

Understanding Congenital Hypothyroidism

Congenital hypothyroidism is a disorder that affects the thyroid gland. This handout will explain congenital hypothyroidism and its causes and symptoms. It will also explain how we test for and treat congenital hypothyroidism and explain the long-term outlook for babies with congenital hypothyroidism.

WHAT IS CONGENITAL HYPOTHYROIDISM?

Congenital hypothyroidism is a condition in which the thyroid gland cannot make the right amounts of thyroid hormone at birth or even before birth.

Congenital hypothyroidism is fairly common, affecting 1 out of every 1,500 to 2,000 babies. It is among the most common preventable causes of mental retardation.

WHAT IS THE THYROID GLAND?

The thyroid gland is a butterfly-shaped gland located in the front of the neck. It makes the thyroid hormones T4 (thyroxine) and T3 (triiodothyronine).

The thyroid hormones are essential for:

- Normal body growth
- Brain development
- Maintaining the body's temperature
- Regulating the body's metabolism

WHAT IS THE PITUITARY GLAND?

The pituitary gland is a pea-sized gland in the brain that makes thyroid stimulating hormone (TSH). TSH regulates the release of thyroid hormones from the thyroid.

WHAT CAUSES CONGENITAL HYPOTHYROIDISM?

In congenital hypothyroidism, a baby's thyroid makes too little of the hormones T4 and T3. This causes the pituitary gland to increase its production of TSH to push the thyroid to work harder.

The most common causes of congenital hypothyroidism are:

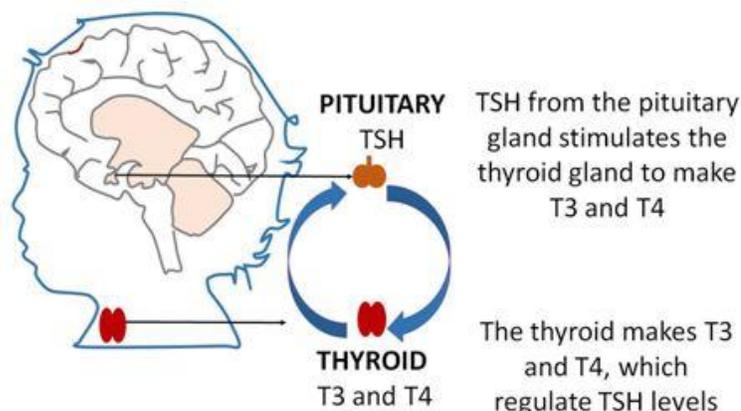
- A baby's thyroid is not in its normal location
- A baby's thyroid hasn't developed properly
- A baby's thyroid hasn't developed at all

Most of the time, we do not know why this happens.

Sometimes babies have congenital hypothyroidism because there is a problem in the production of thyroid hormones. This is often an inherited condition, which means the condition was passed down from the parents.

Sometimes, babies develop congenital hypothyroidism because the pituitary is not making enough TSH or because the thyroid gland cannot respond to TSH.

The Pituitary and Thyroid Gland



WHAT ARE THE SYMPTOMS OF CONGENITAL HYPOTHYROIDISM?

Babies with congenital hypothyroidism typically look normal and do not show symptoms. This is because the mother's thyroid hormones help the baby before birth and because the newborn screening program is able to diagnose hypothyroidism within days of birth.

Some symptoms you might notice if treatment is delayed or if the dose of thyroid hormone being given to your baby is too low include:

- Jaundice (yellowing of the skin) in a newborn for longer than usual
- Puffy face or swollen tongue
- Hoarse (raspy) cry
- Not eating well
- Cold arms and legs
- Low muscle tone
- Umbilical hernia (the belly button sticks out more than usual)
- Constipation (having trouble passing stool or having hard stools)
- Sleeping more than usual
- Poor growth

HOW DO WE TEST FOR CONGENITAL HYPOTHYROIDISM?

Most often, congenital hypothyroidism is discovered through the newborn screening test. All babies have this test when they are born. A newborn screening test screens your baby for congenital hypothyroidism as well as other diseases that can affect newborns.

For the newborn screening test, we take a few drops of blood from your baby's heel soon after he or she is born. The test results are sent to your baby's pediatrician.

When a baby's newborn screening test shows possible congenital hypothyroidism, we repeat the thyroid labs using a blood test. For this, we take a small amount of blood from a vein in your baby's arm. The blood test will check the level of thyroid hormone in your baby's blood. Blood tests also help us figure out how much medication to give your baby, and whether the amount of medication we are giving is the right amount.

Sometimes, we might also have your baby get a thyroid scan. This helps us determine how well your baby's thyroid gland has developed and if it is in its usual position in the front of the neck. When the thyroid gland is not in the right position, we call this an ectopic thyroid gland. This is important in determining whether your baby might receive a trial off medication at 3 years.

HOW DO WE TREAT CONGENITAL HYPOTHYROIDISM?

We treat your baby's congenital hypothyroidism with a medication called **levothyroxine**. This is the natural thyroid hormone that your baby's thyroid cannot make and is taken in the form of a pill. There are different brand names for levothyroxine.

The dose of levothyroxine is adjusted as your baby grows to keep your baby's thyroid hormones at a normal level.

HOW DO WE MONITOR TREATMENT FOR CONGENITAL HYPOTHYROIDISM?

Your baby will need blood tests so we can check his or her thyroid hormone levels and adjust his or her levothyroxine dose accordingly.

Your baby's endocrinologist or pediatrician will ask you to bring your baby in for blood tests every 1-2 months during the first year of life, every 2-3 months in the second and third years, and every 4-6 months after your baby's third year.

During your baby's first 3 years, it is very important that his or her thyroid hormone levels are in the normal range. This will help make sure that your baby's brain develops properly.

Most children will need treatment throughout their life. For some children, hypothyroidism is mild and temporary. Sometimes, your child might be given a brief trial off medications after 3 years of age. Your child would then be retested to see if medication needs to be restarted.

It is very important that you keep your doctor's appointments and give your baby the medicine every day.

WHAT IS THE LONG-TERM OUTLOOK FOR CONGENITAL HYPOTHYROIDISM?

Your baby should do well and be healthy if we treat him or her early and with the right dose of medication.

If your baby's treatment is delayed or the dose of thyroid hormone given is too low, it may affect his or her development, learning and school performance.

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