



**NEWBORN SCREENING
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Congenital Hypothyroidism

Information for Physicians and Other Health Care Professionals

Definition Congenital hypothyroidism (CH) 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.

Clinical Symptoms Although the clinical signs of hypothyroidism may be subtle, infants with CH may exhibit some of the following symptoms: feeding problems, lethargy, prolonged postnatal jaundice, delayed stooling and constipation, enlarged protruding tongue, hoarse cry, protruding abdomen with an umbilical hernia, cold mottled skin, sluggish reflexes, patent posterior fontanelle with widely spread cranial sutures or delayed skeletal maturation for gestational age.

Newborn Screening and Definitive Diagnosis In Illinois, primary newborn screening for CH utilizes fluorometric assay to determine the thyroid stimulating hormone (TSH) level. If the TSH is elevated, the T4 level also is tested. False positive and false negative results are possible with this screening. Specimen collection prior to 24 hours of age, prematurity and illness can affect this screening. **Infants with a presumptive positive screening test (seriously elevated TSH and/or low T4) require prompt follow-up and, when notified of these results, the clinician should *immediately* check on the clinical status of the baby and refer the infant to a pediatric endocrinologist. Collection of serum TSH and T4 level also is recommended.** Suspect abnormal results (moderately elevated TSH) indicate the need for repeat filter paper screening. Nearly 90 percent of CH cases are detected by newborn screening; however, the remaining 10 percent must be detected clinically. A small number of children may test normal on the newborn screen but later develop hypothyroidism. Clinicians must remain alert to signs indicative of possible hypothyroidism and clinical symptoms and/or family history of thyroid disorders indicate the need for thyroid testing, regardless of newborn screening results. Same birth siblings (twins, triplets) of infants diagnosed with CH should be re-screened; additional testing of these siblings also may be indicated.

Treatment Immediate diagnosis and treatment of congenital hypothyroidism in the neonatal period is critical to normal brain development and physical growth. Treatment is usually effective if started within the first few weeks of life. Delayed treatment may result in decreased intellectual capacity. Recommended treatment is lifetime daily administration of levo-thyroxine. Only the tablet form of levo-thyroxine should be prescribed. The U.S. Food and Drug Administration has not approved liquid suspensions. Suspensions prepared by pharmacists may lead to unreliable dosage. The tablets should be crushed daily, mixed with a few milliliters of water, formula or breast milk and fed to the infant. **Levo-thyroxine should not be mixed with soy formula or with formula containing iron, as these products interfere with absorption of the medication.** Dosage will need to be gradually increased as the infant grows.

Incidence Congenital hypothyroidism occurs in one of every 3,500 to 5,000 births; it is twice as common in females as in males. CH also is more common in Caucasians than African Americans by 5:1. The incidence of CH may be 40 percent higher among Hispanic populations than among Caucasians. Incidence is believed to be still greater among Native American and Asian populations. Illinois began screening for congenital hypothyroidism in 1979, and, on average, the Newborn Screening Program identifies 60-70 new cases of CH each year.

Inheritance Pattern Congenital hypothyroidism occurs sporadically and is not usually an inherited disorder. The disorder is not associated with any prenatal lifestyle or risk factors. A more rare form of CH (about 15 percent of the cases) does involve an inborn (autosomal recessive) error in thyroid hormone synthesis.

Physiology The thyroid gland produces triiodothyronine (T3) and thyroxine (T4) in response to pituitary gland stimulation. The body can convert T3 to T4, and a biofeedback mechanism maintains adequate levels of thyroxine for body metabolism and, in children, normal growth and brain development. Thyroxine, which has no specific target organ sites, is vital to normal function in all organs, tissues and cells in the body. T4 controls the body's metabolic rate. Thyroxine deficiency in infancy can cause severe, irreversible mental and physical retardation, a condition known as cretinism.

There are several types of primary CH, the most common form resulting from abnormal fetal development of the thyroid gland. The thyroid gland may be absent, mislocated (ectopic) or malformed. Transient hypothyroidism may occur in some infants as a result of maternal exposure to excess iodine, antithyroid medications (propylthiouracil or PTU), or exposure of the infant to maternal antithyroid antibodies. The use of iodine-based skin disinfectants on neonates, especially premature neonates, can inhibit thyroxine production resulting in transient hypothyroidism. Untreated maternal hypothyroidism also can result in low fetal levels of thyroxine.

Key Points for Parents Avoid overly alarming the child's parents if the diagnosis of CH has not yet been confirmed. If the child needs additional testing or diagnostic evaluation, make certain the parents understand the importance of following the pediatrician's and /or specialist's recommendations for additional testing and referrals.

Follow-up After Confirmation of Diagnosis These guidelines should be followed after a diagnosis of congenital hypothyroidism has been confirmed:

1. **Parents should understand that treatment for primary congenital hypothyroidism will be lifelong.**
2. Parents should understand that treatment is not curative and that all morbidity cannot necessarily be prevented. Long-term management, monitoring and compliance with treatment recommendations are essential to the child's well-being. A multidisciplinary approach is recommended and should include the following specialties: pediatrics and endocrinology. Infants and children with congenital hypothyroidism should have regular follow-up appointments with a pediatric endocrinologist. Periodic hearing evaluations also are recommended for children with CH, as hearing disorders are sometimes associated with congenital hypothyroidism.
3. Genetic counseling services may be indicated. A list of counselors and geneticists, whose services are available through the Illinois Department of Public Health, should be given to the parents if they have not already seen a geneticist.
4. Provide a list of support services available within the community, such as the local health department and Early Intervention service providers.
5. Additional information about newborn screening can be found at:
 - Baby's First Test: <http://www.babysfirsttest.org/>
Health Resource and Service Administration (HRSA), Grant no. U36MC16509, Quality Assessment of the Newborn Screening System.
 - National Center for Biotechnology Information: <http://www.ncbi.nlm.nih.gov/gtr/>
National Center for Biotechnology Information, U.S. National Library of Medicine, 8600 Rockville Pike, Bethesda MD, 20894 USA.