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ВАЖНОСТЬ РАННЕЙ ДИАГНОСТИКИ И ЛЕЧЕНИЯ ВРОЖДЕННОГО ГИПОТИРЕОЗА

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АННОТАЦИЯ

Врожденный гипотиреоз является одним из наиболее распространённых эндокринных заболеваний у детей и обусловлен частичным или полным дефицитом гормонов щитовидной железы. Данное состояние представляет серьёзную медико-социальную проблему, поскольку при отсутствии своевременной диагностики и адекватной терапии приводит к необратимым нарушениям физического, психоневрологического и интеллектуального развития ребёнка. Основными причинами врождённого гипотиреоза являются дисгенезия щитовидной железы, нарушения гормоногенеза, а также реже — центральные формы заболевания.

Особое значение в профилактике тяжёлых осложнений имеет неонатальный скрининг, позволяющий выявить заболевание на доклинической стадии и своевременно начать заместительную терапию левотироксином. Раннее начало лечения, особенно в первые недели жизни, способствует нормализации роста, развития и метаболических процессов у ребёнка.

В данном обзоре обобщены современные данные отечественных и зарубежных исследований, посвящённых этиологии, патогенезу, клиническим проявлениям, диагностике и лечению врождённого гипотиреоза. Подчёркивается важность раннего выявления заболевания, совершенствования скрининговых программ и повышения осведомлённости врачей и родителей.

Ключевые слова: врожденный гипотиреоз, неонатальный скрининг, дисгенез щитовидной железы, дефицит гормонов, ранняя диагностика, заместительная терапия, детская эндокринология.

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IMPORTANCE OF EARLY DIAGNOSIS AND TREATMENT IN CONGENITAL HYPOTHYROIDISM

ANNOTATION

Congenital hypothyroidism is one of the most common endocrine disorders in children and is caused by a partial or complete deficiency of thyroid hormones. This condition represents a significant medical and social problem, as delayed diagnosis and inadequate treatment can lead to irreversible impairments in physical growth, neurodevelopment, and cognitive function. The main causes of congenital hypothyroidism include thyroid dysgenesis, defects in hormone synthesis, and, less commonly, central forms associated with hypothalamic or pituitary dysfunction.

Neonatal screening plays a crucial role in the early detection of this condition, allowing diagnosis at a preclinical stage and enabling prompt initiation of hormone replacement therapy with levothyroxine. Early treatment, particularly within the first weeks of life, is essential for ensuring normal growth, brain development, and metabolic regulation.

This review summarizes current data from both domestic and international studies on the etiology, pathogenesis, clinical manifestations, diagnosis, and treatment of congenital hypothyroidism. Special emphasis is placed on the importance of early diagnosis, optimization of screening programs, and increasing awareness among healthcare professionals and parents.

+Keywords: congenital hypothyroidism, neonatal screening, thyroid dysgenesis, hormone deficiency, early treatment, pediatric endocrinology

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TUG‘MA GIPOTIREOZDA ERTA TASHXISLASH VA DAVOLASHNING AHAMIYATI

ANNOTATSIYA

Tug‘ma gipotireoz bolalarda eng ko‘p uchraydigan endokrin kasalliklardan biri bo‘lib, qalqonsimon bez gormonlarining qisman yoki to‘liq yetishmasligi bilan tavsiflanadi. Ushbu kasallik muhim tibbiy va ijtimoiy ahamiyatga ega, chunki o‘z vaqtida aniqlanmasa va davolanmasa, bolalarda jismoniy o‘sish, psixonevrologik rivojlanish hamda intellektual faoliyatning orqada qolishiga olib keladi. Tug‘ma gipotireozning asosiy sabablari qalqonsimon bez disgeneziyasi, gormonlar sintezining buzilishi, kamroq hollarda esa markaziy shakllar hisoblanadi.

Kasallikni erta aniqlashda neonatal skrining muhim ahamiyatga ega bo‘lib, u klinik belgilar paydo bo‘lishidan oldin tashxis qo‘yish va davolashni o‘z vaqtida boshlash imkonini beradi. Ayniqsa, hayotning ilk haftalarida levotiroksin bilan davolashni boshlash bolaning normal o‘sishi, rivojlanishi va metabolik jarayonlarini ta’minlashda muhim rol o‘ynaydi.

