CONDITION

Neonatal hypothyroidism

Neonatal hypothyroidism means a newborn has too little thyroid hormone for healthy growth and brain development.

WHAT IT IS
Low thyroid hormone levels in a newborn

BODY SYSTEM
Hormone system, brain, growth, and metabolism

MAIN SYMPTOMS
Prolonged jaundice, poor feeding, sleepiness, constipation

WHAT CAUSES IT
Thyroid development, inherited, maternal, or temporary factors

HOW IT IS DIAGNOSED
Newborn screening and confirmatory thyroid blood tests

MAIN TREATMENT
Thyroid hormone replacement

CAN IT BE CURED?
Sometimes - temporary forms resolve with age

IS IT AN EMERGENCY?
Sometimes - breathing trouble or extreme difficulty waking

LONG-TERM OUTLOOK
Usually excellent with early, consistent treatment

Overview

Neonatal hypothyroidism means that a baby has too little thyroid hormone during the first weeks of life. It is usually present from birth and is often called congenital hypothyroidism.

The thyroid is a small gland in the neck. It produces hormones that control metabolism, which is how the body uses energy. In a newborn, thyroid hormone is especially important for brain development, growth, body temperature, digestion, and normal activity.

Most affected babies look healthy at birth. Symptoms may not appear until thyroid hormone has been low for several weeks. This is why newborn blood spot screening is important: it can identify possible hypothyroidism before there are obvious signs.

An abnormal screening result does not always mean a baby has permanent hypothyroidism.

Further blood tests are needed. Some babies have a lifelong condition, while others have a temporary form that improves as they mature.

Early treatment gives the developing brain the thyroid hormone it needs. Follow-up remains important because both too little and too much replacement hormone can affect a baby's health.

Symptoms

Many newborns with hypothyroidism have no clear symptoms. Hormone received from the pregnant parent and the baby's own remaining thyroid function may temporarily reduce the signs.

When symptoms occur, they may include:

These signs can have many causes and do not confirm hypothyroidism. Newborn screening and thyroid blood tests are needed to find out whether thyroid hormone levels are low.

- Yellow skin or eyes that remain yellow longer than expected
- Poor feeding or difficulty staying awake for feeds
- Unusual sleepiness or low activity
- Constipation
- A weak or hoarse-sounding cry
- Cool, dry, or mottled skin
- A large tongue or puffy-looking face
- A swollen belly or prominent belly button
- Low muscle tone, meaning the baby feels unusually floppy
- Slow growth or poor weight gain

Causes and risk factors

The most common cause is a thyroid gland that did not develop in its usual position or did not fully form before birth. The gland may be absent, smaller than expected, or located higher in the neck. This usually happens without a clear inherited cause.

Some babies have a normally located thyroid that cannot make hormone properly. These conditions, called thyroid hormone synthesis disorders, are often inherited. A family history of

congenital hypothyroidism or a previous affected child can increase the chance of this type.

Less common causes include problems with the pituitary gland or hypothalamus in the brain. These structures normally signal the thyroid to produce hormone. This is known as central hypothyroidism.

Hypothyroidism can also be temporary. Possible reasons include:

Being born early, having certain chromosome conditions such as Down syndrome, or having other pituitary problems may increase risk. However, neonatal hypothyroidism can occur without any known risk factor.

- Thyroid-blocking antibodies passed across the placenta
- Certain medicines taken during pregnancy
- Too little or too much iodine exposure
- Premature birth or serious newborn illness
- An immature thyroid system that begins working more effectively later

Diagnosis

Newborn blood spot screening is the usual first test. A small blood sample is taken from the baby's heel during the first days after birth. The exact timing and tests differ between screening programs.

Screening usually measures thyroid-stimulating hormone, known as TSH, and may also measure thyroxine, or T4. TSH is made by the pituitary gland and tells the thyroid to work. T4 is one of the main thyroid hormones.

A high TSH level can mean the thyroid is not producing enough hormone. A low T4 result can also suggest hypothyroidism, although it may have other explanations.

An abnormal screen must be followed promptly by a blood sample from a vein. This usually measures TSH and free T4, the portion of T4 available for the body to use. These confirmatory tests help show whether treatment is needed.

Babies born prematurely or seriously unwell may have delayed changes in TSH. Some newborn programs arrange another screening sample for babies at higher risk. Central hypothyroidism may not be detected by screening systems that measure only TSH.

An ultrasound or a nuclear medicine thyroid scan may be used to check the gland's size and location. These tests can help explain the cause, but they should not delay treatment when blood results show that thyroid hormone is needed.

Treatment and management

The main treatment is levothyroxine, a manufactured form of T4 thyroid hormone. It replaces the hormone that the baby's thyroid cannot provide. Treatment begins promptly once confirmatory results show hypothyroidism, sometimes while further testing is still underway.

Levothyroxine is given by mouth. Depending on the product and local practice, a tablet may be crushed and given in a small amount of water or milk, or an appropriate liquid preparation may be used. It should not be mixed into a full bottle because the baby may not finish it and could receive less than intended.

The care team will explain how to give the specific product consistently. Iron, calcium, soy, and some medicines can reduce levothyroxine absorption when given close together. Families should discuss feeds, supplements, and other medicines with the baby's clinical team rather than changing the routine independently.

Regular blood tests measure TSH and free T4. Testing is frequent during infancy because babies grow quickly and their hormone needs change. Monitoring continues through childhood, but visits and blood tests usually become less frequent when levels are stable.

Some children need treatment for life. If the cause is uncertain and permanent hypothyroidism has not been established, the specialist may later arrange a supervised trial without treatment, often after the period of most rapid early brain development. Levothyroxine should never be stopped for this purpose without medical supervision.

