**Congenital Hypothyroidism**

**What is congenital hypothyroidism?**
Congenital hypothyroidism is a condition where the thyroid gland does not make enough thyroid hormone. The thyroid gland is a small butterfly-shaped gland located in the neck. Normally, it uses iodine from food we eat to make thyroid hormone. This thyroid hormone is also known as thyroxine (T4). T4 is needed for normal growth and development. If congenital hypothyroidism is left untreated, it can lead to growth failure, mental retardation and other serious health problems.

About one baby in 3,000 is born with congenital hypothyroidism in the United States. The condition can be found in all ethnic groups.

**How does congenital hypothyroidism affect a child?**
The bad effects often do not appear until a baby is three to four months old. Some babies may have newborn jaundice, poor growth and weight gain, lengthy sleeping habits that require a baby to be awakened for feeding and severe constipation. If the baby is not treated, it can result in mental retardation, growth failure, deafness and other neurological problems.

**What causes congenital hypothyroidism?**
The most common cause of congenital hypothyroidism is the incorrect growth of the thyroid gland. Early in fetal development, a baby’s thyroid gland begins to form in the brain and then moves to the neck. In some cases this process is interrupted, resulting in a small thyroid gland (dysgenesis) or even the lack of a thyroid gland (agenesis). This is a permanent condition that a baby does not outgrow.

Congenital hypothyroidism does not appear to have a clear pattern of inheritance. There are some types of inherited hypothyroidism that are autosomal recessive. Most often, the parents of a child with an autosomal recessive condition are not affected because they are “carriers,” with one copy of an altered gene and one copy of the normal gene. Your endocrinologist and/or regional genetic center will let you know if your baby has this type of hypothyroidism and the chance of having another child with this condition.

**Is there a test for congenital hypothyroidism?**
Yes. Babies are tested (newborn screening) for congenital hypothyroidism before they leave the hospital. The baby’s heel is pricked and a few drops of blood are taken. The blood is sent to the state laboratory to look at the thyroid-stimulating hormone (TSH, a precursor to thyroxine (T4)).

**Can congenital hypothyroidism symptoms be prevented?**
Yes. In most cases, the bad effects of congenital hypothyroidism can be prevented by replacing the missing T4 with medication given by mouth. The medication form of thyroxine is needed throughout an individual’s lifetime. Follow up with an endocrinologist may be done to evaluate the amount of thyroid gland present, its location, and watch how much thyroxine is being made. Consult with your endocrinologist and/or regional genetic center to determine what tests and follow up are needed.

**DISCLAIMER:** The information contained on this page is not intended to replace the advice of a genetic metabolic medical professional.
Resources:
MUMS National Parent-to-Parent Network
Julie J. Gordon 150 Custer Court Green Bay, Wisconsin 54301-1243
Phone: 1-877-336-5333 (Parents only please)
Phone: 1-920-336-5333
Fax: 1-920-339-0995
E-mail: mums@netnet.net
www.netnet.net/mums/

American Thyroid Association
6066 Leesburg Pike, Suite 650
Falls Church, VA 22041
www.thyroid.org

The Hormone Foundation
8401 Connecticut Avenue, Suite 900
Chevy Chase, MD 20815-5817
Phone: 1-800-HORMONE
www.hormone.org

References:
