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Original Research Article

Evaluation of Thyroid Disorders in Children with Emphasis on Congenital and Acquired Hypothyroidism

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Abstract:

Background: Thyroid disorders are among the most common endocrine problems in children and can significantly affect growth, metabolism, and neurodevelopment. Early identification is essential to prevent long-term complications, particularly in cases of congenital and acquired hypothyroidism.

Aim: To evaluate thyroid disorders in children with special emphasis on congenital and acquired hypothyroidism.

Methodology: A prospective observational study was conducted in the Department of Pediatrics, Shree Narayan Medical Institute & Hospital, Saharsa, Bihar. A total of 90 children aged 0–18 years with suspected thyroid dysfunction were included. Clinical evaluation and laboratory investigations, including serum TSH, Total T4, and Free T4 levels, were performed. Additional diagnostic tests such as ultrasonography and antibody assays were used when required.

Results: The majority of participants were aged 6–10 years (24.4%), with a slight female predominance (53.3%). Acquired hypothyroidism was the most common disorder (55.6%), followed by congenital hypothyroidism (26.7%) and subclinical hypothyroidism (11.1%). Growth retardation (42.2%), lethargy (35.6%), and constipation (31.1%) were the most common clinical manifestations. Mean TSH levels were elevated (14.8 ± 6.3 mIU/L) with reduced Total T4 and Free T4 levels.

Conclusion: Acquired hypothyroidism predominates among pediatric thyroid disorders. Early diagnosis through clinical assessment and thyroid function testing is essential for timely management and prevention of developmental complications.

Keywords: Thyroid disorders, Hypothyroidism, Congenital hypothyroidism, Acquired hypothyroidism, Children, Thyroid function test.

Introduction

Thyroid disorders represent one of the most widespread endocrine diseases that affect people from all age groups throughout the world [1]. The thyroid gland controls human growth and body metabolism and human development through its production of thyroid hormones which include thyroxine (T4) and triiodothyronine (T3) as its primary hormones. The hormones play a vital role in supporting normal physical development and brain growth and metabolic processes throughout the first two years of life and childhood. The thyroid gland will produce hazardous health effects during these critical growth times because its malfunctioning results in growth issues and puberty delays and cognitive development problems. The increasing rates of thyroid disorders which present lasting health problems when people do not receive diagnosis or treatment have turned thyroid diseases into a serious public health issue during the past few decades.

Thyroid diseases which rank as one of the most prevalent endocrine disorders throughout the world have become a significant health issue in India. According to projections from various epidemiological studies on thyroid disorders, it has been estimated that approximately 42 million people in India suffer from thyroid diseases [2]. The scale of the problem requires immediate solutions through both early detection and proper treatment methods. The main approach to treating thyroid disorders in children requires both early diagnosis and specific medical interventions because treatment delays can result in permanent damage to their physical and mental development.

Thyroid disorders in children show multiple manifestations which include hypothyroidism and hyperthyroidism together with thyroiditis and thyroid gland structural abnormalities. Among these, hypothyroidism is the most frequently encountered condition [3]. The body experiences hypothyroidism

when the thyroid gland produces inadequate thyroid hormones needed for basic metabolic functions. The body experiences two effects from this deficiency because it causes metabolic processes to slow down and it damages multiple organ systems. Pediatric patients with hypothyroidism present a major threat because thyroid hormones play an essential role in brain development and bone growth and overall body development. Therefore, the evaluation and early detection of thyroid dysfunction in children are essential components of pediatric healthcare.

Hypothyroidism in children can broadly be classified into two major categories: congenital hypothyroidism and acquired hypothyroidism [4]. Congenital hypothyroidism (CH) is a condition present at birth and is considered one of the most common endocrine disorders in the neonatal period. The condition of congenital hypothyroidism leads to severe intellectual disability and growth retardation and various developmental deficiencies when it goes undetected for an extended period. The implementation of neonatal screening programs across multiple countries enables identification of congenital hypothyroidism at an early stage, which leads to better outcomes because healthcare providers initiate thyroid hormone replacement therapy without delay.

Various etiological factors lead to the development of congenital hypothyroidism. The majority of cases stem from thyroid gland dysgenesis, which encompasses three conditions: agenesis and hypoplasia and ectopia of the thyroid gland [5]. Other causes include defects in thyroid hormone synthesis which medical professionals commonly refer to as dyshormonogenesis. The majority of patients with congenital hypothyroidism need to undergo permanent thyroid hormone replacement therapy throughout their entire lives. The condition of congenital hypothyroidism exists as a temporary form because it has the potential to disappear without treatment as time passes. Some patients with transient congenital hypothyroidism recover during follow-up without requiring continuous replacement therapy whereas others may need temporary thyroxine therapy until the condition resolves [6]. The differentiation between permanent and transient forms of congenital hypothyroidism is therefore important for determining long-term treatment strategies.

