



Congenital Hypothyroidism

WHAT IS CONGENITAL HYPOTHYROIDISM?

Newborn babies who are unable to make enough thyroid hormone have congenital hypothyroidism, meaning they are born with an underactive or absent thyroid gland. If the condition is not found and treated, children can have mental retardation and growth failure. But the good news is that early diagnosis and proper treatment can prevent these problems. In most cases, the condition is permanent and your child will need lifelong treatment.

WHAT CAUSES CONGENITAL HYPOTHYROIDISM?

The most common causes are

- An underdeveloped thyroid gland
- A thyroid gland that's not located where it should be (in the neck below the voice box or larynx)
- A missing thyroid gland

These abnormalities are not inherited from the parents.

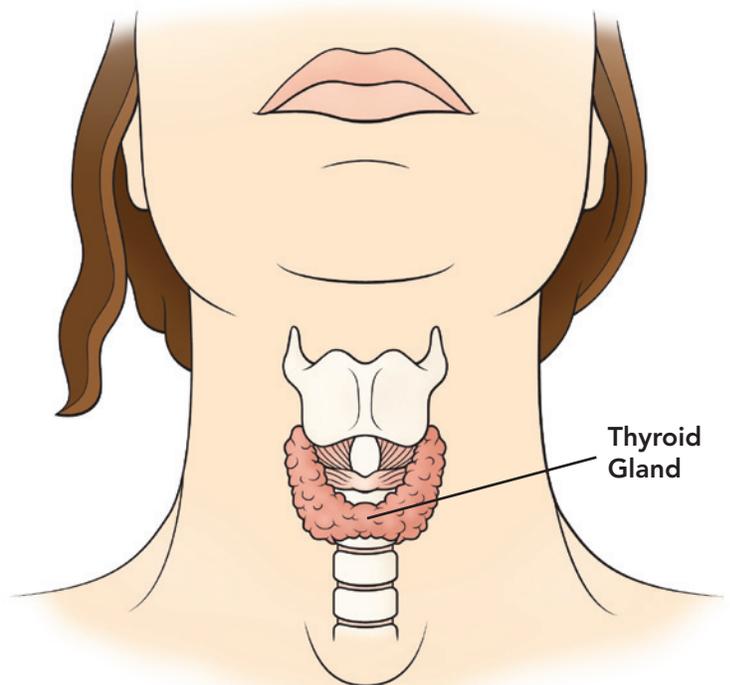
Other possible causes include

- Defective production of thyroid hormone (an inherited condition)
- Problems with the pituitary gland (located at the base of the brain), which tells the thyroid to make thyroid hormone

Less commonly, a mother's thyroid disease or medicines taken during pregnancy can cause congenital hypothyroidism.

DID YOU KNOW?

About 1 in every 2,000 to 4,000 babies is born with congenital hypothyroidism. It is twice as common in girls as in boys.



WHAT DOES THYROID HORMONE DO?

Thyroid hormone helps your baby's brain and nervous system develop normally. It also helps the heart, muscles, and other organs work properly. Thyroid hormone regulates how the body uses and stores energy and how the body grows. The thyroid gland makes two forms of thyroid hormone, T4 (thyroxine) and T3 (triiodothyronine).

WHAT ARE THE SIGNS AND SYMPTOMS OF CONGENITAL HYPOTHYROIDISM?

Many babies with congenital hypothyroidism appear normal at birth or for several months after birth. But others may have these signs and symptoms:

- Jaundice (yellowing of the skin and eyes)
- Constipation
- Poor muscle tone
- Sluggishness
- A hoarse cry

They might also have

- Feeding problems
- A thick, large tongue
- A puffy-looking face
- A swollen abdomen, sometimes with "outpouching" of the belly button
- Large soft spots of the skull

Thyroid hormone deficiency can also occur in older babies or young children, even if test results at birth were normal. If your child shows signs and symptoms of hypothyroidism, contact your doctor right away.

HOW IS CONGENITAL HYPOTHYROIDISM DIAGNOSED?

Most newborns in the United States are tested a few days after birth for congenital hypothyroidism as part of standard screening tests. These tests detect almost all cases of congenital hypothyroidism. Doctors check blood levels of

- T4, one of the thyroid hormones
- Thyroid-stimulating hormone (TSH), a substance produced by the pituitary gland that tells the thyroid gland to make thyroid hormone

Low levels of T4 and high levels of TSH suggest a diagnosis of congenital hypothyroidism. Doctors also might do a thyroid scan or an ultrasound exam to look at the size and location of the thyroid or to find out if it is missing altogether.

HOW IS CONGENITAL HYPOTHYROIDISM TREATED?

Every day, you will give a synthetic (laboratory-made) form of thyroid hormone to your baby to replace the missing thyroid hormone. You'll crush the hormone pill and mix it with a small amount of water, breast milk, or formula. (Do not mix it with soy protein-based formulas, since they interfere with absorption of thyroid hormone.) Then you'll give it to your baby by mouth, using a medicine dropper, syringe, or open nipple. Treatment begins as soon after birth as possible. In most cases, your child will need to take daily thyroid hormone throughout life.

Talk with your doctor about how often to have follow-ups. The doctor will recommend blood tests to check your child's hormone levels regularly, including one month after any dose change, to make sure your child is getting the right amount of hormone. The doctor also will check your child's growth and brain development.

Questions to ask your doctor

- How do I give thyroid medicine to my child? When?
- How often does my child need check-ups?
- What foods and minerals can affect how my child's body uses thyroid hormone? What should I do about them?
- Should my child see a pediatric endocrinologist?

RESOURCES

- Find-an-Endocrinologist: www.hormone.org or call 1-800-HORMONE (1-800-467-6663)
- American Thyroid Association: www.thyroid.org or call 1-800-THYROID (1-800-849-7643)
- Pediatric Endocrine Society: www.lwpes.org or call 1-703-556-9222
- MedlinePlus: www.medlineplus.gov (search for congenital hypothyroidism)
- The Magic Foundation: www.magicfoundation.org (search for congenital hypothyroidism)

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The Hormone Health Network offers free, online resources based on the most advanced clinical and scientific knowledge from The Endocrine Society (www.endo-society.org). The Network's goal is to move patients from educated to engaged, from informed to active partners in their health care. This fact sheet is also available in Spanish at www.hormone.org/Spanish.

