



Other Disorders

Screening, Technology, and Research in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn screening and genetic testing – [http:// www.newbornscreening.info](http://www.newbornscreening.info)

Disorder name: Congenital Hypothyroidism

Acronym: CH

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This fact sheet contains general information about congenital hypothyroidism (CH). Every child is different and some of these facts may not apply to your child specifically. Certain treatments may be advised for some children but not others.

What is CH?

CH stands for “congenital hypothyroidism.” Congenital means present at birth. Hypothyroidism is a condition in which the person does not make enough thyroid hormone.

The thyroid gland is a butterfly-shaped organ at the base of the neck. Its job is to make specific hormones that help the cells of the body function correctly. The main hormone made by the thyroid gland is thyroid hormone, also called ‘thyroxine,’ or T4. It is released by the thyroid gland into the bloodstream whenever it is needed by the body. It helps cells work more efficiently and also

helps maintain our body temperature. In babies and young children, thyroid hormone is crucial for normal growth and development of the body and brain.

People with hypothyroidism have specific symptoms and health issues. Babies who do not have enough thyroid hormone are often slow to grow, are sluggish, and have learning delays and other specific health problems. There are a number of different causes for CH which are mentioned below.

What causes CH?

CH can be the result of a number of different underlying causes.

Missing or misplaced thyroid gland

Most babies with CH are missing their thyroid gland or have a thyroid that did not develop properly. In some cases, the thyroid gland may be smaller than usual or may not be located in the correct place.

In healthy people, the thyroid gland is located in the center of the front of the neck, near the top of the windpipe. In some children with CH, the thyroid gland may instead be under the tongue or on the side of the neck. If the thyroid gland is in the wrong place, or if it is underdeveloped, it often does not work well and makes less thyroid hormone than needed by the body. If the thyroid gland is missing, the baby cannot make any of its own thyroid hormone. A missing, underdeveloped, or misplaced thyroid gland is a birth defect that happens for unknown reasons and is usually not inherited.

Hereditary causes

Less often, CH is caused by inherited changes in a gene or pair of genes. This is explained in more detail below. Children with the inherited type of CH do not make enough thyroid hormone even though their thyroid gland appears normal in size and shape. About 15% of children with CH are thought to have an inherited type.

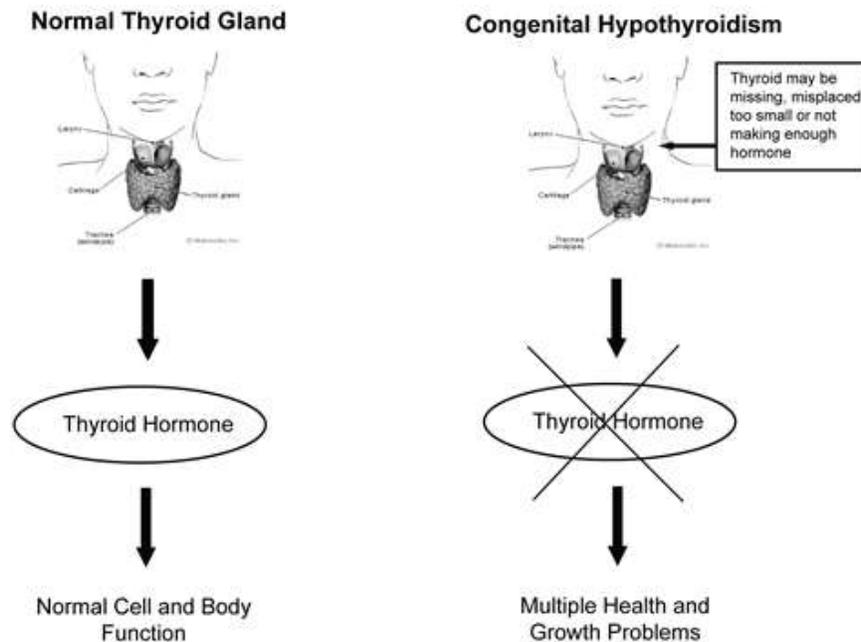
Maternal iodine deficiency

If the mother is deficient in iodine during the pregnancy, the fetal thyroid gland may not be able to make enough thyroid hormone. The baby is then born with CH. This is a problem in some parts of the world where people do not get enough iodine in their diet. This is a very rare cause for CH in the United States because our table salt is supplemented with iodine ('iodized salt'). Other foods, in particular dairy products, contain iodine, as well.