The onset of acquired hypothyroidism occurs during the later stages of childhood or adolescence while its development typically occurs due to autoimmune or inflammatory disorders that impact the thyroid gland [7]. Autoimmune thyroiditis which includes Hashimoto thyroiditis serves as the primary reason for acquired hypothyroidism to occur. Hashimoto thyroiditis leads to autoimmune destruction of thyroid tissue which results in decreasing thyroid hormone production over time. This condition represents the most prevalent type of thyroiditis that affects children and serves as the primary reason for thyroid dysfunction cases which occur in children who live in iodine-sufficient regions throughout the globe. The condition may show symptoms including fatigue weight gain cold intolerance constipation and poor growth velocity together with delayed puberty and goiter. The disease remains asymptomatic for extended periods in most children which creates a need for doctors to use laboratory tests as the primary method to confirm the diagnosis.

The clinical presentation of hypothyroidism in children varies depending on the age at onset and severity of hormone deficiency [8]. In neonates with congenital hypothyroidism, early symptoms may be subtle or absent, which is why neonatal screening programs are essential for early detection. Untreated infants may develop symptoms which include prolonged jaundice together with hypotonia and feeding difficulties and macroglossia and developmental delay. The symptoms of acquired hypothyroidism in older children include poor growth and delayed skeletal maturation and decreased academic performance and lethargy and weight gain. The nonspecific nature of these attributes requires biochemical tests to establish an accurate diagnosis.

The development of diagnostic technologies has brought about substantial progress in assessing thyroid disorders. The primary method for diagnosing hypothyroidism relies on laboratory tests that include thyroid profile tests which measure serum thyroid stimulating hormone (TSH) and free thyroxine (FT4) and triiodothyronine (T3) levels [9]. The combination of biochemical tests with imaging techniques such as ultrasonography and radionuclide scintigraphy enable medical professionals to assess both the structural and functional aspects of the thyroid gland. The diagnostic methods assist in identifying the specific type of hypothyroidism which enables doctors to select suitable treatment methods.

Thyroid gland ultrasonography functions as a non-invasive imaging method which medical professionals use to determine the thyroid gland's dimensions and configuration and internal structure. The test detects all types of structural defects which include hypoplasia and ectopia and nodules and inflammatory changes. Thyroid scintigraphy helps assess thyroid tissue function while it serves as an effective method to detect thyroid dysgenesis in cases of congenital hypothyroidism. The combination of diagnostic tools and laboratory tests enables medical professionals to determine the cause of hypothyroidism while developing treatment plans for extended patient care.

Early identification and appropriate treatment of hypothyroidism are essential to prevent complications. Levothyroxine replacement therapy remains the standard treatment for both congenital and acquired hypothyroidism. When initiated early, particularly in neonates with congenital hypothyroidism, this

therapy can prevent intellectual disability and ensure normal growth and development. Regular follow-up and monitoring of thyroid hormone levels are necessary to adjust the dosage and ensure optimal therapeutic outcomes.

Thyroid disorders in children remain difficult to treat because medical advances in diagnosis and treatment methods have not solved this problem in developing countries that lack proper awareness and screening programs and healthcare facilities. The majority of cases maintain their hidden status until patients start showing signs of the condition which leads to permanent health problems that already exist. The medical field requires complete assessment of thyroid disorders in children because it will help doctors detect and treat these conditions better.

In this context, the present study aims to evaluate thyroid disorders in children with special emphasis on congenital and acquired hypothyroidism. Understanding the clinical profile, diagnostic findings, and underlying causes of these conditions will help improve early detection, guide appropriate management strategies, and ultimately enhance health outcomes in the pediatric population.

Methodology

Study Design: The present study was conducted as a prospective observational study aimed at evaluating thyroid disorders in children with particular emphasis on congenital and acquired hypothyroidism.

Study Area: The study was carried out in the Department of Pediatrics, Shree Narayan Medical Institute & Hospital, Saharsa, Bihar, India from January 2024 to December 2024

Study Participants

Inclusion Criteria

- Children from birth to 18 years of age attending the pediatric outpatient department (OPD) or admitted to the inpatient department (IPD).
- Children with clinical suspicion of thyroid disorders, particularly hypothyroidism.
- Children showing abnormal thyroid function test results suggestive of congenital or acquired hypothyroidism.
- Children whose parents or guardians provided informed consent for participation in the study.

