Overview

Congenital hypothyroidism (CH) is a condition that affects infants from birth (congenital) and results from a partial or complete loss of thyroid function (hypothyroidism).

What is congenital hypothyroidism?

Congenital hypothyroidism occurs when the thyroid gland fails to develop or function properly. The thyroid gland is a butterfly-shaped gland in the lower neck. It makes hormones that are important for regulating growth, brain development, and metabolism (chemical reactions in the body).

In 80 to 85 percent of all babies with congenital hypothyroidism, the thyroid gland is absent, abnormally located, or too small. In the remaining 15-20% of cases, the thyroid gland looks normal, but it does not produce any or enough hormones. If untreated, congenital hypothyroidism can lead to developmental delay or intellectual disabilities and poor growth. If treatment begins within the first month after birth, infants usually develop normally.

Why is newborn screening done for congenital hypothyroidism?

Newborn screening is done for congenital hypothyroidism so that babies with this condition can be diagnosed and treated quickly. Immediate diagnosis and treatment of congenital hypothyroidism is important for normal brain development and physical growth. Without prompt diagnosis and treatment, infants with congenital hypothyroidism will develop varying degrees of intellectual disabilities and abnormal growth.

Does a positive result from the Kansas Newborn Screening Lab mean that my baby has congenital hypothyroidism?

No, not necessarily. Newborn screening tests the baby's level of thyroid-stimulating hormone (TSH) but additional tests will need to be done to determine if the baby has congenital hypothyroidism or not.

How common is congenital hypothyroidism?

Congenital hypothyroidism affects 1 in 5,000 infants and is more common in girls. It is more common in Hispanics than in Caucasians and even less common in African Americans.

Is congenital hypothyroidism inherited?

Most cases of congenital hypothyroidism are sporadic, which means that they occur in people without a history of the disorder in their family. A few genes responsible for sporadic congenital hypothyroidism have been identified.

An estimated 15-20% of cases are inherited. Many of these cases are autosomal recessive. This means that both parents of the affected child are carriers of the condition, but they do not have the disease. Each pregnancy between carrier parents has a 25% chance of producing a child affected with congenital hypothyroidism, a 50% chance of producing an unaffected carrier child, and a 25% chance of producing a child who is unaffected and is not a carrier. Some
inherited cases have an autosomal dominant pattern of inheritance, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Multiple genes are associated with congenital hypothyroidism and there may be other associated genes that have yet to be well characterized. Such defective genes prevent the thyroid gland from producing normal amounts of thyroid hormones.

- **What are the signs and symptoms of congenital hypothyroidism?**

Most infants with congenital hypothyroidism appear normal at birth. Some babies have subtle symptoms, but many of these symptoms are also found in babies who do not have congenital hypothyroidism. Symptoms may include feeding problems, prolonged jaundice, constipation, an enlarged or protruding tongue, hoarse cry, protruding abdomen with umbilical hernia, and sluggish reflexes. Without treatment, individuals with congenital hypothyroidism have poor growth and developmental delay or intellectual disabilities.

- **How is congenital hypothyroidism diagnosed?**

False positive and false negative results are possible with this screening. If TSH is elevated or an infant is considered “high risk,” additional confirmatory blood tests will be ordered to test for thyroid function. Diagnostic testing includes looking for reduced free T4 and elevated TSH. Additional thyroid hormone tests may be ordered.

- **Is there a cure for congenital hypothyroidism?**

No, there is no cure for congenital hypothyroidism. However, the outcome is usually excellent in infants who are treated early and continue with lifelong treatment.

- **How is congenital hypothyroidism treated?**

  - Lifelong treatment with oral thyroxine is required to ensure normal growth and development. Individuals with CH need to be followed closely to ensure adequate thyroid hormone levels and to monitor development.
  - Periodic hearing evaluations are recommended as hearing disorders are sometimes associated with CH.
  - Infants with CH have an increased incidence of heart defects, which may require treatment.

- **Where can I get additional information?**

  1. The Thyroid Foundation of America at www.tsh.org

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