



Newborn Screening ACT Sheet

Primary T4-follow-up TSH test/Low T4 and/or Elevated TSH Congenital Hypothyroidism

Differential Diagnosis: Primary and secondary congenital hypothyroidism (CH), transient CH, thyroxine binding globulin (TBG) deficiency.

Condition Description: Lack of adequate thyroid hormone production.

You Should Take the Following Actions

- Contact family **IMMEDIATELY** to inform them of the newborn screening test result.
- Consult pediatric endocrinologist; refer to endocrinologist if considered appropriate. (See attached list.)
- Evaluate infant (see clinical considerations below).
- Initiate timely confirmatory/diagnostic testing as recommended by the specialist.
- Initial testing should include free T4 and TSH.
- Repeat newborn screen if second screen has not yet been done.
- Initiate treatment as recommended by consultant as soon as possible.
- Educate parents/caregivers that hormone replacement prevents mental retardation.
- Report findings to 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. TSH is reduced or inappropriately normal in secondary (hypopituitary) hypothyroidism. Low total T4 and elevated T3 resin uptake are consistent with TBG deficiency.

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

Additional Information:

New England Newborn Screening Program

<http://www.umassmed.edu/nbs/screenings/disorders/hypothyroidism.cfm>

American Academy of Pediatrics

<http://pediatrics.aappublications.org/cgi/content/abstract/91/6/1203>

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition=congenitalhypothyroidism>



Newborn Screening FACT Sheet

Congenital Hypothyroidism (CH)

What Is CH?

CH is a condition in which the thyroid gland doesn't make enough thyroid hormone. The thyroid gland is in the neck.

What Causes CH?

CH happens when the thyroid gland doesn't make any or enough hormones. This mainly occurs because the thyroid gland didn't develop, or because it developed in a different place in the body. No one knows why the thyroid gland doesn't develop all the way, or why it doesn't develop where it should.

What Symptoms or Problems Occur with CH?

[Symptoms are something out of the ordinary that a parent notices.]

There are different symptoms of CH in babies. The most obvious are:

- **Skin:** Some CH babies have newborn jaundice (a yellow color) longer than usual. Their skin may look pale or blotchy.
- **Appetite and Digestion:** CH babies may not want to feed. It might be hard to keep them awake during feedings. They may be very constipated.
- **Growth:** CH newborns are often large. They usually have poor growth and weight gain if they are not treated.
- **Circulation:** Babies with CH may have a slow heart rate and low blood pressure. Their hands and feet may be cool to the touch. This is because of poor circulation.
- **Activity and Development:** Babies with CH are often quiet and don't cry much. They may not be interested in sights or sounds around them. They may sleep for a long time, then have to be woken up for feeding. They may feel "floppy" when they are picked up.

What is the Treatment for CH?

Medications – The doctor will prescribe synthetic (man-made) thyroid hormone to replace your child's missing thyroid hormone. The medicine acts just like the natural hormone made in the body. The synthetic thyroid hormone doesn't cause other problems when the right amount is given.

Things to Remember

Babies will have normal growth and development when they get their thyroid medicine regularly. Your baby has the same chance for a normal life as any other baby.