Exclusion Criteria

- Children already diagnosed with hypothyroidism and receiving treatment before the study period.
- Children with incomplete clinical records or laboratory investigations.
- Children whose parents or guardians refused to provide consent for participation in the study.

Sample Size: A total of 90 children fulfilling the inclusion criteria were included in the study. The sample comprised children presenting with suspected thyroid dysfunction who were evaluated clinically and biochemically for the presence of congenital or acquired hypothyroidism.

Procedure: All eligible children attending the pediatric outpatient department or admitted to the inpatient department during the study period were screened for symptoms and signs suggestive of thyroid disorders. After obtaining informed consent from parents or guardians, the relevant clinical information of each child was recorded using a predesigned proforma. Detailed demographic data, medical history, and clinical presentation were documented carefully.

Each participant underwent a thorough clinical examination, focusing on signs commonly associated with hypothyroidism. In neonates and infants, symptoms such as coarse facial features, feeding difficulties, macroglossia, prolonged neonatal jaundice, constipation, lethargy, dry skin, and delayed developmental milestones were noted. In older children and adolescents, clinical features such as short stature, growth retardation, delayed dentition, obesity, fatigue, poor scholastic performance, delayed puberty, irregular menstrual cycles in adolescent girls, and mental retardation were evaluated.

Following the clinical assessment, all participants were subjected to thyroid function tests to determine the hormonal status. The laboratory investigations included measurement of Thyroid Stimulating Hormone (TSH), Total Thyroxine (T4), and Free Thyroxine (FT4) levels. Children with elevated TSH levels accompanied by decreased T4 or FT4 levels were considered suggestive of hypothyroidism and were included in further diagnostic evaluation.

Additional diagnostic procedures were carried out where necessary to determine the etiology of thyroid dysfunction. Ultrasonography of the neck was performed to assess the size, position, and structural abnormalities of the thyroid gland. Antibody assays, including anti-thyroid peroxidase antibodies (anti-TPO) and anti-thyroid microsomal antibodies, were conducted to detect autoimmune thyroid disease.

In selected cases, thyroid radionuclide imaging was performed using Technetium-99m thyroid scan to evaluate the functional activity, anatomical location, and structural characteristics of the thyroid gland. This imaging technique helped identify conditions such as thyroid dysgenesis or ectopic thyroid tissue. Radioactive iodine uptake (RAI-U) studies were also used in certain cases to evaluate iodine uptake capacity and detect possible defects in hormone synthesis. In suspected cases of dyshormonogenesis, additional diagnostic tests such as the perchlorate discharge test were conducted to assess defects in iodine organification.

Based on clinical findings, biochemical results, and imaging studies, children were categorized into congenital hypothyroidism or acquired hypothyroidism for further analysis and interpretation.

Statistical Analysis: All collected data were compiled and entered into a Microsoft Excel spreadsheet and subsequently analyzed using Statistical Package for the Social Sciences (SPSS) version 27.0. Descriptive statistics such as frequency, percentage, mean, and standard deviation were used to summarize demographic variables and clinical characteristics. Appropriate statistical tests were applied to determine associations between variables where necessary, and the results were presented in tables and figures for better interpretation.

Result

Table 1 shows the age distribution of the study participants. Out of the total 90 children included in the study, the highest proportion belonged to the 6–10 years age group with 22 children (24.4%). This was followed by the 11–14 years age group, which comprised 19 children (21.1%). The 0–1 year age group included 18 children (20%), while 16 children (17.8%) were in the 2–5 years age group. The lowest proportion of participants was observed in the 15–18 years age group with 15 children (16.7%). Overall, the distribution indicates that children from different age groups were fairly represented in the study, with a slightly higher concentration in the 6–10 years age category.

Table 1: Age Distribution of Study Participants (n = 90)

Age Group (Years)	Number of Children	Percentage (%)
0–1	18	20
2–5	16	17.8
6–10	22	24.4
11–14	19	21.1
15–18	15	16.7
Total	**90**	**100**

Table 2 shows the gender distribution of the study participants. Out of the total 90 children included in the study, 48 were females and 42 were males. Females constituted the slightly higher proportion of the study population, accounting for 53.3%, while males represented 46.7%. This indicates that the participation of female children was marginally greater than that of male children in the present study. Overall, the distribution demonstrates a relatively balanced representation of both genders among the study participants.

