Congenital Hypothyroidism — Causes and Treatment

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

Definition and Background

**Congenital Hypothyroidism**: The deficiency of thyroid hormones in neonates and infants.

**Cretinism**: Severe untreated congenital hypothyroidism resulting in short stature (dwarfism) and mental retardation.

The congenital hypothyroidism is one of the most common preventable causes of intellectual disability (mental retardation). Hence, the early diagnosis and treatment are important to decrease the morbidity from this condition.

The 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

Anatomic defects of the thyroid gland

The thyroid gland develops from the fourth branchial pouch in the buccopharyngeal cavity during fourth to the tenth week of gestation. It later migrates towards 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. Subsequently, a deficiency of iodine 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

In the United States, it is estimated that the annual incidence of congenital hypothyroidism is approximately 1 in 4000 births. The international data from different countries indicates the similar incidence i.e. approximately 1 in 3000–4000 births. However, the incidence is higher in some Middle East countries i.e. 1 in 1400–2000.

The racial and gender preponderance has also been noted. The incidence is higher among Hispanic females. It also appears to be higher in females in comparison to males with a female-to-male ratio of 2:1. Twin births appear to have 12 times higher risk of developing the condition.

Clinical Presentation
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. The common clinical features include:

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

The babies often sleep excessively and rarely cry, and are labeled as “good babies”.
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 countries of the Western world, the screening for congenital hypothyroidism is performed during the first week of life and most of the newborn infants are usually identified 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 the 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
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 in preventing the condition.

Prognosis
The prognosis is excellent if timely treated. Most of the children achieve normal physical and mental growth. 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 newborn infant, who was born at home, is brought to the pediatrician by her mother. This is because she noticed that: she always feels cold, she sleeps too much, and her bowel movements are not very frequent. By physical examination, the pediatrician finds mild jaundice and hypotonia. Which of the following lab findings would you expect to see in this newborn?
   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 you would 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. By physical examination, you notice that he suffers from hypotonia, lethargy, coarse facial features, and a protruding large tongue. What is the most likely cause of his presentation?

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

References

2. Congenital hypothyroidism. Wikipedia.

Correct answers: 1C, 2D, 3D

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