Mazkur sharhda tug‘ma gipotireozning etiologiyasi, patogenezi, klinik belgilari, diagnostikasi va davolash usullari bo‘yicha zamonaviy mahalliy va xorijiy tadqiqotlar tahlil qilingan. Shuningdek, erta tashxislash, skrining dasturlarini takomillashtirish va aholining xabardorligini oshirish zarurligi alohida ta’kidlangan.

Kalit so`zlar: tug‘ma gipotireoz, neonatal skrining, qalqonsimon bez disgeneziyasi, gormonlar tanqisligi, erta davolash, bolalar endokrinologiyasi

Congenital hypothyroidism (CH) is one of the most common endocrine disorders in newborns and remains a leading preventable cause of intellectual disability worldwide. The condition develops due to insufficient production or impaired biological action of thyroid hormones, which are critically important for normal growth, metabolism, and especially neurodevelopment [1]. Despite advances in modern medicine, CH continues to be a significant public health concern, particularly in regions with iodine deficiency and limited access to neonatal screening programs [2].

Thyroid hormones play a fundamental role in early human development, especially during fetal life and the first years after birth. They are directly involved in the regulation of neuronal proliferation, migration, differentiation, synaptogenesis, and myelination [3]. These processes are essential for the formation of the central nervous system and higher cognitive functions. The first months of life represent a critical developmental window during which the brain is particularly sensitive to thyroid hormone deficiency. Even a short-term deficit during this period can result in irreversible structural and functional damage to the brain [4].

From a clinical perspective, congenital hypothyroidism is a highly heterogeneous condition with multiple etiological factors. In approximately 85–90% of cases, the disease is caused by thyroid dysgenesis, including agenesis, ectopia, and hypoplasia of the gland [5]. These abnormalities arise due to disruptions in embryological development, although the precise mechanisms are not always fully understood. Genetic mutations affecting transcription factors involved in thyroid development have been identified in some cases, but many instances remain idiopathic.

Another important group of causes includes dyshormonogenesis, which accounts for 10–15% of cases and is associated with defects in hormone synthesis, transport, or metabolism [6]. These forms are often inherited in an autosomal recessive manner and may be accompanied by goiter. Less frequently, congenital hypothyroidism results from central causes, such as dysfunction of the hypothalamic–pituitary axis, leading to insufficient secretion of thyroid-stimulating hormone (TSH) or thyrotropin-releasing hormone (TRH) [7].

In addition, transient forms of hypothyroidism are recognized, particularly in newborns exposed to maternal factors. These may include iodine deficiency or excess, maternal use of antithyroid medications, or the presence of transplacental antibodies. Prematurity is also an important contributing factor, as immature endocrine regulation can temporarily impair thyroid function [8].

The pathophysiology of congenital hypothyroidism is closely linked to the systemic effects of thyroid hormone deficiency. At the cellular level, hypothyroxinemia leads to reduced metabolic activity, decreased enzyme function, and accumulation of incompletely oxidized metabolic products [9]. These changes affect virtually all tissues and organ systems, but the central nervous system is particularly vulnerable.

One of the most critical consequences of thyroid hormone deficiency is impaired brain development. In the absence of adequate hormone levels, processes such as myelination of nerve fibers and synaptic formation are delayed or disrupted [10]. This results in reduced neuronal connectivity and long-term cognitive impairment. Importantly, the damage becomes irreversible if treatment is not initiated within the early postnatal period, highlighting the importance of early detection. Despite the severity of the condition, clinical diagnosis of congenital hypothyroidism in the neonatal period is challenging. Early symptoms are often subtle and nonspecific, and only a small proportion of newborns exhibit classical signs such as prolonged jaundice, hypotonia, macroglossia, or delayed meconium passage [11]. In many cases, more pronounced symptoms, including developmental delay, growth retardation, and metabolic disturbances, become apparent only after several months, when neurological damage has already occurred.