Table 2: Gender Distribution of Study Participants (n = 90)

Gender	Number of Children	Percentage (%)
Male	42	46.7
Female	48	53.3
Total	**90**	**100**

Table 3 shows the distribution of different types of thyroid disorders among the 90 children included in the study. The findings indicate that acquired hypothyroidism was the most common disorder, accounting for 50 cases (55.6%) of the total study population. Congenital hypothyroidism was observed in 24 children (26.7%), representing the second most frequent thyroid disorder. Subclinical hypothyroidism was identified in 10 cases (11.1%), suggesting a moderate proportion of children with mild or early thyroid dysfunction. In addition, other thyroid disorders were reported in 6 children (6.6%), constituting the smallest category in the study. Overall, the results highlight that acquired hypothyroidism predominated among the study participants, while congenital and subclinical forms were also present in notable proportions.

Table 3: Distribution of Types of Thyroid Disorders (n = 90)

Thyroid Disorder	Number of Cases	Percentage (%)
Congenital Hypothyroidism	24	26.7
Acquired Hypothyroidism	50	55.6
Subclinical Hypothyroidism	10	11.1
Other Thyroid Disorders	6	6.6
Total	**90**	**100**

Table 4 shows the clinical presentation of children with hypothyroidism among the 90 study participants. The most common clinical feature observed was growth retardation, reported in 38 cases

(42.2%), indicating that impaired physical growth is a major manifestation of hypothyroidism in children. This was followed by lethargy or fatigue, which was present in 32 children (35.6%), and constipation, seen in 28 cases (31.1%). Other notable symptoms included dry skin in 24 children (26.7%) and poor scholastic performance in 19 cases (21.1%), suggesting the impact of hypothyroidism on both physical and cognitive functioning. Delayed puberty was reported in 15 children (16.7%), while macroglossia was observed in 12 cases (13.3%). The least common presentation was prolonged neonatal jaundice, documented in 10 children (11.1%). Overall, the findings highlight those growth-related problems and generalized symptom such as fatigue and constipation were the most frequent clinical manifestations among children diagnosed with hypothyroidism.

Table 4: Clinical Presentation of Children with Hypothyroidism (n = 90)

Clinical Feature	Number of Cases	Percentage (%)
Growth retardation	38	42.2
Lethargy / Fatigue	32	35.6
Constipation	28	31.1
Dry skin	24	26.7
Delayed puberty	15	16.7
Macroglossia	12	13.3
Poor scholastic performance	19	21.1
Prolonged neonatal jaundice	10	11.1

Table 5 shows the thyroid function test parameters among the 90 study participants. The mean serum TSH level was 14.8 ± 6.3 mIU/L, which is markedly elevated, indicating significant thyroid dysfunction among the studied children. In contrast, the mean Total T4 level was 4.9 ± 1.6 µg/dL, which is comparatively lower than the normal reference range, suggesting reduced thyroid hormone production. Similarly, the mean Free T4 level was 0.82 ± 0.24 ng/dL, which is also on the lower side. The combination of elevated TSH with reduced Total T4 and Free T4 levels indicates the presence of hypothyroid status among the participants, supporting the occurrence of thyroid disorders, particularly hypothyroidism, in the study population. These findings highlight the importance of thyroid function testing for early detection and management of thyroid abnormalities in children.

Table 5: Thyroid Function Test Parameters Among Study Participants (n = 90)

Parameter	Mean ± SD
TSH (mIU/L)	14.8 ± 6.3
Total T4 (µg/dL)	4.9 ± 1.6
Free T4 (ng/dL)	0.82 0.24

Discussion

The present study evaluated the pattern of thyroid disorders among children with special emphasis on congenital and acquired hypothyroidism. The study found essential demographic information and clinical data and biochemical details which demonstrated how thyroid dysfunction affected the pediatric population. The study results show similar findings to earlier published research studies although some differences were observed.

The study found that 24.4% of cases occurred in the 6–10 years age group while 21.1% of cases occurred in the 11–14 years age group and 20% of cases occurred in infants aged 0–1 year. Similar findings were reported by Seth et al. (2011) [10], who observed that most children with hypothyroidism were diagnosed during school age due to the appearance of symptoms such as growth retardation and poor academic performance. Their study reported that approximately 27% of cases occurred in children aged 6–10 years, which is comparable to the 24.4% observed in the present study. Venkateswarlu et al. (2015) [11] reported that school-age children represent 25 to 30 percent of pediatric thyroid disorder cases because clinical symptoms become more visible during this age period. The present study found that 20% of participants were infants which exceeds the 12 to 15 percent range of previous studies that focused on older children. The difference exists because of better public knowledge and the implementation of early detection methods.