Maternal thyroid condition and medications

In a small number of cases, CH occurs when the mother is given anti-thyroid drugs during pregnancy to treat her own thyroid problem.

CONGENITAL HYPOTHYROIDISM (CH)



If CH is not treated, what problems occur?

Most babies do not have symptoms right away because they are protected by their mother's thyroid hormone for a few weeks after birth. After about three to four weeks of age, babies must rely solely on their own thyroid hormone. If they don't make enough, symptoms will show up at that time. A small number of babies with CH do show effects at birth, however.

Some babies have a yellow color to their skin or the whites of their eyes. This is called jaundice. Other signs that may occur in early infancy include:

- Low activity level - babies sleep more than usual and don't move as much
- Poor feeding and poor suck
- Fewer bowel movements or constipation
- Floppy muscle tone (hypotonia)
- Swelling around the eyes and a puffy face
- Large swollen tongue
- Cool, pale, dry skin
- Large soft spot on the skull (the fontanel) that closes late
- Large belly with protruding navel ('umbilical hernia')

If left untreated, babies may develop some or all of the following effects over time:

- Coarse, swollen facial features
- Breathing problems
- Hoarse-sounding cry
- Delays in sitting, crawling, walking, talking (delayed milestones)
- Wide, short hands
- Poor weight gain and growth
- Goiter (enlarged thyroid gland causing a lump in the neck)
- Anemia
- Slow heart rate
- Fluid build-up under the skin (called myxedema)
- Hearing loss

Children who remain untreated usually develop intellectual disabilities and are much shorter than average. They may have spasticity and an unsteady gait. Most have speech delay and some have behavior problems.

What is the treatment for CH?

Your baby's doctor may work with a pediatric endocrinologist, a doctor with training in treating children with thyroid and other hormone problems, to care for your child.

The main treatment for CH is thyroid hormone replacement. It is safe and easy to take. If it is begun immediately after your child is diagnosed, treatment can prevent many or all of the effects of CH. If damage to the brain and nerves happens because treatment is delayed, it is usually permanent and cannot be reversed.

1. Medication

L-thyroxine is a synthetic form of thyroid hormone naturally made by the body. Its chemical structure is identical to that produced by the normal thyroid gland. This medication is given in tablet form to all babies with CH. Your doctor and endocrinologist will decide how much L-thyroxine your baby needs and how often. Your doctors will increase the amount of medication as your child grows. L-thyroxine needs to be taken on a daily basis throughout your child's whole life.

L-thyroxine tablets are small and can be crushed into food or dissolved into a small amount of formula, juice, or other liquid. Do not dissolve them into a full bottle or glass of liquid because your baby may not finish the whole bottle and will not get the full dose of medicine. Young children can easily chew and swallow the pills. There is no approved liquid form of thyroid hormone for babies.

It is important to give your child the correct amount of L-thyroxine. Giving your child more than he or she needs can cause body functions to speed up. Some of the signs that occur when a child takes too much L-thyroxine are:

- Rapid heart rate
- Diarrhea
- Lack of sleep
- Shakiness

Synthetic L-thyroxine is the safest form of medication to use. In the past, before synthetic forms were available, children were treated with dried thyroid hormone from pooled animal tissue. This is called 'desiccated thyroid' and is still available. Do not use desiccated thyroid as the dose of hormone is not consistent.

Soy-based formulas and iron supplements can reduce the amount of thyroid hormone your baby absorbs from the pills. Separate the time you administer your baby's thyroid medication by at least one hour from the time you feed soy formula or iron medication. Tell your doctor if you feed your baby a soy-based formula or iron supplements so the medication can be monitored and increased if necessary.

