Congenital Hypothyroidism

WHAT IS THE THYROID GLAND?

The thyroid gland is a butterfly-shaped endocrine gland that is located in the lower front of the neck, just above the collarbone. The thyroid's job is to make thyroid hormones, which are released into the blood and then carried to every tissue in the body. In children, thyroid hormone helps to ensure that growth and development occur normally and that the body's energy, metabolism, heart, muscles, and other organs are working properly.

WHAT DOES THYROID HORMONE DO?

Thyroid hormone plays an important role in growth and development. Before a baby is born and up to 2 to 3 years of life, thyroid hormone is very important for brain development. After this time, thyroid hormone is important for growth as well as enabling the body to use energy and stay warm (metabolism) and to help the brain, heart, muscles, and other organs work as they should.

WHAT IS CONGENITAL HYPOTHYROIDISM?

When thyroid hormone deficiency is detected at birth it is called "congenital hypothyroidism". Babies with congenital hypothyroidism are born with an underactive or absent thyroid gland. Because thyroid hormone plays such an important role in brain development and growth, all babies born in the United States, Canada and other developed countries undergo a screening test to check thyroid function shortly after birth. Early detection and treatment of hypothyroidism generally results in normal growth and development.

WHAT CAUSES CONGENITAL HYPOTHYROIDISM?

The most common causes of congenital hypothyroidism are:

1. A thyroid gland in an abnormal location (ectopic thyroid gland)
2. An underdeveloped thyroid gland (thyroid hypoplasia)
3. A missing thyroid gland (thyroid agenesis)

- As a group, these abnormalities are called *thyroid dysgenesis* and are usually not inherited from parents; there is a low chance that additional children will have the same problem.
- Another explanation for congenital hypothyroidism is that the thyroid is in a normal location but it cannot make a normal amount of thyroid hormone. This is called *thyroid dyshormonogenesis*. This form of congenital hypothyroidism may be inherited with a 25% chance (1 in 4) that a future baby from the same parents will have the same, treatable problem.

LESS COMMON CAUSES ARE:

- A "thyroid blocking antibody" passed from mother to the baby during pregnancy; mothers who have *Hashimoto's thyroiditis* may produce this "blocking antibody". The baby may need thyroid hormone treatment, but this form of congenital hypothyroidism is often temporary, lasting a few weeks to months.
- Medications taken by mother during pregnancy. The most common is an "anti-thyroid drug" that is used for the treatment of *Graves' disease* (hyperthyroidism). If a mom ingests extremely high doses of iodine from the diet or from a supplement this may also result in congenital hypothyroidism.
- The baby cannot make the signal from the brain (pituitary gland) to tell the thyroid to work. This signal is called thyroid stimulating hormone (TSH). This may be associated with other pituitary hormone deficiencies. Additional hormone levels and brain imaging will be performed based on the clinical situation

WHAT ARE THE SIGNS AND SYMPTOMS OF CONGENITAL HYPOTHYROIDISM?

Most babies have no obvious signs or symptoms of thyroid hormone deficiency at birth. This is why newborn screening is so important. Other babies may have some of the following features either at birth or developing slowly over the first few months of life:

- A puffy-looking face
- Large, thick tongue
- Large soft spots of the skull
- Hoarse cry
- Distended stomach with outpouching of the belly button (umbilical hernia)
- Feeding problems, including needing to be awakened for feedings and difficulty swallowing
- Constipation
- "Floppy" (poor muscle tone, also called hypotonia)
- Jaundice (a yellow appearance of the skin and eyes)

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HOW IS CONGENITAL HYPOTHYROIDISM DIAGNOSED?

Babies in the United States, Canada and many other countries are tested for congenital hypothyroidism as part of the standard newborn screening program. A heel–prick blood sample is obtained at 1-2 days of age and mailed to the state screening laboratory. The screening laboratory will measure the level of:

- T4 (thyroxine)
- TSH (thyroid stimulating hormone)

If the heel-prick blood T4 level is low and the TSH is elevated, the results suggest congenital hypothyroidism. These results are sent to the baby's primary care provider. The screening test results must be confirmed by another blood test, one that is taken directly from a vein, not a repeat heel-prick. This test will also measure T4 (more often a "free T4") and TSH. If the free T4 is low and the TSH is elevated, a diagnosis of congenital hypothyroidism is confirmed. The doctor may also recommend some form of imaging, such as an ultrasound exam or thyroid scan, to look for a specific cause of congenital hypothyroidism.

HOW IS CONGENITAL HYPOTHYROIDISM TREATED?

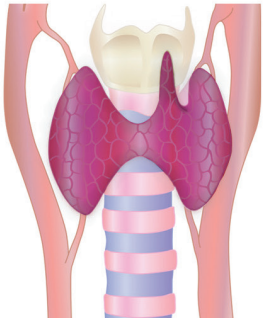
Treatment involves replacing the missing thyroid hormone to restore thyroid hormone levels to normal. The common form of thyroid hormone, considered the best treatment, is called levothyroxine (although it is synthetic, it is identical to theT4 produced by the body). Currently levothyroxine is only available in tablet form. Parents should crush up each day's tablet, and then mix with a small volume (about 1 tsp) of liquid, either expressed breastmilk, water, or formula. This can be given to the baby on a teaspoon or by using a medicine dropper or syringe and squirting the suspension into the baby's mouth (against the side or cheek pad). Levothyroxine should not be mixed with a soy protein formula, as soy protein binds thyroid hormone, reducing absorption from the gut. It is extremely important that parents administer thyroid hormone daily to maintain steady blood levels. In order to do this, parents must fill their baby's levothyroxine prescription in a timely manner, and let their doctor know if they need a refill. Some pharmacists may suggest that they can make a 'suspension' for the baby. These preparations are not stable and they should NOT be used.

HOW OFTEN ARE BLOOD LEVELS CHECKED?

An important part of treatment involves monitoring of blood thyroid hormone levels (TSH and free T4) to make sure that the amount of medication is adjusted to keep up with how fast the baby is growing. Generally, blood tests are checked every 1 to 2 months up to 6 months of age and then every 2 to 3 months thereafter. In general, it is recommended that babies with congenital hypothyroidism be managed in consultation with a pediatric endocrine specialist. The primary care provider or the pediatric endocrine specialist will give instructions for how often the blood tests are monitored.

IS LIFELONG TREATMENT NECESSARY?

For many babies, thyroid hormone replacement therapy will be needed for their entire life. With proper treatment, these children can lead healthy and happy lives, with normal growth and development, and no restrictions as far as activities. For some babies, thyroid hormone deficiency is transient, with treatment required for several months to a few years. In cases where congenital hypothyroidism is thought to be temporary, the baby's doctor (endocrinologist) may recommend a trial off levothyroxine treatment after age 3 years (after the time of critical brain development). Once off of the medication, repeat blood tests to measure the TSH and free T4 levels will help determine if the baby can stay off of thyroid hormone replacement or whether it needs to be restarted.



FURTHER INFORMATION

Further details on this and other thyroid-related topics are available in the patient thyroid information section on the American Thyroid Association® website at *www.thyroid.org*.

For information on thyroid patient support organizations, please visit the *Patient Support Links* section on the ATA website at *www.thyroid.org*