International Journal of Contemporary Pediatrics
Ranabijuli et al. Int J Contemp Pediatr. 2026 Dec;13(12):2563-2567
https://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: https://dx.doi.org/10.18203/2349-3291.ijcp20264326

Case Series

Diverse presentations of goitre in children: a case series

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Received: 03 September 2026
Revised: 05 October 2026
Accepted: 04 November 2026

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ABSTRACT

Goitre in the pediatric population represents a broad spectrum of thyroid disorders ranging from autoimmune thyroiditis to hyperthyroid states such as Graves' disease. In iodine-sufficient regions, autoimmune thyroiditis is the most common etiology. However, clinical presentation varies widely from euthyroid enlargement to overt hypothyroidism or thyrotoxicosis. Early recognition is critical to prevent growth disturbances, neurocognitive impairment, and metabolic complications. This is a retrospective descriptive case series of five pediatric patients presenting with goitre. Detailed clinical history, examination findings, thyroid function tests, antibody profiles, imaging findings, treatment strategies, and follow-up data were analysed. Among the five cases, four were diagnosed with autoimmune thyroiditis and one with Graves' disease. Presentations ranged from euthyroid goitre to severe hypothyroidism and overt hyperthyroidism. Anti-thyroid peroxidase antibodies were elevated in most cases of autoimmune thyroiditis. One case required prolonged dose titration over several years. Another case demonstrated spontaneous reduction in goitre without pharmacological intervention. Pediatric goitre demonstrates significant heterogeneity in clinical and biochemical presentation. Comprehensive evaluation including antibody profiling is essential. Long-term follow-up is mandatory due to risk of progression and fluctuations in thyroid status.

Keywords: Goitre, autoimmune thyroiditis, Graves' disease, Anti thyroid peroxidase antibody

INTRODUCTION

Goitre is defined as enlargement of the thyroid gland beyond the normal size for age and sex and is one of the most common endocrine abnormalities encountered in children and adolescents. It represents a clinical manifestation of diverse thyroid disorders rather than a disease entity itself. Thyroid enlargement may result from physiological, autoimmune, inflammatory, nutritional, congenital, or neoplastic conditions. The clinical presentation ranges from asymptomatic thyroid enlargement detected during routine examination to symptomatic disease associated with thyroid dysfunction or compressive symptoms.[1] Globally, iodine deficiency remains the most common cause of goitre; however, in iodine-sufficient regions, autoimmune thyroid diseases, particularly Hashimoto thyroiditis and Graves' disease, constitute the predominant etiologies in school-aged children and adolescents. Hashimoto thyroiditis is characterised by autoimmune destruction of thyroid tissue and typically presents with a painless diffuse goitre. Patients may initially be euthyroid but can subsequently develop subclinical or overt hypothyroidism. Conversely, Graves' disease is the leading cause of hyperthyroidism in children and is characterised by diffuse thyroid enlargement, thyrotoxicosis, and the presence of TSH receptor antibodies (TRAb).[2]

Less common causes of pediatric goitre include dyshormonogenesis, congenital defects in thyroid

hormone synthesis, thyroid cysts, multinodular goitre, subacute or suppurative thyroiditis, benign thyroid nodules, and thyroid malignancies. Although thyroid cancer is uncommon in childhood, papillary thyroid carcinoma is the most frequent histological subtype and should always be considered in children presenting with thyroid nodules or suspicious ultrasonographic features.[3]

The clinical manifestations of goitre depend on the underlying etiology and thyroid functional status. Children may present with diffuse or nodular thyroid enlargement, symptoms of hypothyroidism such as fatigue, constipation, poor growth, weight gain, delayed puberty, and impaired school performance, or symptoms of hyperthyroidism including weight loss, tachycardia, tremors, heat intolerance, excessive sweating, emotional lability, and behavioural disturbances. Large goitres may occasionally produce compressive symptoms such as dysphagia, dyspnea, or hoarseness of voice due to compression of adjacent cervical structures. Nevertheless, many children remain asymptomatic, with thyroid enlargement detected incidentally during routine physical examination.[4]

