Congenital Hypoth

The thyroid is a butterfly-shaped gland at the base of the neck. Its function is to make and secrete specific hormones, particularly thyroid hormone (also called thyroxine or T4). Congenital Hypothyroidism (CH) is a condition present at birth in which an infant fails to produce enough thyroid hormone, resulting in a deficiency.

There are number of different causes for congenital hypothyroidism. Most common is a defect of development of the thyroid itself, resulting in an absent or underdeveloped gland. CH can also occur due to defects in the production of thyroid hormone within a structurally normal gland. In rare cases, the defect is due to a deficiency of thyroid stimulating hormone (TSH), either on its own or as part of a different condition called congenital hypopituitarism.

People with hypothyroidism have specific symptoms and health issues. Infants born with CH may at first show no effect or mild symptoms that are not recognized as a problem: excessive sleeping, reduced nursing, choking episodes, low muscle tone, infrequent bowel movements, jaundice, and a low hoarse cry. If the thyroid hormone deficiency is severe, infants are more likely to also have a puffy face, a large fontanel (the soft spot on the top of the head), an umbilical hernia, and a large tongue. As infants grow into young children, thyroid hormone is crucial for normal growth and development of the body and brain. Babies who do not have enough thyroid hormone are often slow to grow, sluggish, and have learning delays and other health problems. Untreated CH is the most common preventable cause of mental retardation. Few treatments in the practice of medicine provide such a large benefit for so little effort.

Risk Factors

The majority of CH cases are sporadic, which means they occur without an identifiable genetic mutation in people with no history of the disorder in their family. About 15 to 20 percent of CH cases are inherited, due to changes in a pair of genes from the baby's parents. If another member of the infant's family has CH, it is a good indication that this may be the inherited form of the disorder. Most of these cases are inherited in an autosomal recessive pattern, which means both copies of the gene have a defect (also called a mutation). The parents of a child with an autosomal recessive condition are usually carriers—they each have one copy of the mutated gene, but their other copy is normal, so they do not show signs and symptoms of the disease. Some more rare inherited forms of CH have an autosomal dominant pattern of inheritance, which means that a person needs only one copy of the mutated gene to cause the disorder.

If a pregnant women is treated for thyroid cancer, the thyroid gland may be destroyed in the developing fetus as well. Infants whose mothers have taken such medicines should be observed carefully after birth for signs of hypothyroidism.

For reasons that remain unclear, congenital hypothyroidism affects females about twice as often as males.

Diagnosis and Management

Congenital hypothyroidism can be easily detected by a newborn screening program, which is the most common method of diagnosis in developed countries. This test measures the levels of thyroid hormone and TSH on the second or third day of life.

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If thyroid hormone is too low or TSH is too high, the infant may be referred to a pediatric endocrinologist to confirm the diagnosis and initiate treatment. The goal of a CH newborn screening program is to detect the disease and start proper treatment within the first one to two weeks of life, before the deficiency irreversibly damages the infant's growth and maturation.

Without newborn screening, less than half of cases of severe hypothyroidism are recognized in first month of life. Over time, these infants grow poorly and show delayed development. Persistence of severe, untreated hypothyroidism results in severe mental impairment and low IQ. Many of these children eventually require institutional care.

Treatment for CH is simple, effective, and inexpensive. Infants are given a daily dose of replacement thyroid hormone, available as a small tablet that is crushed and mixed with a small amount of water or milk. After a few weeks, the thyroid hormone and TSH levels are rechecked to confirm that they are being normalized by treatment. As the child grows up, these levels are checked regularly to maintain the right dose, which must increase as the child grows. Individuals with CH need to take this medication their whole lives and should not ever stop daily treatment. Most children with CH who are treated correctly and promptly grow and develop normally, without any signs of developmental or mental deficiency.

Congenital Hypothyroidism in Arab Populations

CH is a one of the most common genetic diseases worldwide. In the United States and Europe, approximately 25-30 babies are born with this condition for each 100,000 live births. Middle Eastern countries tend to have even higher rates of disease: 29/100,000 in Kuwait and Saudi, 32/100,000 in Qatar, 34/100,000 in Bahrain, 45/100,000 in Oman, 71/100,000 in Iran, and 94/100,000 in the UAE. Cases have also been reported in Egypt and Syria, though accurate incidence rates are not available.

The Middle East and North Africa newborn screening initiative has set a goal for all member countries to institute national programs to test neonates for CH. Many countries in the region have already implemented comprehensive programs, including Egypt, Iran, Lebanon, Oman, Palestine, Qatar, Saudi Arabia, and the UAE. Some other countries, including Jordan, Kuwait, and Tunisia, have pilot screening programs that test a portion of the population, with the goal of expanding screening to all babies born in these countries. A few countries are still in the planning stages for routine neonatal CH testing.