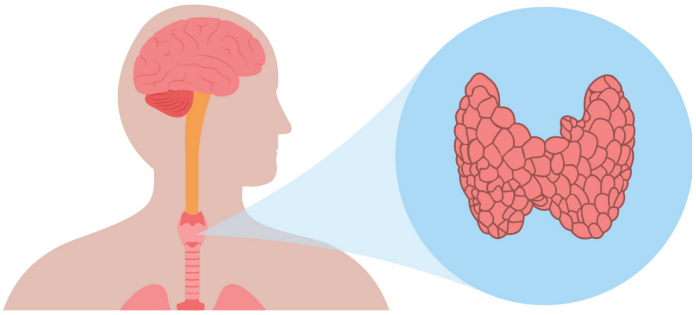
Congenital Hypothyroidism Parent Guidebook

NEXT STEPS AFTER DIAGNOSIS

DEPARTMENT OF HEALTH

What is congenital hypothyroidism?

Congenital hypothyroidism (CH) is a condition present at birth where a child's thyroid gland does not make enough thyroid hormone. The thyroid gland is a butterfly-shaped organ at the base of the neck. It makes iodine-containing hormones that help the cells in the body function properly. The main hormone the thyroid gland makes is called thyroxine (T4). In babies and young children this hormone is very important for normal growth and development of the brain and body.



Thyroid hormones

- Help regulate body temperature.
- Maintain a normal heart rate.
- Help with normal development of the brain and nervous system (spine and nerves).
- Help maintain normal functioning of the digestive track, including bowel movements.

What causes CH?

Missing or misplaced thyroid gland

In about 85% of children with CH, it is caused by a thyroid gland that did not develop or is misplaced. When the gland is missing or in the wrong place, it can't produce enough thyroid hormone. A healthy thyroid gland is located in the center front of the neck, near the top of the windpipe. In some children with CH, the thyroid is located under the tongue or on the side of the neck. This is a birth defect and the reason it happens is still unknown.

Hereditary causes

Less often, CH is caused by changes in a gene or pair of genes. Genes carry the information that decides a person's traits. Genes are passed on to babies from parents. We have no control over which genes are passed on to babies during pregnancy. In this case, even though the thyroid gland is the right size and in the right place, it does not make enough thyroid hormone.

Maternal iodine deficiency

If a mother does not get enough iodine while she is pregnant, her baby might not be able to make enough thyroid hormone. This is very rare in the United States because our table salt is supplemented with iodine ("iodized salt").

Maternal thyroid medication

In very few cases, CH is caused when a mother takes medication to treat her own overactive thyroid while pregnant. It is safe and important for mothers to take this medication (levothyroxine) during pregnancy when needed.

How is CH treated?

Your baby's primary doctor might work with a pediatric endocrinologist, a doctor with training in treating children with thyroid and hormone conditions, to care for your child. The main treatment for CH is thyroid hormone replacement. If this is started immediately after your child is diagnosed, treatment can prevent many or all of the effects of CH.

What happens if CH is left untreated?

If damage to the brain and nerves happens because treatment is delayed, it is usually permanent and cannot be reversed. Children with CH who remain untreated usually become intellectually, mentally, and/or physically disabled. Many will also have speech delays and behavioral problems, along with other physical and cognitive complications.

Thyroid hormone medication

L-thyroxine is a synthetic form of thyroid hormone given in a tablet that is identical to the T4 hormone your thyroid makes. There are also Food and Drug Administration-approved liquid thyroid formulations.
How much your baby needs to take will be determined by your baby's doctor and endocrinologist, and will increase as they grow up. Synthetic thyroid hormone medications need to be taken every day. It is important to tell your child's doctor if you are giving your baby soy-based formula or iron supplements. These can reduce the amount of thyroid hormone your baby absorbs from the medication.

How do you give L-thyroxine to your child?

If prescribed L-thyroxine when your baby is too young to chew tablets, you can:

- Crush the tablets, using the back of a spoon, into a fine powder.
- Wash your hands well, then moisten one of your fingers and dip it into the crushed tablets. Let your baby suck the crushed tablets off. Repeat until the powder is gone.
- Mix the crushed powder with a small amount of formula, breastmilk, or juice. Give this mixture through a dropper on the side of the mouth. Do not mix the crushed tablets into a large amount of liquid, like a bottle. They might not finish the whole bottle and then they won't get all of the medication.

Signs your child's dose is too high

- Trouble sleeping.
- Shaking (tremors).
- Weight loss or poor weight gain.
- Irritability (crabby).
- Diarrhea.
- Very hungry.

Signs your child's dose is too low

- Sleeping too much.
- Constipation.
- Cold, dry skin.
- Weight gained very quickly.
- Low energy/activity level.

How is CH monitored?

Your child will need regular visits to the doctor to check their weight, height, development, and overall health. Regular blood tests will be needed to check the levels of thyroid hormone. Studies support blood tests every 1 to 2 months in the first 6 months of life, every 2 to 3 months in the second 6 months of life, and then every 3 to 4 months between 1 and 3 years of age. After age three these tests can usually be done less often.

Developmental evaluations

Your child's doctor might suggest a formal evaluation of your child's development. If they show delays in certain areas of learning or speech, extra help can be arranged. There are early intervention programs available in Minnesota that provide services for children before they reach school age.

What happens when my baby with CH grows up?

Sometimes, CH is not a permanent condition and no evidence of the disease is present after around 3 years of age. The safest approach is to give your baby medication through the time of crucial brain development.

Do not stop the medication on your own. Your child's endocrinologist may decide to stop the medication for a few weeks to do blood tests. If hormone levels are normal, your child may no longer need medication. If not, your child can start on medication again without any damage during those weeks.

Most children born with CH grow up to lead healthy, happy lives. As your child grows up, it is important to communicate with your child and the rest of your family, and maintain a strong support system of friends and health providers.

Frequently asked questions

How many people have CH?

About one in every 2,000 babies born in the U.S. each year has CH. Twice as many girls are diagnosed with CH than boys.

Does CH occur more in certain ethnic groups?

It is more common in babies of Hispanic or Native American descent. It is less common in babies of African-American descent.

Does CH go by any other names?

- CHT.
- Cretinism.
- Endemic cretinism.
- Congenital myxedema.

Resources

- AAP (American Academy of Pediatrics) | CH: Screening and Management
  www.publications.aap.org/pediatrics/article/151/1/e2022060420/190308/
- Baby's First Test | Congenital Hypothyroidism
  www.babysfirsttest.org
- Major Aspects of Growth in Children (MAGIC) Foundation
  www.magicfoundation.org
- Midwest Genetics Network
  www.midwestgenetics.org
- National Library of Medicine | Genetics Home Reference
  www.medlineplus.gov/genetics

This guide does not provide medical advice. This guide has general information about congenital hypothyroidism (CH). Every child and family is different and some of these facts may not apply to your child. Check with your child's doctor before trying a new treatment or medication.

The information in this guide was adapted from materials by the Star G Project and California Department of Health Services.

DEPARTMENT OF HEALTH

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