CONGENITAL HYPOTHYROIDISM: IMPORTANCE OF EARLY DIAGNOSIS AND TREATMENT

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Abstract. *Congenital hypothyroidism (CH) is a prevalent endocrine disorder in newborns, primarily caused by thyroid dysgenesis or dyshormonogenesis. The deficiency of thyroid hormones from birth can severely impair neurological development and physical growth if left untreated. Since clinical signs may be subtle or absent in the neonatal period, universal newborn screening programs are vital for early detection. Timely diagnosis and prompt initiation of levothyroxine therapy can prevent irreversible cognitive impairment and ensure normal development. This article highlights the etiology, clinical presentation, diagnostic approach, treatment strategies, and prognosis of CH.*

Keywords: *congenital hypothyroidism, neonatal screening, thyroid hormones, levothyroxine, early diagnosis.*

Introduction

Congenital hypothyroidism (CH) is one of the most common preventable causes of intellectual disability in children. It is defined as thyroid hormone deficiency present at birth.

The condition can result from absent, underdeveloped, or ectopically located thyroid gland (thyroid dysgenesis), or from defects in hormone synthesis (dyshormonogenesis). Early diagnosis and intervention are critical, as untreated CH can lead to irreversible mental retardation, poor growth, and delayed development. The implementation of neonatal screening programs worldwide has significantly improved outcomes.

Etiology and Pathogenesis

CH can arise from various anatomical and functional abnormalities. The most frequent cause (approximately 85%) is thyroid dysgenesis, including agenesis, hypoplasia, or ectopia. Dyshormonogenesis, often inherited in an autosomal recessive manner, involves enzymatic defects in thyroid hormone production. Central hypothyroidism, due to pituitary or hypothalamic dysfunction, is less common. In some regions, iodine deficiency during pregnancy remains a major risk factor.

Clinical Manifestations

Symptoms of CH in newborns are often mild or nonspecific. Common features include prolonged jaundice, constipation, lethargy, feeding difficulties, macroglossia, umbilical hernia, and hypotonia. In the absence of early treatment, infants may develop coarse facial features, delayed milestones, and severe intellectual disability. Due to this subtle presentation, clinical suspicion alone is insufficient; hence, the need for routine screening.

Diagnosis

Diagnosis is primarily based on newborn screening using TSH and/or T4 levels. Elevated TSH with low T4 confirms primary hypothyroidism. Follow-up tests may include serum free T4, total T4, and thyroid ultrasonography or scintigraphy to determine the gland's presence and position. Early diagnosis within the first two weeks of life is essential to initiate timely treatment and prevent complications.

Treatment

Levothyroxine is the treatment of choice. Therapy should start as soon as CH is confirmed, ideally within the first 14 days of life. The recommended starting dose is 10–15 μg/kg/day, adjusted based on TSH and free T4 levels. Regular follow-up and dosage adjustments are essential to ensure normal growth and neurodevelopment. Treatment may be lifelong, though some cases of transient CH can be re-evaluated at 3 years of age to confirm persistence.

Prognosis

When diagnosed early and treated adequately, children with CH can achieve normal intellectual and physical development. Delayed diagnosis and insufficient treatment increase the risk of permanent cognitive deficits, growth retardation, and motor impairments. Public health efforts should focus on universal screening, parental education, and healthcare provider training to improve outcomes.

Conclusion

Congenital hypothyroidism is a serious but manageable condition. Universal neonatal screening and early initiation of levothyroxine therapy are critical steps to prevent irreversible complications. With timely intervention and proper monitoring, affected children can lead healthy, normal lives.

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