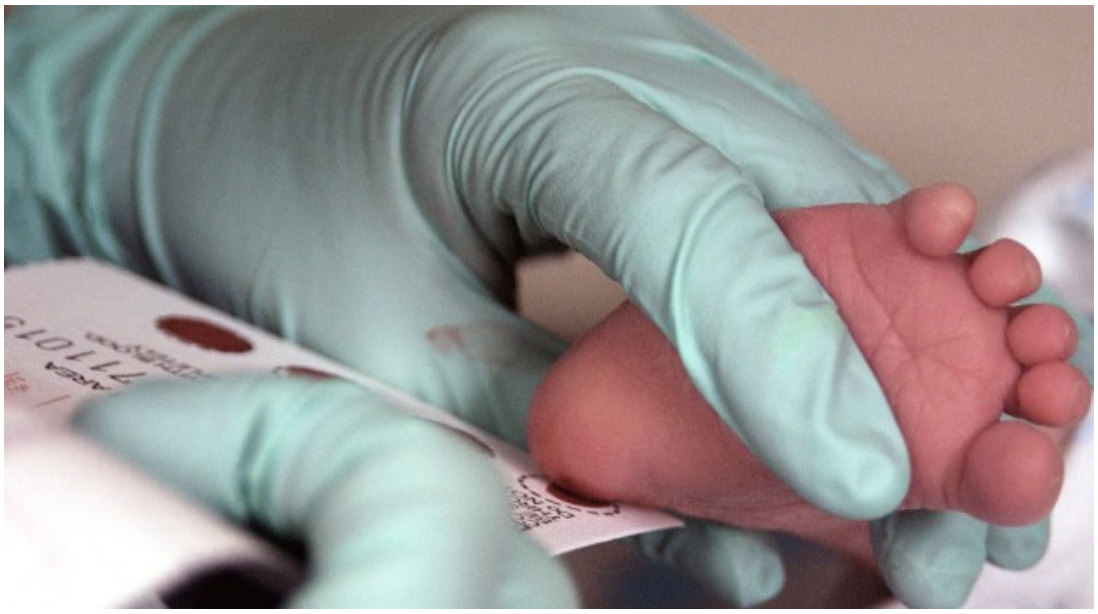
Congenital Hypothyroidism — Causes and Treatment

See online here

Congenital hypothyroidism is a condition caused by lack of thyroid hormones. This condition is found in newborn infants. If left untreated, it may lead to severe complications including permanent intellectual disability or growth failure.



Definition and Background of Congenital Hypothyroidism

Congenital hypothyroidism is the deficiency of thyroid hormones in neonates and infants.

Cretinism is severe, untreated congenital hypothyroidism; it results in short stature (dwarfism) and intellectual disabilities.

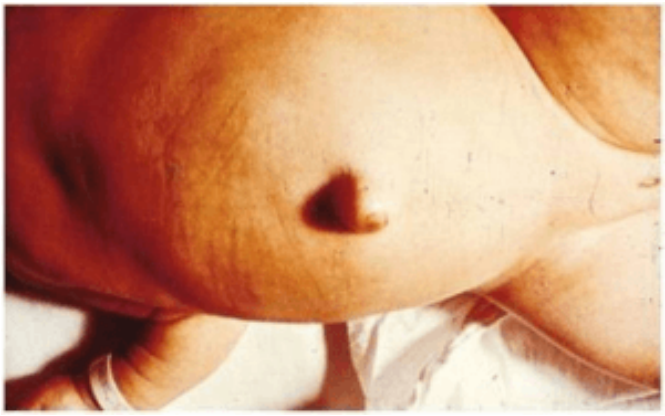


Image: “3-month-old infant with untreated CH; close up showing abdominal distension and umbilical hernia” by Rastogi and LaFranchi. License: CC BY 2.5

Congenital hypothyroidism is one of the most common preventable causes of intellectual disabilities. Hence, early diagnosis and treatment are important to decrease morbidity from this condition.