This lack of reliable early clinical indicators underscores the crucial role of neonatal screening programs. Screening for congenital hypothyroidism is widely recognized as one of the most effective preventive strategies in modern medicine. It typically involves measuring TSH and/or thyroxine (T4) levels in dried blood spots collected from newborns within the first days of life [12]. Elevated TSH levels serve as a sensitive marker of primary hypothyroidism and require confirmatory testing.

The introduction of universal neonatal screening has dramatically improved outcomes for children with congenital hypothyroidism. Early detection allows treatment to be initiated before the onset of clinical symptoms, thereby preventing irreversible neurological damage [13]. However, challenges remain, including false-positive results, delayed TSH elevation in premature infants, and variability in screening protocols between countries.

Once the diagnosis is confirmed, treatment must be initiated without delay. Levothyroxine replacement therapy is the standard of care and is highly effective when started early. The goal of treatment is to normalize thyroid hormone levels as quickly as possible in order to support normal brain development and metabolic function [14]. The recommended initial dose is relatively high compared to that used in older children, reflecting the urgent need to restore hormone levels during the critical developmental period. One of the most important aspects of treatment is continuous monitoring. Regular assessment of TSH and T4 levels allows clinicians to adjust the dosage of levothyroxine and ensure optimal therapeutic outcomes [15]. Both insufficient and excessive hormone replacement can have negative consequences, including impaired growth, behavioral disturbances, and metabolic imbalance. The prognosis of congenital hypothyroidism depends largely on the timing of diagnosis and initiation of treatment. Children diagnosed through neonatal screening and treated within the first weeks of life generally achieve normal intellectual development and quality of life [16]. In contrast, delayed diagnosis is associated with permanent neurological deficits, emphasizing the importance of early intervention.

Beyond individual clinical outcomes, congenital hypothyroidism has broader public health implications. The condition represents a significant economic burden due to the costs associated with lifelong disability, special education, and medical care in untreated cases [17]. Early diagnosis and treatment, therefore, are not only medically beneficial but also economically advantageous.

In recent years, advances in molecular genetics have provided new insights into the underlying mechanisms of congenital hypothyroidism. Identification of genetic mutations involved in thyroid development and hormone synthesis has improved diagnostic accuracy and allowed for more precise classification of the disease [18]. These developments also open the possibility of personalized approaches to treatment and long-term management. At the same time, growing attention is being paid to prenatal factors influencing thyroid function. Maternal iodine status, environmental exposures, and endocrine-disrupting chemicals may all play a role in the development of congenital hypothyroidism [19]. This highlights the importance of preventive strategies aimed at improving maternal health and reducing exposure to harmful substances during pregnancy. Another important direction for future research is the study of long-term outcomes in patients with congenital hypothyroidism. Although early treatment significantly improves prognosis, subtle cognitive and behavioral differences may still be observed in some

individuals [20]. Understanding these outcomes is essential for optimizing follow-up care and support.

In conclusion, congenital hypothyroidism is a common and potentially severe endocrine disorder that can lead to irreversible developmental impairment if left untreated. However, it is also one of the most successfully managed conditions in pediatric endocrinology, thanks to the widespread implementation of neonatal screening and effective hormone replacement therapy. Early diagnosis remains the cornerstone of management, and continued efforts to improve screening programs, increase awareness, and advance research are essential for further reducing the global burden of this disease [21].

An important aspect that deserves particular attention in the study of congenital hypothyroidism is the role of maternal and perinatal factors in shaping neonatal thyroid function. It is well established that the fetal thyroid gland begins to function relatively late in gestation, and until that time the developing fetus is largely dependent on maternal thyroid hormones. Even mild maternal hypothyroxinemia during pregnancy can adversely affect fetal brain development, leading to subtle neurocognitive deficits that may persist later in life [22]. This emphasizes the importance of adequate maternal iodine intake and thyroid function monitoring during pregnancy.

