of Health and Senior Services

Congenital Hypothyroidism (CH)

Information for Health Professionals

Description

Congenital hypothyroidism (CH) represents one of the most common preventable causes of mental retardation. It occurs when infants are unable to produce sufficient amounts of thyroid hormone (thyroxine, or T4), which is necessary for normal metabolism, growth and brain development. Causes of CH may be sporadic or familial, goitrous or nongoitrous. The most common causes of CH are total or partial failure of the thyroid gland to develop (aplasia or hypoplasia), or its development in an ectopic location.

Incidence

The prevalence of congenital hypothyroidism has been found to be 1:4,000 infants worldwide; lower in African-Americans (1:20,000). It is twice as common in girls as in boys.

Clinical Features

Most infants with CH are asymptomatic at birth. They may appear clinically normal up to three months of age. By that time, if treatment has not been instituted, some brain damage has usually occurred. Birth weight and length are normal, but head size may be slightly increased. Affected infants have a decreased, hoarse cry, increased sleep, decreased muscle tone and feed poorly. There may be prolonged neonatal jaundice, constipation, a distended abdomen, and umbilical hernia. Many infants present with a large fontanelle and a puffy face. The temperature is subnormal and the skin may be cold and mottled. Edema of the genitals and extremities may be present. The pulse is slow, and heart murmurs, cardiomegaly, and asymptomatic pericardial effusion are common.

Infants with CH appear to be at increased risk for congenital anomalies, which occur in approximately 10% of these infants as compared to 3% in the general population. Cardiovascular anomalies are the most common association; these include pulmonary stenosis, atrial septal defect, and ventricular septal defect

Screening

New Jersey has a two-tier screening protocol for CH. Measurement of the thyroxine (T4) level is the initial screening test, which is performed on a dried blood spot on filter paper. All specimens in the lowest 10th percentile for T4 are re-assayed for thyroid stimulating hormone (TSH) concentration.

A premature or sick newborn may have a physiological reduction in blood T4 levels; the TSH levels are usually normal. Premature infants have about the same incidence of hypothyroidism as full term infants. **Repeat T4 and TSH screenings should be done to determine whether abnormal values are related to true thyroid dysfunction or are secondary to prematurity or neonatal illness.**

Regardless of newborn screening results, if clinical signs and symptoms suggest hypothyroidism, serum T4 and TSH analysis should be carried out to rule out the possibility of late-onset disease.

Confirmatory Testing

All infants with abnormal screening results need to have confirmatory testing. An infant with a low T4 level and an elevated TSH concentration (greater than 40 uIU/ml) is considered to have primary hypothyroidism until proven otherwise. Infants with elevated TSH results should be immediately examined and have confirmatory serum testing to verify the diagnosis.

Treatment

Treatment of CH is simple and effective, but should be made in consultation with a pediatric endocrinologist. Sodium L-thyroxine given orally is the treatment of choice. It must be in tablet form as liquid preparations are not stable. Tablets should be crushed and dissolved in a teaspoon of water or less and administered by spoon or dropper. Families should be advised NOT to administer the medication by bottle or through a nipple. Frequent laboratory evaluations of thyroid function ensure optimal thyroxine dosage.

Interpretations/Recommendations:

- **Expected Results: (Age > 24 hours) Normal T4 level with TSH not tested or normal TSH (<20 µIU/ml)**
- **Initial Borderline Results: (Any age) Low T4 level with normal TSH (<20 µIU/ml); repeat filter paper specimen within 2 days**
- **Initial Borderline Results: (Any Age) Normal or Low T4 level with elevated TSH (20–40 µIU/ml)
Recommend: obtain serum specimen within 2 days (T4, free T4, and TSH assay preferred)**
- **Repeat Borderline Results: Any baby with repeat borderline abnormal results should be immediately evaluated; obtain serum T4, free T4 and TSH within 2 days; consultation with a pediatric endocrinologist is strongly recommended**

Interpretations/Recommendations (cont.):

- **Presumptive-Positive Results:** (Any Age) Normal or Low T4 level with elevated TSH (>40 uIU/ml)
Recommend: **Immediate** examination of baby; **immediate** consultation with pediatric endocrinologist; serum T4, free T4, and TSH testing should be performed.

Note: Newborn screening tests are an adjunct to clinical assessment, which is paramount. CH should be considered in infants with any of the signs.

Additional Information:

eMedicine

<http://www.emedicine.com/ped/topic501.htm>

Illinois Department of Public Health Newborn Screening Program

http://www.idph.state.il.us/HealthWellness/fs/congenital_hypo.htm

Texas Department of State Health Services Newborn Screening

http://www.tdh.state.tx.us/newborn/p_thyroi.htm

For questions, contact:

Inborn Errors of Metabolism Laboratory at (609) 292-3090

Newborn Screening and Genetic Services at (609) 292-1582

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