The current research found that more females than males participated in the study because females made up 53.3% of the cases while males constituted 46.7% of the cases. De Luca et al. (2013) [12] found that approximately 55 to 60 percent of pediatric hypothyroidism cases occurred in female patients which supports this finding. Unnikrishnan and Menon (2011) [13] discovered that thyroid disorders affected women more than men especially in cases of autoimmune thyroiditis. The study showed that both sexes experienced thyroid disorders during

childhood since males and females both experienced the condition but females experienced it more often.

The distribution of thyroid disorders in the present study showed that acquired hypothyroidism was the most common type, which accounted for 55.6% of cases while congenital hypothyroidism and subclinical hypothyroidism followed with respective rates of 26.7% and 11.1%. The results of this study match the findings of Venkateswarlu et al. (2015) who demonstrated that acquired hypothyroidism made up approximately 58% of pediatric thyroid disorder cases while congenital hypothyroidism represented about 24%. The study conducted by Seth et al. (2011) discovered that congenital hypothyroidism occurred at a higher rate than acquired forms because their research examined numerous children who received late congenital disease diagnoses. The present study discovered that acquired hypothroidism occurred more frequently because it showed an increasing trend which resulted from rising cases of autoimmune thyroiditis and other acquired conditions in kids with advanced developmental stages. De Luca et al. (2013) confirmed that Hashimoto's thyroiditis serves as the primary reason for acquired hypothyroidism among children and adolescents since it produces approximately 50-60% of all cases.

The current study demonstrated clinical symptoms which matched the findings of earlier research. The most frequently observed clinical symptom among children was growth retardation which affected 42.2% of the studied population. Seth et al. (2011) found that almost 40% of children with hypothyroidism showed either short stature or growth delay. The thyroid hormone deficiency leads to skeletal development and growth disruption which results in this symptom appearing frequently. The present study showed that 35.6% of cases experienced lethargy or fatigue which matches the 32-38% rate found in other pediatric research studies. Grüters and Krude (2012) [14] reported that 31.1% of children experienced constipation which matched their findings that gastrointestinal symptoms occurred in approximately 30% of pediatric hypothyroid cases.

The study found other symptoms which included dry skin (26.7%), poor scholastic performance (21.1%), and delayed puberty (16.7%). De Luca et al. (2013) found similar results because 20% of hypothyroid children showed cognitive or academic difficulties. The present study found 13.3% of cases exhibited macroglossia which doctors consider a traditional symptom of congenital hypothyroidism. Tamam et al. (2009) [15] found macroglossia in 15% of children with congenital hypothyroidism which matches the current research results. The study found that 11.1% of cases developed prolonged neonatal jaundice which matches previous research findings that showed neonatal jaundice occurred in about 10-12% of congenital hypothyroidism cases.

The present study's biochemical results established an additional confirmation of hypothyroidism diagnosis. The mean serum TSH level showed a substantial increase to 14.8 ± 6.3 mIU/L, whereas the mean total T4 level measured 4.9 ± 1.6 µg/dL, and free T4 level showed 0.82 ± 0.24 ng/dL. The results of this study match the results of Tamam et al. (2009), who found that children with hypothyroidism had mean TSH levels exceeding 12 mIU/L and decreased T4 levels. Monroy-Santoyo et al. (2011) [16] found that children with congenital hypothyroidism showed increased TSH levels which reached an average of 13 to 15 mIU/L, while their free T4 levels showed significant decreases. The biochemical tests demonstrate primary hypothyroidism because thyroid hormone production has decreased, which causes the pituitary gland to produce more TSH.

The study results confirm earlier research which examined different groups of people. The study results show that acquired hypothyroidism predominates in the population and that school-age children show higher rates of the condition and that both growth retardation and fatigue represent common clinical symptoms which doctors observe. The study results show that congenital cases and specific clinical symptoms display minor differences which researchers attribute to variations in their study groups and age distribution and diagnostic methods. The study results demonstrate that doctors need to identify thyroid disorders in children at an early stage while children who show growth delays and developmental issues and other symptoms should undergo regular testing and biochemical testing.

Conclusion

The present study concluded that thyroid disorders are an important endocrine problem among children, with acquired hypothyroidism being the most common form followed by congenital hypothyroidism and subclinical hypothyroidism. The findings demonstrated that children of different age groups were affected, although a slightly higher proportion was observed in the 6–10 years age group, and a marginal female predominance was noted. Growth retardation, lethargy, constipation, and dry skin were the most frequently observed clinical manifestations, indicating the significant impact of thyroid hormone deficiency on physical growth and general health. The biochemical findings showed elevated TSH levels with reduced Total T4 and Free T4, confirming hypothyroid status among the participants. These results emphasize the importance of early clinical evaluation, routine thyroid function testing, and timely management to prevent long-term developmental and metabolic complications in children.

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