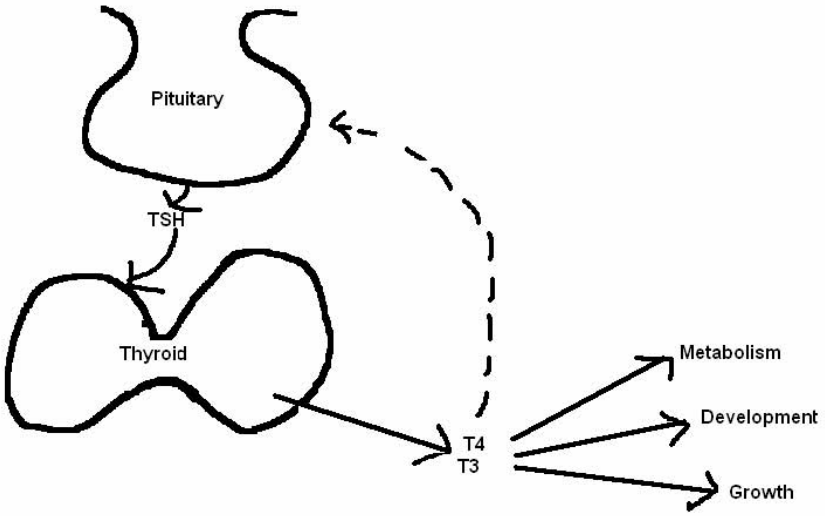
CONGENITAL HYPOTHYROIDISM

Congenital hypothyroidism is a condition in which children are born with an inability to make adequate amounts of thyroid hormones. Thyroid hormones are necessary for normal brain growth during the first 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 brain growth and development. In the past, many children with congenital hypothyroidism were not diagnosed early enough and their brain development was already abnormal by the time treatment was started. Over the past few years, babies born in the USA have had their thyroid hormone levels checked soon after birth so that children with congenital hypothyroidism can be promptly detected and treated. This pamphlet answers a few of the more common questions about congenital hypothyroidism.

WHAT ARE THYROID HORMONES?

Thyroid hormones are produced by the thyroid gland, which is usually located in the front of the neck. The main type of thyroid hormone is called T4. The thyroid gland releases T4 into the bloodstream. Most of the T4 in the blood is stuck onto a protein in the bloodstream called thyroid binding globulin (TBG), while the rest of the T4 floats freely in the blood, and is called free T4. It is not known why we have two forms of T4 in the body, because only free T4 is important for promotion of growth, development, and bodily function.



WHAT IS CONGENITAL HYPOTHYROIDISM?

Congenital hypothyroidism is a condition in which a child is born with an inability to make adequate amounts of thyroid hormones. "Congenital" refers to something that you are born with. "Hypothyroidism" means low thyroid hormone levels.

WHAT CAUSES CONGENITAL HYPOTHYROIDISM?

There are many things that can cause congenital hypothyroidism. In the old days, a deficiency of iodine before or after birth could cause hypothyroidism. This is because iodine is a key ingredient in the production of thyroid hormones. This form of congenital hypothyroidism is still common in some parts of the world. However, it is very **uncommon** in the USA since our modern diet provides adequate amounts of iodine.

The most common cause for congenital hypothyroidism in the USA today is a malformation of the thyroid gland. The development of the thyroid gland

occurs before birth and is a very complicated process. If any part of the process is not right, the thyroid gland will not function properly after birth. Some children with congenital hypothyroidism are born without a thyroid gland; others are born with only part of the gland, or the gland doesn't develop in the right place. We do not completely understand what causes this malformation to occur. This type of congenital hypothyroidism usually does **NOT** run in families.

Very rarely, congenital hypothyroidism can be due to a disorder in which the thyroid gland is present but lacks the ability to manufacture thyroid hormones. This type of problem tends to run in families.

HOW COMMON IS CONGENITAL HYPOTHYROIDISM?

In the USA, about 1:4000 children are born with congenital hypothyroidism.

HOW IS CONGENITAL HYPOTHYROIDISM DIAGNOSED?

In all 50 states, babies have thyroid testing done soon after birth, usually within the first week of life. This testing is usually done in the nursery before a baby goes home. Small drops of blood from a heel stick or fingerstick are soaked into a piece of paper. This is dried and sent to a state laboratory where the level of T4 is measured in the dried blood spot. If this level is low, another hormone called TSH is measured. Both of these levels are then reported to the baby's doctor and to special consultants for the State Health Department. These doctors then work together to begin proper treatment.

WHAT IS TSH?

TSH stands for thyroid stimulating hormone. TSH is produced by the pituitary gland in the brain, released into the bloodstream, and travels to the thyroid gland where it stimulates the production of thyroid hormones. If the thyroid gland is malformed and not working properly, the pituitary gland senses the lack of thyroid hormone in the blood, and produces extra amounts of TSH in an effort to make the thyroid work properly. In most cases of congenital hypothyroidism, the thyroid gland cannot respond to the extra TSH. The TSH levels then become very high.