The evaluation of pediatric goitre requires a systematic approach comprising detailed clinical history, physical examination, thyroid function testing, thyroid autoantibody assessment, and imaging studies. Measurement of serum thyroid-stimulating hormone (TSH), free thyroxine (FT4), and triiodothyronine (T3), together with anti-thyroid peroxidase (anti-TPO) and anti-thyroglobulin antibodies, is essential for diagnosing autoimmune thyroiditis . Measurement of TSH receptor antibodies (TRAb) is particularly useful in confirming Graves' disease . Thyroid ultrasonography remains the imaging modality of choice for assessing thyroid size, echotexture, vascularity, and the presence of nodules. Radionuclide thyroid scanning and fine-needle aspiration cytology are reserved for selected cases. Because pediatric goitre encompasses a wide spectrum of disorders with variable clinical and biochemical presentations, early recognition and appropriate evaluation are crucial for timely diagnosis and management. Delayed diagnosis may adversely affect growth, pubertal development, cognitive function, and overall quality of life.

The present case series highlights the diverse clinical spectrum of pediatric goitre encountered at a tertiary care centre and emphasises the importance of comprehensive clinical evaluation, antibody profiling, and long-term follow-up in achieving optimal outcomes.

CASE SERIES

Case 1: grave's disease (hyperthyroid goitre)

A 10-year-old girl presented with a one-month history of anterior neck swelling associated with significant weight loss (approximately 8 kg), intermittent abdominal pain, heat intolerance, hot flushes, and behavioural changes, including increased fearfulness. There was no history of palpitations, increased appetite, tremors, or ophthalmic symptoms such as staring look or diplopia. There was no family history of thyroid disease. On examination, the child was conscious and hemodynamically stable. Local examination revealed a WHO Grade 2 diffuse goitre. No cervical lymphadenopathy or thyroid bruit was noted.

Laboratory investigations demonstrated markedly elevated serum T3 (7.08 ng/ml) and T4 (26 µg/dl) levels with suppressed TSH (0.009 µIU/ml), consistent with overt hyperthyroidism. TSH receptor antibody (TRAb) level was significantly elevated at 29.08 IU/l (reference <1.99 IU/l), confirming Graves' disease. Thyroid ultrasonography revealed diffuse enlargement of both lobes with heterogeneous echotexture and increased vascularity suggestive of thyroiditis. A 99mTc-pertechnetate thyroid scan demonstrated diffusely increased tracer uptake throughout the gland, consistent with Graves' disease.

The patient was initially started on propranolol for symptomatic control, followed by carbimazole. During follow-up, her symptoms gradually improved, thyroid swelling decreased, and thyroid function tests progressively normalised. She is currently receiving carbimazole (0.5 mg/kg/day) and propranolol (0.5 mg/kg/day) with regular endocrinology follow-up.

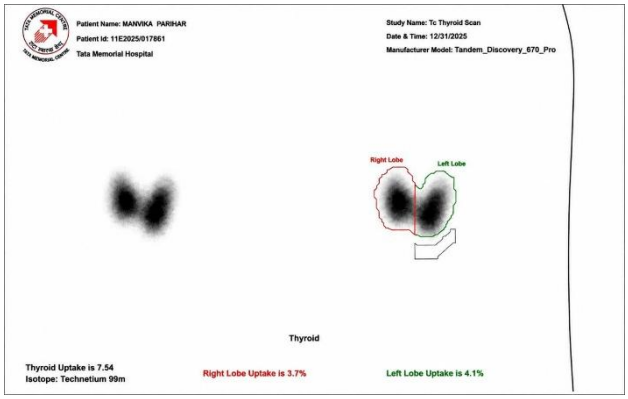


Figure 1: Tc-99m-pertechnetate thyroid scan s/o diffuse increased tracer uptake in the thyroid gland, consistent with grave's disease.

Case 2: severe autoimmune hypothyroidism

A 16-year-old girl presented with anterior neck swelling for two months, accompanied by leg cramps, poor concentration at school, easy fatigability, and irritability for one month. There was no family history of thyroid disease.

General physical and systemic examinations were unremarkable. Neck examination revealed a WHO Grade 2 diffuse goitre without tenderness or cervical lymphadenopathy. Thyroid function tests demonstrated

severe primary hypothyroidism with TSH >100 µIU/ml and low T4 (4.91 µg/dl). Anti-thyroid peroxidase (anti-TPO) antibody levels were markedly elevated (>1300 IU/ml; reference <34 IU/ml), confirming autoimmune thyroiditis.

The patient was started on levothyroxine 100 µg once daily. Serial monitoring demonstrated progressive normalization of thyroid function and gradual reduction in thyroid gland size. Dose adjustments were made according to serial TSH values. At the most recent follow-up, she remains clinically euthyroid on levothyroxine 75 µg once daily.