Congenital hypothyroidism may occur as a result of an inborn error of the thyroid metabolism, iodine deficiency, or a structural defect in the thyroid gland.

Etiology and Pathophysiology of Congenital Hypothyroidism

Anatomic defects of the thyroid gland

The thyroid gland develops from the fourth branchial pouch in the buccopharyngeal cavity during the fourth to tenth week of gestation. It later migrates toward the neck to rest at its anatomic position. An error in the migration or formation of the thyroid tissue (dysplasia, aplasia, or ectopy) may result in congenital hypothyroidism.

Iodine deficiency

Iodine is required for the production of thyroid hormones triiodothyronine (T3) and thyroxine (T4) from the thyroid gland. A deficiency of iodine, therefore, results in decreased production of thyroid hormones.

Inborn errors of thyroid metabolism

Children with normal anatomic thyroid glands may still suffer from congenital hypothyroidism due to other inborn errors of thyroid metabolism. These inborn errors may include:

- Lack of iodide uptake
- Abnormalities in TSH receptors
- Defects in thyroglobulin
- Deiodinase defect
- Defects in thyroid peroxidase

Epidemiology of Congenital Hypothyroidism

In the United States, it is estimated that the annual incidence of congenital hypothyroidism is approximately **1 in 4,000 births**. Data from a number of other countries indicates a similar incidence, at approximately 1 in 3,000–4,000 births. However, the incidence is higher in some Middle Eastern countries, at approximately 1 in 1,400–2,000 births.

Racial and gender disparities have also been noted; for example, the incidence of congenital hypothyroidism is higher among Hispanic females. It is also higher in girls than boys, with a female-to-male ratio of 2:1. Twin births appear to have a 12-times-higher risk of developing the condition.

Clinical Presentation of Congenital Hypothyroidism

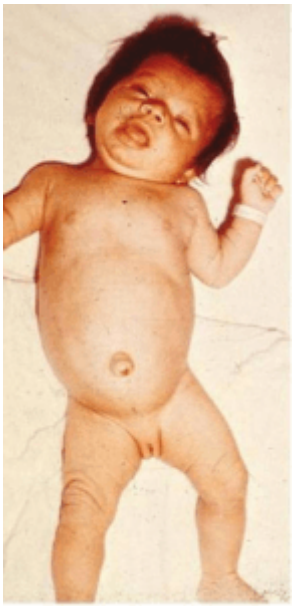


Image: “3-month-old infant with untreated CH; picture demonstrates hypotonic posture, myxedematous facies, macroglossia, and umbilical hernia” by Rastogi and LaFranchi. License: CC BY 2.5

Newborn infants with congenital hypothyroidism are usually born at normal gestational term or even post-term and may be initially asymptomatic, as maternal hormones pass on to the fetus through the placenta.

Birth length and weight are usually within the normal range, although head circumference may be increased. The knee epiphyses often lack calcification. Common clinical features include:

- Poor growth
- Small stature
- Poor feeding resulting in poor weight gain
- Decreased activity
- Constipation
- Hoarse cry
- Jaundice

Affected babies often sleep excessively and rarely cry, and are thus labeled as ‘good babies.’

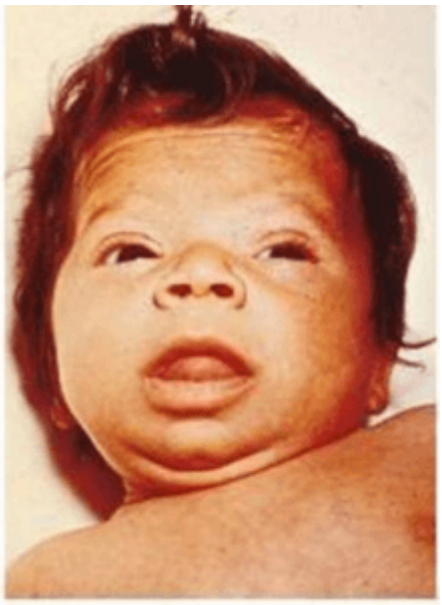


Image: “3-month-old infant with untreated CH; close up of face, showing myxedematous facies, macroglossia, and skin mottling” by Rastogi and LaFranchi. License: CC BY 2.5

