Disease Name	**Congenital Hypothyroidism**
Acronym	CH
Disease Classification	Endocrine Disorder
Symptom onset	Clinical signs of hypothyroidism often do not appear until the infants 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.
Symptoms	An affected infant may have prolonged neonatal jaundice, growth failure, lethargy, poor appetite and constipation.
Natural history without treatment	Even mild hypothyroidism can lead to severe mental delays and growth delays if untreated. Development is delayed early on, often indicated by failure to meet normal milestones.
Treatment	Daily oral thyroxine medication to prevent problems, treatment must begin shortly after birth and is lifelong.
Inheritance	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.
General population incidence	Estimated to affect 1 in 4,500 births
OMIM Link	http://www.ncbi.nlm.nih.gov/omim/201910
Genetests Link	www.genetests.org
Support Group	MAGIC Foundation for Children's growth (MAGIC) http://www.magicfoundation.org National Organization for Rare Diseases http://www.rarediseases.org

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 T4 and thyroid stimulating hormone (TSH); consultant may also recommend total T4 and T3 resin uptake. Test results include reduced free T4 and elevated TSH in primary hypothyroidism; if done, reduced total T4 and low or normal T3 resin uptake

Clinical Considerations: Most neonates are asymptomatic, though a few can manifest some clinical features, such as prolonged jaundice, puffy facies, large fontanels, 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"