Case 3: early childhood autoimmune thyroiditis with long-term follow-up

A 3-year-old girl presented with progressive neck swelling of six months' duration. Her mother had a history of gestational hypothyroidism and was receiving levothyroxine therapy. General and systemic examinations were normal. Neck examination revealed diffuse thyroid enlargement. Initial laboratory evaluation showed severe hypothyroidism with TSH >100 µIU/ml, low T4 (1.40 µg/dl), anti-thyroglobulin antibody level of 501 IU/ml (reference 10–115 IU/ml), and anti-TPO antibody level of 14.1 IU/ml (reference 5–34 IU/ml).

The child was started on levothyroxine 75 µg once daily. During long-term follow-up, repeated dose adjustments were required because of fluctuations in thyroid function. Despite these variations, the patient remained compliant with therapy. At the latest follow-up, she was clinically euthyroid with normal thyroid function tests, highlighting the importance of long-term surveillance in autoimmune thyroiditis diagnosed during early childhood.

Case 4: subclinical autoimmune thyroiditis

A 7-year-old girl presented with painless neck swelling for four months without symptoms suggestive of thyroid dysfunction. Her mother had been diagnosed with hyperthyroidism ten years earlier and was receiving carbimazole therapy. General physical and systemic examinations were within normal limits. Neck examination demonstrated a WHO Grade 1B diffuse goitre.

Laboratory evaluation revealed mildly elevated TSH with normal serum T3 and T4 levels, consistent with subclinical hypothyroidism. Anti-TPO antibody level exceeded 1300 IU/ml (reference <60 IU/ml), while anti-thyroglobulin antibody level was 309.16 IU/ml (reference <4.5 IU/ml), confirming autoimmune thyroiditis. Thyroid ultrasonography demonstrated a few sub centimeter reactive cervical lymph nodes at level V without other significant abnormalities.

As thyroid function remained largely preserved, the patient was managed conservatively without levothyroxine therapy. Serial follow-up demonstrated stable thyroid function with gradual reduction in goitre size.

Case 5: euthyroid autoimmune thyroiditis

An 11-year-old girl presented with painless anterior neck swelling for four months without systemic symptoms. There was no family history of thyroid disease. Clinical examination revealed diffuse thyroid enlargement without cervical lymphadenopathy.

Thyroid function tests were within the normal range, although free T4 was mildly elevated (21.40 pmol/l; reference 11.10–18.10 pmol/l). Anti-TPO antibody level was greater than 1300 IU/ml (reference <60 IU/ml), while anti-thyroglobulin antibody level measured 348.29 IU/ml (reference <4.5 IU/ml). Thyroid ultrasonography was suggestive of autoimmune thyroiditis.

A diagnosis of euthyroid autoimmune thyroiditis was established. As thyroid function was preserved, no pharmacological treatment was initiated. Follow-up demonstrated a decline in antibody titers (anti-TPO 138 IU/ml and anti-thyroglobulin antibody 132 IU/ml). Serial follow-up demonstrated stable thyroid function with gradual reduction in goitre size.

Table 1: Comparative clinical features.

Parameter	Case 1	Case 2	Case 3	Case 4	Case 5
Age/Sex	10 yr/F	16 yr/ F	3yr/F	11yr/F	7yr/F
Goiter grade	Grade 2	Grade 2	Grade 1B	Grade IB	Grade 2
Thyroid status	Hyperthyroid	Hypothyroid	Hypothyroid	Subclinical Hypothyroidism	Euthyroid
Antibody profile	TRAb +	Anti-TPO +	Anti-Tg +	Anti-TPO + Anti Tg +	Anti-TPO + Anti Tg +
Family history	No	No	Maternal gestational hypothyroidism	Maternal Hyperthyroidism	No
Diagnosis	Grave's disease	autoimmune thyroiditis	autoimmune thyroiditis	autoimmune thyroiditis	autoimmune thyroiditis
Treatment	Carbimazole	thyroxine	Thyroxine	Observation	Observation
Outcome	Improving	Stable	Controlled	Stable	Stable

DISCUSSION

Goitre is one of the most common endocrine disorders encountered during childhood and adolescence. Although thyroid enlargement may result from several etiologies, autoimmune thyroid disease has become the predominant cause of pediatric goitre in iodine-sufficient regions. Our case series demonstrates the broad clinical and biochemical spectrum of autoimmune thyroid disorders in children, ranging from euthyroid autoimmune thyroiditis to overt hypothyroidism and Graves' hyperthyroidism, highlighting the importance of comprehensive clinical and laboratory evaluation.

