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MODERN METHODS OF DIAGNOSTICS OF THYROID DISEASES IN CHILDREN

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Abstract

Thyroid diseases in children are becoming increasingly recognized as a significant health concern, with diagnoses of conditions such as hypothyroidism, hyperthyroidism, and thyroid cancer on the rise. Early diagnosis is essential to mitigate the impact of these diseases on growth, development, and overall well-being. This article aims to provide an overview of modern diagnostic methods for thyroid diseases in pediatric populations. These include clinical evaluation, biochemical tests, imaging techniques, and molecular diagnostic tools, all of which contribute to a more accurate and timely diagnosis. This review also highlights advancements in genetic screening, ultrasonography, and other innovative approaches to enhancing diagnostic accuracy.

Keywords: Thyroid diseases, pediatrics, diagnostics, hypothyroidism, hyperthyroidism, thyroid cancer, imaging, genetic screening.

Introduction

The prevalence of thyroid diseases in children is generally lower than in adults, but these conditions are still a significant concern due to their impact on growth,

development, and overall health. The main thyroid diseases in children include congenital hypothyroidism, autoimmune thyroid disorders like Hashimoto's thyroiditis and Graves' disease, and thyroid cancer. The prevalence of these conditions varies across regions and is influenced by genetic, environmental, and nutritional factors [2,5].

Thyroid diseases in children can have profound effects on growth, cognitive development, and overall health. Early detection and accurate diagnosis are critical to prevent long-term complications such as developmental delays, growth disturbances, and cardiovascular issues. The spectrum of thyroid conditions in children includes congenital hypothyroidism, autoimmune thyroiditis, hyperthyroidism (commonly Graves' disease), and thyroid malignancies. This article explores the current methodologies employed in diagnosing thyroid diseases in children, including clinical assessment, biochemical tests, imaging techniques, and emerging molecular tools.

The diagnostic process often begins with a detailed clinical history and physical examination. Physicians should carefully assess for symptoms such as growth failure, delayed development, fatigue, weight changes, and alterations in heart rate, which are commonly associated with thyroid dysfunction. The clinical presentation may vary depending on the specific thyroid disorder. For example, children with hypothyroidism may present with lethargy, constipation, and cold intolerance, whereas those with hyperthyroidism might show symptoms like irritability, weight loss, and tachycardia.

Physical signs such as goiter (an enlarged thyroid gland), skin changes, and delayed or accelerated puberty can also provide important clues. Although clinical evaluation is essential, it often requires confirmation through biochemical and imaging studies.

It is impossible to diagnose thyroid disease without laboratory tests. The first-line diagnostic approach for thyroid diseases is through biochemical testing. The primary hormones measured are: thyroid stimulating hormone, free thyroxine FT4, free triiodothyronine FT3 and thyroid antibodies [1,4].

Thyroid stimulating hormone TSH. Elevated TSH levels are indicative of hypothyroidism, whereas suppressed TSH levels suggest hyperthyroidism.

Free thyroxine FT4 and free triiodothyronine FT3. These hormones are measured alongside TSH to assess thyroid function. A low FT4 and high TSH suggest primary hypothyroidism, while elevated FT3 and FT4 levels with low or undetectable TSH indicate hyperthyroidism.

Thyroid antibodies. In children suspected of having autoimmune thyroiditis, measurement of anti-thyroid peroxidase anti-TPO and anti-thyroglobulin anti-Tg antibodies can help in diagnosing autoimmune thyroid diseases such as Hashimoto's thyroiditis and Graves' disease [2,3].

In pediatric cases, it is important to establish reference ranges for these tests as they can vary with age. For example, neonates and infants may have slightly different normal values compared to older children.

Instrumental research methods are of great importance for the diagnosis of thyroid diseases. Imaging plays an essential role in the evaluation of thyroid disease, especially when a structural abnormality is suspected. Modern imaging techniques used in pediatric thyroid diagnosis include:

Ultrasonography US. Ultrasonography is a non-invasive, widely used, and highly sensitive technique for evaluating the size, shape, and texture of the thyroid gland. It is particularly useful for detecting thyroid nodules, goiters, and cysts. High-resolution ultrasound can also help in assessing the vascularity of the thyroid gland, which may be abnormal in conditions like hyperthyroidism.

Radioactive iodine scintigraphy RAI. This imaging technique is primarily used in the diagnosis of hyperthyroidism. It helps assess the functional activity of the thyroid gland by visualizing iodine uptake. RAI is often utilized in children with suspected Graves' disease or toxic thyroid adenoma. However, it is generally avoided in young children due to radiation exposure concerns [5].

CT and MRI scanning. While not commonly used as first-line diagnostic tools, computed tomography CT and magnetic resonance imaging MRI may be employed in complex cases, such as the evaluation of large goiters or suspected malignancies. Currently, new methods of diagnosing thyroid pathology are being introduced in medicine, such as genetic screening and molecular diagnostics.

Recent advancements in molecular diagnostics have improved the understanding of genetic factors contributing to thyroid diseases. Genetic screening is particularly

useful in the diagnosis of congenital hypothyroidism, where mutations in thyroid hormone synthesis genes, such as TSHR, TPO, or DUOX2 can be identified. Genetic testing is also increasingly being applied to familial cases of autoimmune thyroid diseases to identify predispositions to thyroid conditions.

Molecular techniques like next-generation sequencing NGS can provide a comprehensive analysis of genetic mutations associated with thyroid diseases. Additionally, the use of circulating thyroid cancer biomarkers such as circulating tumor DNA ctDNA and specific microRNAs has shown promise in early cancer detection, though this is still an emerging field [1,4].

Conclusion

The diagnosis of thyroid diseases in children has advanced significantly with the development of modern diagnostic tools. While clinical evaluation remains a crucial first step, biochemical testing and imaging have become integral to confirming thyroid dysfunction and identifying structural abnormalities. Emerging technologies, such as genetic screening, molecular diagnostics, and AI-enhanced imaging, hold promise for further improving the accuracy and efficiency of pediatric thyroid disease diagnosis. Early and accurate diagnosis is vital in managing thyroid disorders effectively, ensuring that children receive the necessary interventions to support optimal growth and development.

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