

Congenital Hypothyroidism in Newborns: Early Detection, Clinical Implications, and Screening Strategies

Shaira Khalilovna Atadjanova¹, Barno Shamsuddinovna Makhmudova²

Abstract

Congenital hypothyroidism (CH) is one of the most common preventable causes of intellectual disability worldwide. It results from inadequate production of thyroid hormones at birth, which are essential for normal brain maturation and physical growth. The global incidence ranges from approximately 1 in 2,000 to 1 in 4,000 live births, with regional variability influenced by iodine status and genetic factors. Most affected newborns appear clinically normal at birth, making early detection through neonatal screening critical. Without timely treatment, CH may lead to irreversible neurodevelopmental impairment, growth retardation, and metabolic disturbances. Measurement of thyroid-stimulating hormone (TSH) and/or thyroxine (T4) levels within the first days of life forms the cornerstone of screening programs. Early initiation of levothyroxine therapy significantly improves cognitive outcomes and ensures normal growth. This article reviews the epidemiology, etiology, clinical features, screening protocols, and management strategies for congenital hypothyroidism, emphasizing the importance of universal newborn screening and early therapeutic intervention.

Keywords: Congenital hypothyroidism, Newborn screening, Thyroid-stimulating hormone, Neonatal endocrinology, Levothyroxine therapy, Iodine deficiency.

^{1,2} Andijan State Institute Department of the Faculty of Pediatrics and Neonatology

Introduction. Congenital hypothyroidism (CH) is a neonatal endocrine disorder characterized by insufficient thyroid hormone production at birth. Thyroid hormones—particularly thyroxine (T4) and triiodothyronine (T3)—play a crucial role in central nervous system development, myelination, and somatic growth during early life. Even short-term deficiency in the neonatal period can result in permanent neurocognitive impairment.

Globally, CH occurs in approximately 1:2,000–1:4,000 live births, though incidence rates vary depending on population iodine sufficiency, genetic background, and screening sensitivity. The majority of cases are caused by thyroid dysgenesis (agenesis, ectopy, hypoplasia), while others result from dyshormonogenesis or transient thyroid dysfunction.

Clinically, newborns with CH often present with subtle or nonspecific signs, including prolonged jaundice, hypotonia, feeding difficulties, macroglossia, and constipation. However, most infants appear asymptomatic at birth due to transplacental transfer of maternal thyroid hormones. This underscores the necessity of universal newborn screening programs.

Early biochemical screening—typically performed 24–72 hours after birth—allows for prompt diagnosis and immediate initiation of levothyroxine therapy. When treatment begins within the first two weeks of life, normal cognitive and physical development can be achieved in the majority of cases.

Literature Review: National Scientific Contribution. Scientific contributions to the understanding and management of congenital hypothyroidism (CH) in Uzbekistan have developed primarily through national endocrine research institutions, public health surveillance systems, and iodine deficiency control programs. Historically, Uzbekistan was recognized as a region affected by iodine deficiency disorders (IDD), which stimulated early national research on thyroid dysfunction, endemic goiter, and maternal–child iodine status. National academic centers and pediatric endocrinology departments have contributed clinical data on neonatal thyroid disorders, including diagnostic patterns, treatment protocols, and follow-up outcomes of children receiving levothyroxine therapy. Although much of this research has been disseminated in regional or national journals, it has informed local clinical guidelines and strengthened neonatal endocrine management practices.

A major institutional scientific milestone was the implementation and expansion of iodine deficiency prevention programs, particularly universal salt iodization. National monitoring initiatives evaluating household iodized salt coverage and urinary iodine concentration in vulnerable groups provided epidemiological evidence necessary for policy adjustments. These surveillance efforts contributed to measurable improvements in population iodine status and reduction of severe iodine deficiency outcomes.