On physical examination, the following signs may be present:

- **Macroglossia** and coarse facial features
- **Hypotonia**
- Large anterior fontanelle
- **Umbilical hernia**
- Slow development
- Goiter
- Pallor
- **Myxedema**
- Dry skin

Screening newborns for congenital hypothyroidism

In the United States and many other developed countries, screening for congenital hypothyroidism is performed during the first week of life, which identifies those children suffering from the condition during this asymptomatic stage. A careful history and physical examination should be performed and testing should be repeated in order to confirm the diagnosis.

Early diagnosis and treatment of congenital hypothyroidism are important to prevent long-term mental disability from this condition, as thyroid hormones are crucial for normal growth and maturation of the brain.

Differential Diagnosis of Congenital

Hypothyroidism

- Panhypopituitarism
- Thyroxine-binding globulin deficiency
- Pediatric hypopituitarism
- Beckwith-Wiedemann syndrome

Diagnosis of Congenital Hypothyroidism

Laboratory studies

- Elevated levels of thyroid-stimulating hormone (TSH)
- Decreased levels of total and free thyroid hormone (T4)
- Neonatal and maternal antithyroid antibodies may be found

Imaging studies

- **Technetium-99m** or **iodine-123** may be useful in determining the cause of hypothyroidism
- Ectopic thyroid may be demonstrated by thyroid scans.
- **Ultrasonography** may be used as an alternative to thyroid scans

Management of Congenital Hypothyroidism

Medical care

Early diagnosis of the condition is very important for management. Medical treatment involves **thyroid hormone replacement** (oral levothyroxine is used).

Diet

In endemic areas where iodine deficiency is common, **dietary iodide supplementation** is advised and may help prevent this condition.

Prognosis

Prognosis is excellent if treatment is timely. Most children achieve normal physical and mental growth status. Regular follow-ups and blood thyroid hormone measurements are needed for optimum management.

Review Questions

The correct answers can be found below the references.

1. A 9-week-old female infant who was born at home is brought to the pediatrician by her mother. The mother notes that the child always feels cold to the touch, sleeps too much and that her bowel movements are infrequent. On physical examination, the pediatrician finds mild jaundice and hypotonia. Which of the following lab findings would you expect to see in this infant?

A. Inhibitory anti-TSH receptor antibodies

B. Normal TSH, elevated reverse T3, and decreased total T4 and T3
C. Elevated TSH, and decreased free and total T4
D. Decreased TSH, and elevated free and total T4
E. Stimulatory anti-TSH receptor antibodies

2. An obstetrician who works in a developing country delivers a baby who is suspected to have congenital hypothyroidism due to maternal iodine deficiency. Which sign of the following would you NOT expect to find in this newborn?

A. Umbilical hernia
B. Macroglossia
C. Hypotonia
D. Diarrhea
E. Mild jaundice

3. A 7-week-old American infant was born at term, following a normal and uncomplicated pregnancy. His chart reveals that he had indirect hyperbilirubinemia at birth. According to his mother, he suffers from poor feeding. On physical examination, the pediatrician notices that the infant suffers from hypotonia, lethargy, coarse facial features, and a protruding, large tongue. What is the most likely cause of this infant's presentation?

A. Rett syndrome
B. Toxoplasmosis
C. Iodine deficiency
D. Thyroid dysgenesis
E. Trisomy 21

References

Maala S Daniel. Congenital Hypothyroidism. Medscape.

Congenital hypothyroidism. Wikipedia.

Hypothyroidism. Mayo Clinic.

Correct answers: 1C, 2D, 3D

Notes