2. Monitoring

Your child will need regular visits to the doctor to check his or her weight, height, development, and overall health. Your child will also likely need regular blood tests to check the level of thyroid hormone. Blood tests are usually done every one to three months until age one, and then every two to four months until age three. They can usually be done less often after age three.

3. Developmental Evaluation

Your doctor may suggest a formal evaluation of your child's development. If your child shows delays in certain areas of learning or speech, extra help can be arranged. Early intervention programs are available in most states to provide services to children before they reach school age.

What happens when CH is treated?

Children with CH who start treatment soon after birth, usually have normal growth and intelligence and can live typical and healthy lives. Some children, even when treated, have problems with school work and may need extra help. Some may have delayed growth compared to other children their age.

If treatment is not started until several months after birth, delays or learning problems may occur. The level of delay varies from child to child.

How do I know if my child's CH is inherited?

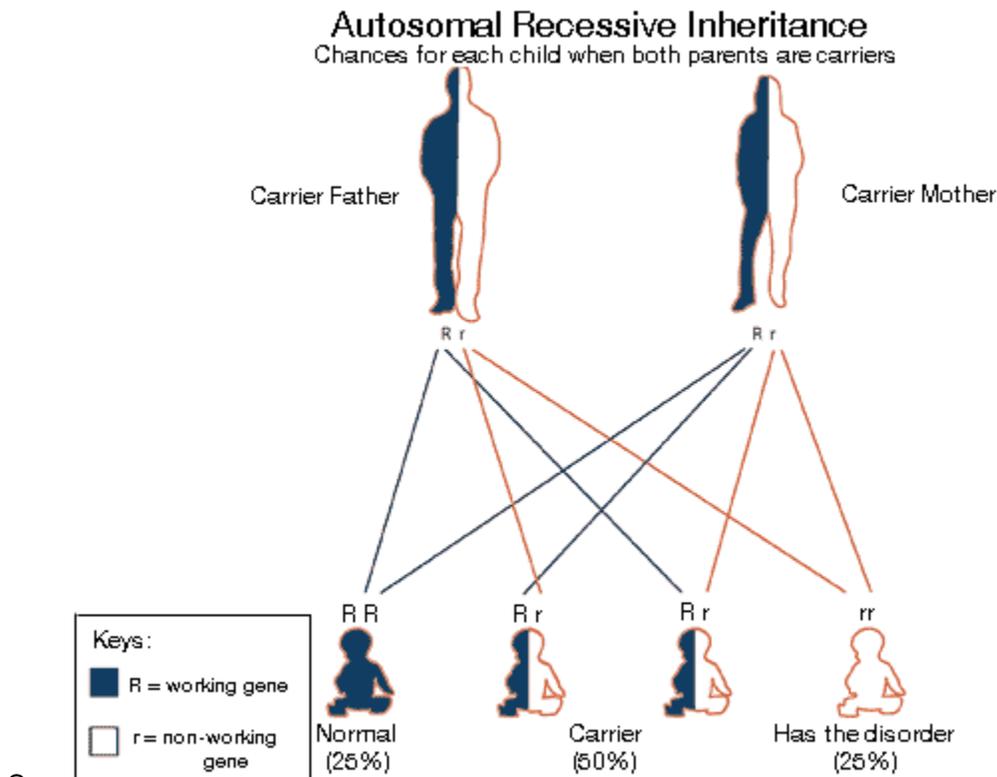
About 80 to 85% of the time, CH is caused when the thyroid gland does not develop at all, is misplaced, or is too small. Most of the time, these cases are not thought to be caused by inherited factors.

In about 15% of cases of CH, the thyroid gland appears normal but the amount of thyroid hormone made is reduced. These cases are more likely to be inherited, but not always. If an inherited type of CH is suspected, you may be referred to a genetic doctor or genetic counselor to determine whether the CH is inherited.