Complications

Without enough thyroid hormone, brain development and physical growth can be affected. Delayed treatment or prolonged low hormone levels may contribute to learning difficulties, developmental delay, movement or coordination problems, hearing problems, and reduced growth.

The likelihood and severity of these effects are influenced by how low the hormone level was, how early treatment began, and how consistently levels were controlled. Early diagnosis and effective treatment greatly reduce these risks.

Too much replacement hormone can also cause problems. Possible signs include irritability, poor sleep, sweating, rapid heartbeat, frequent stools, or difficulty gaining weight. Long periods of excessive treatment may affect bone development and growth.

A single unexpected blood result does not necessarily mean harm has occurred. The clinical team will interpret it alongside previous results, growth, symptoms, and how the medicine has

been given.

Prevention

Most cases cannot be prevented. A thyroid gland that is absent, underdeveloped, or unable to make hormone usually results from developmental or inherited factors that parents could not have controlled.

Adequate iodine is needed during pregnancy for thyroid hormone production, but both iodine deficiency and excessive iodine can cause problems. Pregnancy supplements and iodine-containing products should be discussed with a qualified healthcare professional rather than started in high amounts.

People who take thyroid medicine, antithyroid medicine, or medicines that may affect the thyroid need appropriate care during pregnancy. Good pregnancy care may reduce some temporary causes, but it cannot prevent most cases.

Newborn screening does not prevent hypothyroidism. It helps prevent avoidable complications by finding affected babies before symptoms become clear.

Living with it and what to expect

During the first months, daily medicine and frequent blood tests can feel demanding. A consistent routine often makes treatment easier. Families may use reminders or keep a record of doses, especially when several people care for the baby.

Most babies treated early grow and develop normally. They can usually feed, play, attend school, exercise, and take part in everyday activities without special restrictions. Thyroid hormone replacement does not sedate a child or change their personality; it supplies a hormone the body needs.

Medicine amounts may change as the child gains weight and grows. Illness, feeding changes, missed doses, or switching between formulations can sometimes affect blood results. Families should tell the care team if doses have been missed rather than trying to compensate without advice.

Development, hearing, growth, and progress at school may be reviewed over time. This does not mean that difficulties are expected. It allows any concern to be recognised and supported early.

Temporary hypothyroidism may resolve in infancy or early childhood. Permanent forms do not

usually go away, but they can generally be controlled with lifelong replacement hormone. No single course can be promised, and ongoing blood tests show whether treatment remains appropriate.

When to get medical help

An abnormal newborn screening result needs prompt follow-up. Contact the newborn screening team, maternity service, or baby's clinician the same day if you were told that confirmatory testing is needed but have not received a clear plan. This is urgent follow-up, although the baby may appear well and may not need an emergency department.

Seek prompt medical advice if a baby:

Call emergency services if the baby has trouble breathing, turns blue or grey, has a seizure, becomes limp, or is extremely difficult to wake. These signs can occur with many serious newborn conditions and need immediate assessment.

Routine questions about giving medicine, arranging blood tests, feeds, supplements, or mild constipation can usually be discussed with the baby's regular clinical team during office hours.

- Is feeding much less than usual
- Is becoming increasingly sleepy
- Has repeated vomiting or cannot keep medicine down
- Develops worsening or prolonged jaundice
- Misses several doses of levothyroxine
- Has possible signs of too much hormone, such as persistent rapid heartbeat or marked irritability

Common questions

Is neonatal hypothyroidism the same as congenital hypothyroidism?

They are often used to describe the same condition. "Neonatal" refers to hypothyroidism during the newborn period. "Congenital" means it was present at birth. Occasionally, low thyroid hormone is first recognised shortly after birth because of temporary illness or another cause.

Will my baby look unwell?

Not necessarily. Many affected babies appear healthy and have no obvious symptoms. That is

why screening is more reliable than appearance alone. A baby who looks well still needs prompt confirmatory testing after an abnormal screen.

Will my baby need thyroid medicine for life?

Some babies will, especially when the thyroid is absent, severely underdeveloped, or unable to make hormone. Others have temporary hypothyroidism. If it is unclear, a specialist can reassess the need for treatment when it is developmentally safe to do so.

Can I breastfeed a baby taking levothyroxine?

Yes. Breast milk does not replace the baby's prescribed thyroid hormone, but breastfeeding can usually continue. The medicine should be given as instructed so the full amount is received consistently.

What happens if a dose is missed?

Follow the instructions provided by the baby's clinical team or pharmacist. Do not give extra medicine unless they advise it. One missed dose is not usually an emergency, but repeated missed doses can leave hormone levels too low and should be discussed promptly.

Can neonatal hypothyroidism affect intelligence?

Severe or prolonged thyroid hormone deficiency can affect brain development. When the condition is found through newborn screening and treated promptly, most children have typical development. Regular testing helps keep hormone levels in the intended range.

Sources

- American Academy of Pediatrics. Congenital Hypothyroidism: Screening and Management . Pediatrics . 2023;151(1):e2022060420.
- van Trotsenburg P, Stoupa A, Léger J, et al. Congenital Hypothyroidism: A 2020-2021 Consensus Guidelines Update . Thyroid . 2021;31(3):387-419.
- UK National Screening Committee. Newborn blood spot screening: congenital hypothyroidism . UK Government.
- Rastogi MV, LaFranchi SH. Congenital hypothyroidism . Orphanet Journal of Rare Diseases . 2010;5:17.
- Great Ormond Street Hospital for Children. Congenital hypothyroidism . Patient information.