Autoimmune thyroiditis accounted for four of the five cases in our series. This observation is consistent with previous studies, particularly the longitudinal cohort reported by Wasniewska et al who reported Hashimoto thyroiditis as the most common cause of acquired thyroid disease in school-aged children and adolescents.[5] Autoimmune thyroiditis exhibits considerable heterogeneity, with patients presenting as euthyroid, subclinically hypothyroid, or overtly hypothyroid at diagnosis.[6] The age at presentation in our patients ranged from 3 to 16 years, illustrating that autoimmune thyroid disease can occur throughout childhood. Although the incidence increases during adolescence, younger children may also be affected, as demonstrated by our third case. Early-onset disease requires prolonged surveillance because thyroid function may fluctuate during growth and development.[7]

One of the distinguishing features of this series is the diversity of thyroid functional status at presentation. Two children presented with overt hypothyroidism requiring levothyroxine replacement, one child had subclinical hypothyroidism that remained stable during follow-up, one child remained euthyroid despite markedly elevated thyroid autoantibodies, and one child presented with overt Graves' hyperthyroidism. These findings support previous reports demonstrating that autoimmune thyroid disease represents a dynamic process rather than a static disorder.[8]

Anti-thyroid peroxidase antibodies were positive in almost all patients with autoimmune thyroiditis, confirming their value as the most sensitive serological marker of autoimmune thyroid disease. Anti-thyroglobulin antibodies were also elevated in several patients, supporting the diagnosis. In contrast, markedly elevated TSH receptor antibodies (TRAb) confirmed the diagnosis of Graves' disease in the hyperthyroid patient. These observations are consistent with current European Thyroid Association and American Thyroid Association recommendations.[9]

Ultrasonography proved valuable in all patients by demonstrating diffuse thyroid enlargement and altered echotexture suggestive of autoimmune thyroiditis. In the child with Graves' disease, increased thyroid vascularity together with diffuse uptake on 99mTc-pertechnetate thyroid scintigraphy established the diagnosis. Imaging therefore serves as an important adjunct to biochemical and immunological evaluation.[10]

Management was individualised according to thyroid functional status. Patients with overt hypothyroidism received levothyroxine replacement, whereas the child with Graves' disease responded well to carbimazole with propranolol for symptomatic control. Children with euthyroid or subclinical autoimmune thyroiditis were managed conservatively with periodic clinical and biochemical follow-up. Current international guidelines recommend treatment decisions based primarily on thyroid function rather than antibody positivity alone.

Long-term follow-up remains an essential aspect of management. One patient in our series required repeated levothyroxine dose adjustments over several years because of fluctuating thyroid function, whereas another child demonstrated spontaneous reduction in thyroid size without pharmacological intervention. Such findings emphasize the unpredictable natural history of pediatric autoimmune thyroid disease and reinforce the need for continued surveillance even in clinically stable children. Although our study includes only five patients, it effectively illustrates the wide spectrum of pediatric goitre encountered in routine clinical practice. The principal limitation of this study is its retrospective design and relatively small sample size, which limits generalizability. Nevertheless, the detailed clinical characterization and long-term follow-up provide valuable insights into the diagnosis and management of pediatric thyroid disorders.

CONCLUSION

Goitre in children encompasses a broad spectrum of disorders with diverse clinical presentations and thyroid functional states. Autoimmune thyroiditis remains the predominant cause in iodine-sufficient regions, while Graves' disease should be considered in children presenting with features of thyrotoxicosis. Comprehensive evaluation, including thyroid function tests, thyroid autoantibody profiling, and ultrasonography, is essential for accurate diagnosis. Management should be individualized according to thyroid functional status, and long-term follow-up is necessary because thyroid function may evolve over time.

ACKNOWLEDGEMENTS

The authors would also like to thank medical director and hospital administration of Jagjivan Ram Hospital, Mumbai and the scientific and ethical committee of the hospital.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Ranabijuli PK, Lodi N, Kadam P, Deshmukh SN. Diverse presentations of goitre in children: a case series. Int J Contemp Pediatr 2026;13:2563-7.