

Congenital Hypothyroidism Fact Sheet for Providers

What is Congenital Hypothyroidism?

Congenital hypothyroidism (CH) is a condition present at birth where a child's thyroid gland does not make the right amount of thyroid hormones (important chemicals in the body). The thyroid gland is found at the base of the neck. Most of the time, babies with CH have a thyroid gland that did not fully develop. Thyroid hormone, called thyroxine (T4) is very important for normal growth and development of the brain and body. Babies with congenital hypothyroidism make too little T4 with either too high or too low TSH.

If left untreated after the first few weeks of life, this condition can lead to developmental issues.

What are the symptoms of Congenital Hypothyroidism?

Newborns with CH are often asymptomatic. Symptoms of CH may include:

- Lethargy
- Vomiting
- Poor feeding
- Large tongue
- Jaundice
- Umbilical hernia

How is Congenital Hypothyroidism identified and diagnosed?

All infants with an abnormal CH newborn screening will require a confirmatory serum testing within 24 hours to measure free T4 and TSH levels.

How is Congenital Hypothyroidism treated?

Hormone replacement is used to treat CH. The baby will need to take a daily dose of Levothyroxine. Regular thyroid function tests (TFTs) and follow-up with pediatric endocrinology will be required to ensure correct hormone levels are maintained.

How do I handle an abnormal screen for Congenital Hypothyroidism?

Early treatment is important to make sure the child's mental function is not impacted. If one of your patients has an abnormal CH newborn screen, take the following steps WITHIN 24 HOURS:

- Collect a confirmatory serum sample for free T4 and TSH for your local hospital or reference laboratory to run.
- Examine the newborn for symptoms of CH. The newborn may be asymptomatic.
- If confirmatory testing remains abnormal, consult a pediatric endocrinologist required to ensure correct hormone levels are maintained.

Where do I go for more information?

Use your smart phone's camera to scan the QR codes below.



American Thyroid Association: <https://www.thyroid.org/wp-content/uploads/patients/brochures/congenital-hypothyroidism-brochure.pdf>



Pediatric Endocrine Society: <https://pedsendo.org/wp-content/uploads/2020/06/E-Congenital-Hypothyroidism.pdf>



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