

Congenital Hypothyroidism

What is the thyroid gland?

The thyroid gland is shaped like a bow tie or butterfly and is located just below the “Adam's apple” in the neck. It makes two important hormones: thyroxin (T4) and triiodothyronine (T3). Another hormone, thyroid-stimulating hormone (TSH) is produced in the brain by the pituitary gland. Thyroid-stimulating hormone’s purpose is to send a message to the thyroid gland to make T3 and T4. Thyroid hormones (T3, T4) control growth and development of the body and brain, influence energy use (metabolism), and directs the function of the heart, liver, kidneys and skin.

What is Congenital Hypothyroidism?

Hypothyroidism is a condition in which the thyroid gland does not make enough thyroid hormone. If a baby is born with hypothyroidism, it is called congenital hypothyroidism. It occurs in approximately one out of every 4000 births. It is twice as common in girls. Most cases occur sporadically and are not inherited. Thyroid hormones are necessary for normal growth of the brain during the first 2 to 3 years of life. Therefore, proper treatment of children with congenital hypothyroidism must be started as soon as possible after birth in order to ensure normal growth and development of the brain. Babies have their thyroid hormone levels checked soon after birth so that congenital hypothyroidism can be promptly detected and treated.

What causes congenital hypothyroidism?

There are several causes of congenital hypothyroidism. The most common cause is that a baby is born without a functioning thyroid. The gland is not present, or is in the wrong place and not functioning properly. This type of congenital hypothyroidism usually does not run in families. It is not related to anything the mother did or may not have done during pregnancy. Less commonly, the thyroid gland is present but the hormones are not being made. This type of problem does tend to run in families.

What are some of the signs and symptoms?

Most babies with hypothyroidism appear normal at birth. The signs and symptoms of hypothyroidism are often absent in the newborn baby because the mother passes along some of her thyroid hormone to the baby during pregnancy. Some babies develop signs and symptoms of hypothyroidism weeks to months after birth. They can have a large soft spot on the head (fontanel), low temperature, feeding difficulties, constipation, jaundice (yellow skin), sleepiness, or color changes. Often, however, there are no reliable signs. If untreated congenital hypothyroidism can cause permanent mental retardation and/or brain damage.

How is it diagnosed?

Hypothyroidism is diagnosed by blood tests which measure the amount of thyroid hormones in the body. This testing is done in the nursery as part of the newborn screen before a baby goes home and then again at 2 weeks of age. Small drops of blood from a heel stick are soaked into a piece

of paper. This is sent to the state laboratory where the level of T4 is measured. If this level is low, another hormone called TSH is measured. Both these levels are reported to the baby's doctor and to special endocrinology consultants for the State Health Department. The doctors work together to begin proper treatment.

- Thyroxine (T4) is generally referred to as "thyroid hormone."
- Free T4 is thyroid hormone that is available and active.
- Thyroid-stimulating hormone (TSH) is made in the pituitary gland in the brain; it sends a message to the thyroid gland to make T4.

How is it treated?

The treatment of congenital hypothyroidism is simple and effective. Your baby will be given thyroid hormone replacement medication. It is very important that the medicine be taken every day, preferably at about the same time. The pill can be crushed and given in a tablespoon of applesauce, water, expressed breast milk or formula (do not mix into an entire bottle).

It is important that the pharmacist does not make a suspension/liquid form. Do not give the medication in any soy-based formula as soy proteins can decrease the absorption of the thyroid hormone medication. Be sure to have the prescription filled before you are completely out of medication so doses are not missed. Also, check with your doctor before taking the "generic" form of thyroid medicine. Some doctors want their patients to take a brand name medication. .

You may see the following symptoms if your baby is getting too much thyroid medicine:

- difficulty sleeping
- tremors (shaking)
- weight loss
- irritability
- frequent bowel movements
- excessive hunger

If your baby is not getting enough thyroid medication you may see:

- sleepiness for long periods of time
- constipation
- cold mottled, dry skin
- excessive weight gain or decreased energy/activity

If these symptoms occur, contact your doctor's office. The amount of medication your child is taking may need to be adjusted. Never adjust the dosage of thyroid medication yourself. A blood test is necessary before changing the dose.

When beginning therapy, your baby may sleep less and seem more active. This is normal. Often parents become concerned with the changes in their baby's behavior who was previously a good baby. Actually, the hypothyroid baby just didn't have enough energy to interact with his/her environment. Remember to have patience as you and your baby adjust to his/her increased energy level.

When should our doctor see my child?

Routine office visits are necessary for physical examination, growth checks, measuring thyroid hormone blood levels, and possibly changing medication dosage. Most children with congenital hypothyroidism are seen in the endocrine office every 2-4 months for the first year and then every 3-6 months until growth is completed. Visits may vary depending on your child's particular situation. Remember, you will also need to follow up with your child's primary care doctor for routine health care.

What is the future for my child?

With proper and early treatment, your baby has an excellent chance for normal development. It is important to follow your doctor's orders for giving thyroid replacement medication. Remember, low levels of thyroid in an infant cause permanent mental retardation, so never adjust or stop your child's medication unless instructed to do so by your doctor. Depending on the individual child, your doctor may recommend a trial period off of medication after the age of 2-3 to see if your child will still need thyroid hormone medication therapy. Congenital hypothyroidism is almost always a lifelong condition which requires treatment throughout life. Even after the brain has stopped developing, thyroid hormone is necessary for normal growth, normal energy levels, and normal adolescent and adult development.