In parallel, the gradual development of neonatal screening systems for thyroid dysfunction has marked an important step in translating research evidence into preventive practice. Laboratory-based TSH screening and early diagnostic confirmation have improved detection rates of congenital hypothyroidism and facilitated timely initiation of treatment, thereby reducing the risk of intellectual disability. Despite progress, gaps remain in peer-reviewed international publication output, longitudinal outcome research, and nationally consolidated incidence reporting for CH. Strengthening research infrastructure, enhancing international collaboration, and integrating endocrine surveillance data into global scientific platforms would further elevate Uzbekistan’s contribution to pediatric thyroid research.

Foreign Scientific Contribution: The global scientific foundation of congenital hypothyroidism (CH) has been shaped by decades of endocrinological research, epidemiological modeling, and neonatal screening innovation. International studies have established CH as one of the most preventable causes of intellectual disability, with early thyroid hormone replacement shown to normalize neurodevelopmental outcomes when initiated within the first two weeks of life. Seminal contributions from European and North American pediatric endocrinology groups standardized neonatal screening protocols based on thyroid-stimulating hormone (TSH) and thyroxine (T4) measurement. Large-scale newborn screening programs

implemented in the 1970s and 1980s demonstrated a dramatic reduction in severe cognitive impairment associated with untreated CH. These findings provided the empirical basis for universal screening recommendations now adopted worldwide. Research conducted by international thyroid and iodine experts further clarified the relationship between iodine deficiency and neonatal thyroid dysfunction. Studies led by global health authorities identified maternal iodine insufficiency as a determinant of transient neonatal hypothyroidism and subclinical thyroid dysfunction. The development of universal salt iodization guidelines by the World Health Organization and UNICEF was grounded in this body of evidence, linking population iodine sufficiency to reductions in congenital thyroid-related morbidity.

Results analysis: The reviewed literature indicates that congenital hypothyroidism (CH) remains a preventable but structurally sensitive neonatal condition whose burden in Uzbekistan must be interpreted within the broader public health trajectory of iodine deficiency control and maternal–child health reforms. Globally, CH incidence ranges from approximately 1 in 2,000 to 1 in 4,000 live births; however, incidence is influenced by iodine sufficiency, screening coverage, and diagnostic sensitivity. In Uzbekistan, although nationally consolidated peer-reviewed incidence figures remain limited, available institutional data suggest regional variability in neonatal thyroid dysfunction, likely reflecting historical iodine deficiency patterns and differences in health system capacity.

From a public health perspective, the most significant determinant in the Uzbek context has been iodine nutrition. Uzbekistan historically experienced moderate iodine deficiency disorders (IDD), particularly in inland and mountainous regions. The introduction of universal salt iodization significantly improved household iodized salt coverage and reduced severe iodine deficiency outcomes. Nevertheless, the literature suggests that monitoring compliance and quality control remain uneven, and maternal iodine sufficiency during pregnancy is not consistently documented across regions. Because fetal thyroid development depends entirely on adequate maternal iodine intake, even mild gestational deficiency may contribute to elevated neonatal TSH levels and transient hypothyroidism. This indicates that congenital hypothyroidism prevention cannot be isolated from maternal nutrition surveillance systems.

Neonatal screening represents the second critical public health pillar. International evidence demonstrates that early TSH-based screening followed by treatment within the first 14 days of life virtually eliminates the risk of intellectual disability associated with CH. In Uzbekistan, screening infrastructure has expanded, and laboratory capacity for TSH testing is available in major centers. However, the literature highlights gaps in uniformly published screening coverage rates, timeliness of confirmatory testing, and long-term developmental follow-up. Public health effectiveness is therefore determined not only by the existence of screening but also by system performance indicators such as turnaround time, referral pathways, and treatment initiation intervals.

