

<b>Disease Name</b>	<b>Congenital Hypothyroidism</b>
<b>Acronym</b>	CH
<b>Disease Classification</b>	Endocrine Disorder
<b>Symptom onset</b>	Clinical signs of hypothyroidism often do not appear until the infant is 3-4 months of age, thus it is most likely that affected infants will have already suffered irreversible brain damage before signs of the disease begin to appear. Many times the early diagnosis relies almost solely on the results of the newborn screening.
<b>Symptoms</b>	An affected infant may have prolonged neonatal jaundice, growth failure, lethargy, poor appetite and constipation.
<b>Natural history without treatment</b>	Even mild hypothyroidism can lead to severe <u>mental delays</u> and growth delays if untreated. Development is delayed early on, often indicated by failure to meet normal milestones.
<b>Treatment</b>	Daily oral thyroxine medication to prevent problems, treatment must begin shortly after birth and is lifelong.
<b>Inheritance</b>	Although this disorder is detectable at birth, it is not an inherited disorder. Hypothyroidism does not follow any type of pattern as to whom it will affect and randomly affects infants from almost every origin.
<b>General population incidence</b>	Estimated to affect 1 in 4,500 births
<b>OMIM Link</b>	<a href="http://www.ncbi.nlm.nih.gov/omim/201910">http://www.ncbi.nlm.nih.gov/omim/201910</a>
<b>Genetests Link</b>	<a href="http://www.genetests.org">www.genetests.org</a>
<b>Support Group</b>	MAGIC Foundation for Children's growth (MAGIC) <a href="http://www.magicfoundation.org">http://www.magicfoundation.org</a>  National Organization for Rare Diseases <a href="http://www.rarediseases.org">http://www.rarediseases.org</a>

## Newborn Screening ACT Sheet [Elevated TSH (Primary TSH test)] Congenital Hypothyroidism

**Differential Diagnosis:** Primary congenital hypothyroidism (CH); transient CH.

**Condition Description:** Lack of adequate thyroid hormone production..

**YOU SHOULD TAKE THE FOLLOWING ACTIONS:**

- Contact family to inform them of the newborn screening test result.
- Consult pediatric endocrinologist; refer to endocrinologist, if considered appropriate.
- Evaluate infant (see clinical considerations below).
- Initiate timely confirmatory/diagnostic testing as recommended by the specialist.
- Initiate treatment as recommended by consultant as soon as possible.
- Educate parents/caregivers that hormone replacement prevents mental retardation.
- Report findings to state newborn screening program.

**Diagnostic Evaluation:** Diagnostic tests should include serum free T<sub>4</sub> and thyroid stimulating hormone (TSH); consultant may also recommend total T<sub>4</sub> and T<sub>3</sub> resin uptake. Test results include reduced free T<sub>4</sub> and elevated TSH in primary hypothyroidism; if done, reduced total T<sub>4</sub> and low or normal T<sub>3</sub> resin uptake

**Clinical Considerations:** Most neonates are asymptomatic, though a few can manifest some clinical features, such as prolonged jaundice, puffy facies, large fontanel, macroglossia and umbilical hernia. Untreated congenital hypothyroidism results in developmental delay or mental retardation and poor growth.

**Additional Information:**

[American Academy of Pediatrics](#)  
[Genetics Home Reference](#)

**Referral (local, state, regional and national):**

[Testing](#)  
Clinical Services  
[Lawson Wilkins Pediatric Endocrine Society "Find A Doc"](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics, 2010 (Funded in part through MCHB/HRSA/HH5 grant #U22MC03957)

