Pediatric Endocrinology Fact Sheet

Congenital Hypothyroidism: A Guide for Families

What is congenital hypothyroidism?

Hypothyroidism refers to an underactive thyroid gland. Congenital hypothyroidism occurs when a newborn infant is born without the ability to make normal amounts of thyroid hormone. Congenital hypothyroidism occurs in about 1 in 3000-4000 children, is most often permanent and treatment is lifelong. Thyroid hormone is important for your baby's brain development as well as growth, therefore, untreated congenital hypothyroidism can lead to mental retardation and growth failure. However, because there is excellent treatment available, with early diagnosis and treatment, your baby is likely to lead a normal, healthy life.

What causes congenital hypothyroidism?

Congenital hypothyroidism most often occurs when the thyroid gland does not develop properly, either because it is missing, is too small, or ends up in the wrong part of the neck. Sometimes the gland is formed properly but does not produce hormone in the right way. Also, sometimes the thyroid is missing the signal from the pituitary (master) gland, which tells it to produce thyroid hormone. In a small number of cases, medications taken during pregnancy, mainly medications for treating an overactive thyroid, can lead to congenital hypothyroidism, which is temporary in most cases. Congenital hypothyroidism is usually not inherited through families. This means if one child is affected, it is unlikely that other children you may have in the future will suffer from the same condition.

What are the signs and symptoms of congenital hypothyroidism?

The symptoms of congenital hypothyroidism in the first week of life are not usually obvious. However, sometimes when hypothyroidism is severe, there may be poor feeding, excessive sleeping, weak cry, constipation, and prolonged jaundice (yellow skin) after birth. In these babies, the doctor may find a puffy face, poor muscle strength, and a large tongue with a distended abdomen and larger-than-normal fontanelles (soft spots) on the head.

How is congenital hypothyroidism diagnosed?

Given the difficulty in diagnosing congenital hypothyroidism in the newborn period based on signs and symptoms, all hospitals in the United States, under the supervision of state health departments, screen for this disease using blood collected from your baby's heel before discharge from the hospital. This process is called newborn screening. When there is a positive result (a low level of thyroid hormone with a high level of thyroid-stimulating hormone, called TSH, from the pituitary), the screening program immediately notifies the baby's doctor, usually before the baby is 2 weeks old. Before starting treatment, your baby's doctor will order a blood sample from a vein to confirm the diagnosis of congenital hypothyroidism. In some cases, the doctor may order a thyroid scan to see if the thyroid gland is missing or too small.

What is the treatment for congenital hypothyroidism?

Congenital hypothyroidism is treated by giving thyroid hormone medication in a pill form called levothyroxine. Many children will require treatment for life. Levothyroxine should be crushed and given once daily, mixed with a small amount of water, formula, or breast milk using a dropper or syringe. Giving your baby his/her thyroid hormone EVERY DAY and having regular checkups with a pediatric endocrinologist will help ensure that your baby will have normal growth and brain development. Your doctor will do periodic thyroid function tests so that the dose of medication can be properly adjusted as your child grows. Please see the handout "Thyroid hormone administration practical tips" for a list of foods to avoid giving at the same time as thyroid medicine. This includes soy milk, vitamins with iron, and calcium.

The hormone in the pill is identical to what is made in the body, and you are just replacing what is missing. In general, side effects occur only if the dose is too high, which the endocrinologist can avoid by checking blood levels on a periodic basis.

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