Unveiling the Impact of Early Detection in Congenital Hypothyroidism

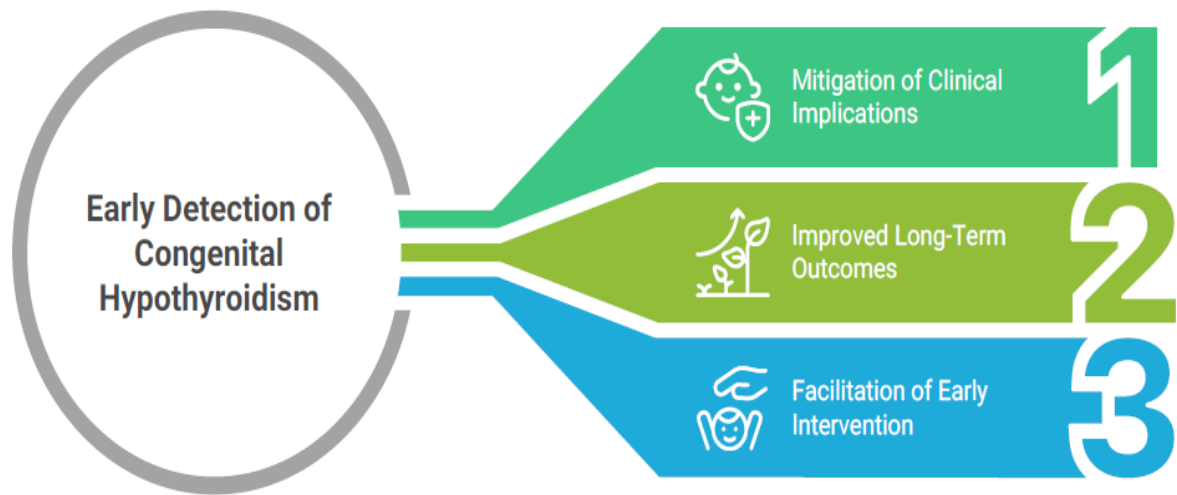


Figure 2. Clinical and Long-Term Outcomes Associated with Early Detection of Congenital Hypothyroidism

Early detection of congenital hypothyroidism through universal newborn screening significantly reduces the risk of irreversible neurodevelopmental impairment. Timely identification—typically via TSH and/or T4 measurement within the first days of life—enables rapid initiation of levothyroxine therapy. Early treatment mitigates immediate clinical complications such as hypertonia, feeding difficulties, and metabolic instability. More importantly, it ensures normal brain maturation, cognitive development, and physical growth. From a public health perspective, early detection strengthens preventive health systems by facilitating structured follow-up, reducing long-term disability burden, and lowering socioeconomic costs associated with intellectual impairment. Therefore, neonatal screening is considered a cornerstone intervention in congenital hypothyroidism control programs.

Treatment access itself is not reported as the primary constraint; levothyroxine therapy is clinically established and effective when initiated early. The greater public health vulnerability lies in delays between birth, screening, confirmation, and therapy, particularly in rural or resource-limited regions. This suggests that health system logistics, rather than pharmaceutical availability, may represent the more critical intervention target.

An additional structural gap identified in the literature is the limited publication of nationally consolidated endocrine surveillance data in international peer-reviewed journals. While institutional monitoring likely exists, insufficient dissemination reduces global visibility and limits comparative epidemiological analysis. This constrains evidence-based policy refinement and international benchmarking.

Overall, the public health trajectory of congenital hypothyroidism in Uzbekistan reflects substantial progress in iodine deficiency control and neonatal screening expansion, yet persistent system-level vulnerabilities remain. The burden of CH appears less a reflection of untreated endemic iodine deficiency and more a function of screening performance, maternal micronutrient surveillance, and regional equity in health service delivery. Strengthening iodine monitoring among pregnant women, standardizing national screening performance metrics, and implementing longitudinal neurodevelopmental follow-up would further consolidate gains and minimize preventable thyroid-related disability.

Conclusion: The public health evolution of congenital hypothyroidism (CH) in Uzbekistan reflects a broader transition from historically iodine-deficient conditions toward a more structured, prevention-

oriented maternal and neonatal health system. Significant progress has been achieved through universal salt iodization and the gradual expansion of neonatal screening services. These measures have reduced the risk of severe iodine deficiency disorders and improved early detection capacity for neonatal thyroid dysfunction.