Most of the hereditary types of CH are inherited in an autosomal recessive manner. This type of inheritance affects both boys and girls equally. In children with autosomal recessive CH, a specific pair of genes is not working correctly and too little thyroid hormone is made. These children inherit one non-working gene for the condition from each parent. These children have a normal appearing thyroid that is in the correct place in the neck, but does not make enough thyroid hormone.

Parents of children with autosomal recessive CH rarely have the condition themselves. Instead, each parent has a single non-working gene for CH. They are called carriers. Carriers do not have CH because their other gene is working correctly.

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have CH. There is a 50% chance for the child to be a carrier, just like the parents. And, there is a 25% chance for the child to have two working genes.



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In very rare cases, CH may be inherited in a different way, either by an X-linked recessive or autosomal dominant gene. If your child has one of these rare inherited types of CH, your genetic counselor or genetic doctor will explain how it is inherited and who else in the family may have a chance to pass on the gene for CH.

Genetic counseling is available to families who have children with CH that might be inherited. Genetic counselors can answer your questions about how the CH could be inherited in your family and chances for CH in future offspring. If you have questions, ask your doctor for a referral to a genetic counselor.

If my child might have an inherited type of CH, is genetic testing available?

There are a number of different genes that can contribute to hereditary CH. Some of these are known and some have still not been identified.

If a genetic doctor suspects an inherited type of CH, genetic testing may be available to attempt to determine the gene changes that caused the CH. Genetic testing, also called DNA testing, can be done on a blood sample.

DNA testing is not necessary to diagnose your child. If the gene change or changes are identified in your child, this may be helpful for carrier or prenatal testing, discussed below.

What other testing is available?

If your baby has a positive newborn screen for CH, additional tests must be done before you know for sure that he or she has CH. Blood tests to detect the amount of thyroid hormone (T4) and thyroid stimulating hormone (TSH) are routinely done to confirm the diagnosis of CH. Talk to your doctor if you have questions about testing for CH.

Sometimes an imaging test of the thyroid, either an ultrasound examination or another test called a 'thyroid uptake and scan,' is used to help determine the cause for the CH. This lets doctors see if the thyroid is present, where it is located, and if it is misshapen or smaller than normal.

Can you test during a future pregnancy?

CH is not usually detectable before birth, and most children with CH do not have a hereditary type. For those that may have an inherited type, genetic testing can confirm only a portion of the hereditary cases.

If your child has a hereditary type of CH, and if the gene changes, also called variants, have been found in your child, DNA testing is possible during future pregnancies. However, prenatal testing is rarely done for CH because treatment is so effective. If you have questions about prenatal testing, ask your genetic counselor or physician.

Can other members of the family have CH?

Having CH

If they are healthy and developing normally, older brothers and sisters of a baby with CH are unlikely to have the condition. Talk to your doctor if you have questions about your other children.

Future pregnancies

All 50 US states offer newborn screening for CH. However, when a previous child in the family has had CH, newborn screening results may not be sufficient to rule out the condition in a newborn baby. In this case, special diagnostic testing may be advised in addition to newborn screening.

How many people have CH?

About one in every 2,000 to 4,000 babies born in the United States has CH. Twice as many girls have CH than boys. The reason for this is unknown.

Does CH happen more often in a certain ethnic group?

CH occurs in people of all ethnic groups around the world. It happens more often in babies from parts of the world in which there is not enough iodine in the food and water. It is also more common in babies of Hispanic, Asian, South Pacific, and Native American ancestry. It is less common in babies of African-American ancestry.

Does CH go by any other names?

CH is sometimes also called:

- CHT
- Cretinism
- Endemic Cretinism (iodine deficiency)
- Congenital Myxedema

Where can I find more information?

Genetic Home Reference

<https://ghr.nlm.nih.gov/condition/congenital-hypothyroidism>

MAGIC Foundation (Major Aspects of Growth in Children)

www.magicfoundation.org

American Thyroid Association

<https://www.thyroid.org/>

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