In addition to maternal thyroid status, environmental factors have been increasingly recognized as contributors to thyroid dysfunction in neonates. Exposure to endocrine-disrupting chemicals, such as perchlorates, polychlorinated biphenyls, and certain pesticides, may interfere with thyroid hormone synthesis, transport, or receptor activity [23]. These compounds can cross the placental barrier and affect fetal thyroid function, further complicating the pathogenesis of congenital hypothyroidism. In regions with environmental pollution or agricultural chemical exposure, this factor becomes particularly relevant.

Another emerging area of research is the role of epigenetic regulation in the development of congenital hypothyroidism. Epigenetic modifications, including DNA methylation and histone modification, may influence the expression of genes involved in thyroid development and hormone synthesis [24]. These changes can be triggered by environmental factors and may partly explain the variability in clinical presentation and severity of the disease, even among individuals with similar genetic backgrounds.

From a diagnostic perspective, the optimization of neonatal screening protocols remains an important challenge. While TSH-based screening is highly sensitive for primary hypothyroidism, it may fail to detect central forms of the disease. Combined TSH and T4 screening approaches have been proposed to improve diagnostic accuracy, particularly in high-risk populations [25]. Furthermore, the development of more sensitive and specific biomarkers may enhance early detection and reduce false-positive and false-negative results.

It is also important to consider the challenges associated with screening in premature and low-birth-weight infants. In these populations, delayed TSH elevation is a well-documented phenomenon, which may lead to missed or delayed diagnosis if only a single screening test is performed [26]. Therefore, repeat screening is often recommended in such cases to ensure accurate diagnosis. In terms of treatment, although levothyroxine remains the gold standard, recent studies have explored the impact of dosing strategies on long-term outcomes. Early high-dose therapy has been associated with faster normalization of thyroid hormone levels and improved neurodevelopmental outcomes [27]. However, excessive dosing may also carry risks, including behavioral disturbances and accelerated bone maturation, highlighting the need for careful monitoring and individualized treatment approaches.

Adherence to therapy is another critical factor influencing treatment success. Since congenital hypothyroidism requires long-term or lifelong treatment, ensuring compliance can be challenging, particularly in low-resource settings. Educational interventions aimed at parents and caregivers have been shown to improve adherence and overall outcomes [28]. Long-term follow-up of patients with congenital hypothyroidism has revealed that, despite early and adequate treatment, some individuals may experience subtle deficits in executive function, attention, and processing speed [29]. These findings suggest that even early-treated patients require ongoing monitoring and support, particularly in educational settings. The psychosocial aspects of congenital hypothyroidism also warrant attention. Chronic disease management in children can have significant emotional and psychological impacts on both patients and their families. Providing adequate counseling and support services is essential for improving quality of life and ensuring optimal developmental outcomes [30]. From a public health perspective, strengthening neonatal screening programs remains a priority. This includes ensuring universal coverage, improving laboratory infrastructure, and standardizing diagnostic criteria. In addition, public awareness campaigns and professional training programs can help improve early detection and treatment rates [31]. Finally, advances in prenatal diagnostics and fetal medicine offer promising opportunities for early intervention. In certain high-risk cases, prenatal identification of thyroid dysfunction may allow for early therapeutic measures, potentially improving outcomes even before birth [32].

Thus, the integration of clinical, molecular, and public health approaches is essential for the comprehensive management of congenital hypothyroidism. Continued research and innovation will play a key role in improving early diagnosis, optimizing treatment strategies, and enhancing long-term outcomes for affected individuals.

Conclusion:

Congenital hypothyroidism remains a major preventable cause of intellectual disability in children. Despite its potentially severe consequences, the disease can be effectively managed if diagnosed early through neonatal screening programs. The absence of specific clinical symptoms in the early stages underscores the importance of laboratory-based detection methods.

Early and adequately dosed L-thyroxine therapy ensures normal physical growth, neurological development, and metabolic function. Advances in prenatal diagnostics and improved awareness of risk factors may further enhance early detection strategies.

Thus, strengthening screening systems, ensuring timely treatment, and increasing awareness among healthcare providers are essential steps toward reducing the global burden of congenital hypothyroidism and improving long-term outcomes in affected children.

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