However, the literature indicates that prevention of CH in Uzbekistan cannot rely solely on iodized salt policy or laboratory-based screening. Persistent variability in maternal iodine adequacy, regional inequalities in health service access, and inconsistencies in screening follow-up mechanisms represent ongoing structural vulnerabilities. The effectiveness of CH control is therefore dependent not only on the existence of national programs, but on their operational performance—particularly early diagnosis, rapid treatment initiation, and sustained monitoring.

From a public health perspective, congenital hypothyroidism in Uzbekistan is no longer primarily a reflection of endemic iodine deficiency, but rather a test of health system integration and maternal–child surveillance quality. Future progress will require strengthened maternal iodine monitoring, standardized national screening performance indicators, improved data transparency, and enhanced longitudinal developmental follow-up. Addressing these system-level dimensions will be essential to fully eliminate preventable thyroid-related neurodevelopmental impairment and consolidate gains in neonatal health outcomes.

Reference

1. Bhutta, Z. A., Das, J. K., Rizvi, A., et al. (2013). Evidence-based interventions for maternal and child nutrition. *The Lancet*, 382(9890), 452–477. [https://doi.org/10.1016/S0140-6736\(13\)60996-4](https://doi.org/10.1016/S0140-6736(13)60996-4)
2. Delange, F. (2001). Iodine deficiency as a cause of brain damage. *Postgraduate Medical Journal*, 77(906), 217–220. <https://doi.org/10.1136/pmj.77.906.217>
3. Léger, J., Olivieri, A., Donaldson, M., et al. (2014). European Society for Paediatric Endocrinology guidelines for congenital hypothyroidism. *Hormone Research in Paediatrics*, 81(2), 80–103. <https://doi.org/10.1159/000356507>
4. Rastogi, M. V., & LaFranchi, S. H. (2010). Congenital hypothyroidism. *Orphanet Journal of Rare Diseases*, 5(17), 1–22. <https://doi.org/10.1186/1750-1172-5-17>
5. Rovet, J. F. (2014). The role of thyroid hormones for brain development and cognitive function. *Endocrine Development*, 26, 26–43. <https://doi.org/10.1159/000363153>
6. UNICEF. (2019). Situation analysis of children and women in Uzbekistan. UNICEF Uzbekistan.
7. UNICEF Uzbekistan, Ministry of Health of the Republic of Uzbekistan, & partners. (2017). *Uzbekistan Nutrition Survey (UNS) 2017: Executive summary*. UNICEF.
8. World Health Organization. (2006). *WHO child growth standards*. WHO Press.
9. World Health Organization. (2007). *Assessment of iodine deficiency disorders and monitoring their elimination* (3rd ed.). WHO.
10. World Health Organization. (2014). *Guideline: Fortification of food-grade salt with iodine for the prevention and control of iodine deficiency disorders*. WHO.
11. World Health Organization. (2021). *Global anaemia estimates, 2021 edition*. WHO.
12. Rabiev, B. B. (2026). Children’s health in migrant families: Gender and care dynamics. *Central Asian Journal of Medical and Natural Science*, 7(2), 184–189. <https://doi.org/10.51699/cajmns.v7i2.3136>
13. World Health Organization & UNICEF. (2007). *Prevention and control of iodine deficiency in pregnant*

and lactating women and in children less than 2 years old. WHO.

14. Zimmermann, M. B. (2009). Iodine deficiency. *Endocrine Reviews*, 30(4), 376–408. <https://doi.org/10.1210/er.2009-0011>
15. Zimmermann, M. B., & Andersson, M. (2012). Update on iodine status worldwide. *Current Opinion in Endocrinology, Diabetes and Obesity*, 19(5), 382–387. <https://doi.org/10.1097/MED.0b013e328357271a>