

MAGIC NZ

Supporting Children with Growth Disorders
and their Families



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Congenital Hypothyroidism

Congenital Hypothyroidism versus Clinical Hypothyroidism

Congenital hypothyroidism is a disorder that affects infants from birth (congenital), resulting from the loss of thyroid function (hypothyroidism), normally due to failure of the thyroid gland to develop correctly. Sometimes the thyroid gland is absent or ectopic (in an abnormal location). As a result, the thyroid gland does not produce enough thyroxine/T4 after birth. This may result in abnormal growth and development, as well as slower mental function.

At the time of birth the symptoms and signs of hypothyroidism are minimal or absent, and the lack of adequate thyroid hormone from birth until approximately age 2 years is associated with varying degrees of permanent mental retardation. For these reasons most countries in the western world and every state in the United States routinely perform screening tests within the first week of life to detect congenital hypothyroidism so that prompt treatment can be initiated to prevent mental retardation.

By definition, congenital hypothyroidism is present at, or before, birth. Children may also develop primary (clinical) hypothyroidism when aged 2 years or older.

This brochure focuses on congenital hypothyroidism. For information regarding primary hypothyroidism, please refer to the "Clinical Hypothyroidism" brochure from The MAGIC Foundation.

What is the Thyroid Gland?

The thyroid is a bow tie shaped gland located in the neck, below the Adam's apple. The thyroid gland is part of the endocrine system. This gland is responsible for secreting a hormone called thyroxine (T4), which plays a vital role in normal growth and development in children. This gland, like other glands in the endocrine system, is controlled by the pituitary gland. It works very much like a thermostat. The brain senses the amount of T4 and then signals the thyroid with another hormone, thyroid-stimulating hormone (TSH), to produce more or less T4. When the thyroid gland produces enough T4, no extra stimulation is needed and the TSH level remains at a normal level. When there is not enough T4, the TSH rises. These characteristics of the T4 and TSH hormones allow for screening of newborns to assess whether or not they have hypothyroidism (an under-active thyroid gland).

Why did My Child Develop Congenital Hypothyroidism?

In most hypothyroid babies, there is no specific reason why the thyroid gland did not develop normally, although some of these children have an inherited form of this disorder. Congenital hypothyroidism is present in about 1 in 4,000 infants in North America. There are a small proportion of children who have temporary (transient) congenital hypothyroidism for a period of time after birth. Since it is impossible to distinguish these transient hypothyroid babies from those with true congenital hypothyroidism, these infants must be treated as well. Often, in children for whom transient or temporary hypothyroidism is suspected, the medication can be gradually discontinued for a short amount of time on a trial basis after the age of 2 or 3. The child will be retested to see if they can remain off medicine. This is **NOT** the case for true congenital hypothyroidism, where L-thyroxine is necessary throughout your child's life.

Symptoms of Congenital Hypothyroidism

Often babies with congenital hypothyroidism appear perfectly normal at birth, which is why screening is so vital. However, some may have one or more of the following symptoms:

- Large, despite having poor feeding habits, increased birth weight.
- Puffy face, swollen tongue.
- Hoarse cry.
- Low muscle tone.
- Cold extremities.
- Persistent constipation, bloated or full to the touch.
- Lack of energy, sleeps most of the time, appears tired even when awake.
- Little to no growth.

Children born with symptoms have a greater risk of developmental delay than children born without symptoms.

Tests used to find Congenital Hypothyroidism

The usual way to discover congenital hypothyroidism is by a screening process done on all newborns between 24 and 72 hours old. The reason this is done so early is that infants with congenital hypothyroidism usually appear normal at birth and many do not show any of the signs or symptoms noted before. For the screening test, blood is

obtained from your baby's heel and is placed on a filter paper. At a laboratory, the T4 and/or TSH level is measured. If the T4 is low and/or the TSH is elevated, indicating hypothyroidism, your paediatrician is contacted immediately so treatment can begin without delay. It is likely that the blood test will be repeated to confirm the diagnosis.

The physicians may also take an x-ray of the legs to look at the ends of the bones. In babies with hypothyroidism, the bones have an immature appearance, which helps to confirm diagnosis of congenital hypothyroidism. A thyroid scan should be done to determine the location or absence of the thyroid gland. These tests, bone age and thyroid scan can be done at the time of diagnosis.

How Does One Treat Congenital Hypothyroidism?

Treatment for congenital hypothyroidism is replacement of the missing thyroid hormone in pill form. It is extremely important that these pills be taken daily for life because tyroxine/T4 is essential for all the body's functions. In general, the average starting dose for L-thyroxine or Levothyroxine (synthetic T4) in a newborn is between 25 and 50 mcg per day or 10mg/kg to 15mg/kg of body weight. This value increases dependent upon the individual needs of the child. The pill can be crushed and then administered in a small amount of water/formula or breast milk while your child is still an infant.

Please be aware that L-thyroxine should not be mixed with Soy formula as this product interferes with absorption.

Blood tests will be done on a regular basis to ensure that the hormone levels are in a normal range. Thyroid hormone is necessary for normal brain and intellectual development and such development can be delayed when there is a lack of L-thyroxine. With early replacement of adequate thyroid hormone and proper follow-up and care, the outlook for most children with congenital hypothyroidism is excellent.

What Medical Attention My Child Should Receive?

Generally, once normal levels have been established, children are seen every 2-3 months for the first three years. The goal is to maintain the concentration of T4 in the mid to upper half of the normal range (10mg/dL to 16mg/dL) for the first years of life. The TSH level should be maintained within the normal reference range for infants.

The treatment for hypothyroidism is safe, simple and effective; however, successful treatment depends on life-long daily medication with close follow-up of hormone levels.

Making this procedure of taking medication on a routine basis needs to become a part of the lifestyle of you and your child in order to assure optimal growth and development.

A Note to Parents

You have just learned that your baby has congenital hypothyroidism. Suddenly, you have a lot of confusion and certainly may be frightened regarding the well being of your new infant. As a concerned parent, you probably wish to learn as much as you can about the condition and what you and your health care professional can do to help your baby's condition as your child grows and develops.

Advocate for Your Child: Most importantly, be an advocate for your child. Treatment is available to help your child. The appropriate medical care under the proper specialists will greatly improve your child's outcome.

Ask Questions: As you learn about congenital hypothyroidism, it is probable that you will have questions that may be specific to your child. Leave no questions unanswered, even if you think the questions are simple or silly. Don't be afraid to ask questions or get a second opinion. A greater understanding of this condition will allow you to provide optimal care for your child.

Network: If you have any questions, MAGIC NZ can help by putting you in touch with other families affected by congenital hypothyroidism.

For more information visit www.magicnz.org.nz, email jan@magicnz.org.nz
or write to MAGIC NZ, PO Box 1493, Wellington.

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