In very rare cases, the congenital hypothyroidism can be due to a deficiency in TSH production by the pituitary gland. In these cases, the thyroid gland is normally formed but cannot function because of the lack of simulation by TSH. Most of these babies also have other problems with the pituitary gland, and many of them are very sick shortly after birth.

HOW IS CONGENITAL HYPOTHYROIDISM TREATED?

If a baby has an abnormally low thyroid hormone level in the first blood sample, the doctor will recommend that another blood level be checked to make sure that there is a deficiency. The baby will then be started on a medicine called Levo-thyroxine (also known as Levothroid or Synthroid). This is given once a day as a pill which can be crushed up and added to a small amount of formula or baby food. Once your baby is old enough to eat solids, it is better to place the crushed pill in a teaspoon of apple sauce. Levothyroxine is the same as T4.

WHAT HAPPENS IN CONGENITAL HYPOTHYROIDISM IF IT IS NOT TREATED?

One of the main things the thyroid hormones do is to control the pace at which the body functions. If thyroid hormone levels are low, the body tends to slow down all of it's functions. Some babies with congenital hypothyroidism have problems right after birth such as constipation, swelling of the tongue and other body parts, and poor feeding. Most babies with unrecognized congenital hypothyroidism seem to be normal at birth and only later develop problems such as abnormally slow growth and development of teeth, and very weak muscles. In severe cases, the brain does not develop normally and the child will be mentally retarded.

DOES THE TREATMENT WORK?

Hundreds of children with congenital hypothyroidism have now been detected and treated early due to the newborn blood testing program. The children who receive early treatment and who continue to receive proper treatment have **normal growth and development**. Children who are not treated early enough or who do not receive the proper amount of medication may grow and develop at an abnormally slow rate.

HOW IS TREATMENT MONITORED?

At each visit, blood is taken and sent to the laboratory for measurement of thyroid hormones. We usually check the level of total T4 and free T4. At the same time, we measure TSH. Because the pituitary is able to sense the level of T4 any increase in the level of TSH indicates that the amount of T4 in the blood is inadequate. The pituitary is not able to sense whether the T4 is coming from the thyroid gland or from the thyroid tablets your child is taking.

HOW OFTEN SHOULD BLOOD TESTS FOR THYROID HORMONE BE CHECKED?

To make sure that a child is receiving the proper amount of the thyroid medication, a blood test is usually taken about 4-6 weeks after the first dose is given. T4 and TSH levels are measured in this sample. Some laboratories are also able to measure free T4. Once the right dose is established, blood levels of these hormones should be checked about every 3 months until the age of 3 years. This is to make sure that the levels remain normal for the entire period during which the brain is developing. After the age of 3 years, the testing can be performed every 4-6 months. As a child grows, the dose of the thyroid medication needs to be gradually increased according to the laboratory test results and the child's growth measurements.

HOW LONG WILL THE THYROID MEDICATION BE NEEDED?

Congenital hypothyroidism is almost always a life-long condition, and treatment is required throughout life. Even after the brain has stopped developing, thyroid hormone is necessary for normal growth, normal energy levels, and normal sexual development and functioning. Older children with congenital hypothyroidism can be very short, perform poorly in school, and have very late puberty if they are not treated. Adults with untreated hypothyroidism can be very lazy, mentally slow, and overweight. Severe hypothyroidism in either children or adults can lead to very thin and fragile hair, partial baldness, very dry skin, and an increased cholesterol.

In very rare cases, congenital hypothyroidism can be temporary. In these cases, a child will have normal T4 and TSH levels throughout the first 3 years of life without any changes in the dose of thyroid medication. If this happens, the doctor may recommend a 4-6 week period without thyroid medication after the age of 3 years to see whether the medicine is still needed.

HOW OFTEN WILL MY CHILD NEED TO SEE THE DOCTOR?

Your child should see his/her pediatrician or family doctor on the same schedule as for other children for checkups, illnesses, and shots. If your child has congenital hypothyroidism, he/she should visit a pediatric endocrinologist at least every 3 months during the first 3 years of life, and about every 4-6 months after that until he/she has finished growing and developing. A pediatric endocrinologist is a specialist in childhood hormone disorders. These visits are recommended to make sure that the thyroid hormone therapy is adequate.

WILL MY CHILD'S BRAIN DEVELOPMENT BE NORMAL?

If treatment is started before one month of age, there is every anticipation that your child's development will be normal. However, your child's development will need to be monitored regularly by your pediatrician. We recommend that a formal developmental assessment be made at 5 years of age before entry into kindergarten. These measures are to ensure that, if there are any learning disabilities or problems with speech or coordination, your can receive expert assistance to allow him/her to overcome them.

FINAL WORDS

This pamphlet was designed to answer some of the questions which parents have about congenital hypothyroidism. It is not meant to replace us. If you have any questions, please call and ask questions. Our phone number is (303) 783-3883.