International Journal of Contemporary Pediatrics
Patel BG. Int J Contemp Pediatr. 2025 May;12(5):852-854
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: https://dx.doi.org/10.18203/2349-3291.ijcp20251112

Case Report

Primary acquired hypothyroidism with associated myopathy in a 12-year-old female: the hidden face of hypothyroidism

Bhargav G. Patel*

Department of Pediatrics, Sadvichar Parivar Godhra, P. T. Mirani Hospital, Godhra, Gujarat, India

Received: 20 March 2025
Revised: 15 April 2025
Accepted: 19 April 2025

***Correspondence:**
Dr. Bhargav G. Patel,
E-mail: bhargavpatel381@gmail.com

ABSTRACT

Primary acquired hypothyroidism with autoimmune aetiology can present with atypical neuromuscular features in pediatric patients. We reported a case of a 12-year-old female presenting with calf muscle pain, pseudohypertrophy, decreased physical activity, and rapid weight gain. Laboratory investigations revealed markedly elevated thyroid stimulating hormone, low free T4, and significantly raised anti-thyroglobulin antibodies. Early diagnosis and prompt initiation of levothyroxine led to dramatic clinical and biochemical improvement. This case highlights the importance of considering thyroid dysfunction in the differential diagnosis of pediatric myopathies.

Keywords: Case report, Hypothyroidism, Autoimmune thyroiditis, Myopathy, Pediatric endocrinology

INTRODUCTION

Primary acquired hypothyroidism is one of the most common endocrine disorders in children and adolescents, often presenting with fatigue, constipation, weight gain, cold intolerance, and growth retardation.[1] In some pediatric patients, however, thyroid dysfunction may primarily manifest with neuromuscular symptoms such as calf muscle pseudohypertrophy and myopathy, leading to diagnostic confusion.[2,3] These features may resemble primary muscular disorders such as muscular dystrophies or metabolic myopathies, potentially delaying appropriate endocrine evaluation.

Autoimmune thyroiditis, most commonly Hashimoto's thyroiditis, is the leading cause of acquired hypothyroidism in this age group and is diagnosed through elevated levels of anti-thyroglobulin or anti-thyroid peroxidase (TPO) antibodies.[4,5] Cases have been documented where pediatric hypothyroidism initially presented with muscular features, including weakness or pseudohypertrophy, even in the absence of typical systemic hypothyroid symptoms.[6,7] In some instances, the muscular signs were so prominent that they mimicked neuromuscular diseases.[8]

In this report, we described a case of a 12-year-old female presenting with muscle pain, decreased activity, and pseudohypertrophy due to primary acquired hypothyroidism of autoimmune origin. This case emphasizes the importance of thyroid screening in children presenting with unexplained muscle symptoms or abnormal anthropometry.

CASE REPORT

A 12-year-old female presented with a one-month history of progressive calf muscle pain, decreased physical activity, and a notable lack of interest in her surroundings. Her parents also reported a sudden weight gain over the preceding 3 to 4 days, although her appetite and diet had not significantly changed.

On general physical examination, the child appeared short in stature. Anthropometric assessment revealed that both her bone age and chronological age were below the expected height age, consistent with familial short

stature. Systemic examination was unremarkable. Neurological assessment showed marked pseudohypertrophy of the calf muscles, giving the appearance of muscle enlargement without accompanying proximal muscle weakness. Reflexes and sensory examination were within normal limits.

Laboratory investigations revealed a significantly elevated thyroid-stimulating hormone (TSH) level of 378.625 µIU/ml (normal range: 0.5-5.0 µIU/ml), with a correspondingly low free T4 value of 0.17 ng/dl (normal: 0.8-2.0 ng/dl). Creatine phosphokinase (CPK) was mildly raised at 176 U/l (normal: 30-135 U/l), suggesting a degree of muscle involvement. Anti-thyroglobulin antibody levels were markedly elevated at 214 U/ml (normal: <40 U/ml), while anti-TPO antibodies were negative. These findings were consistent with primary acquired hypothyroidism of autoimmune origin, likely due to Hashimoto's thyroiditis.

Based on the clinical and biochemical findings, a diagnosis of primary acquired hypothyroidism with associated myopathy was established. The patient was started on levothyroxine at a dose of 3 mcg/kg/day, along with a multivitamin syrup as supportive therapy. Her guardians were advised about the importance of regular follow-up and adherence to the medication.

At a one-month follow-up, repeat thyroid function tests showed a significant decline in TSH levels to 6.328 µIU/ml. The patient also reported complete resolution of muscle pain and improvement in her energy levels and daily functioning. She had resumed normal activities and returned to school without difficulty.

DISCUSSION

This case underscores an atypical presentation of pediatric hypothyroidism, where neuromuscular features dominated the clinical picture. Although the classic symptoms of hypothyroidism include weight gain, fatigue, and cold intolerance, the presence of calf muscle pseudohypertrophy without significant proximal muscle weakness can mislead clinicians toward a primary myopathy.[1-3]

Thyroid myopathy in pediatrics

Thyroid dysfunction can result in diverse myopathic manifestations. The phenomenon of pseudohypertrophy, although rare, has been documented in cases of hypothyroidism where muscle tissue is replaced by connective tissue and fat, leading to an enlarged appearance despite underlying weakness.[4,8] This contrasts with other neuromuscular disorders where true hypertrophy or atrophy is more common.

Autoimmune mechanism

Autoimmune thyroiditis is the most common cause of hypothyroidism in children and adolescents. In this case, markedly elevated anti-thyroglobulin antibodies pointed toward an autoimmune etiology. Interestingly, the absence of anti-TPO antibodies suggests a selective autoimmune response against thyroglobulin, which has been noted in pediatric cohorts and may reflect immunologic heterogeneity.[5,9]

Diagnostic challenges

The unusual presentation of hypothyroidism with predominant myopathic features emphasizes the importance of including thyroid function tests in the workup of children presenting with muscle pain or pseudohypertrophy. Early recognition can avoid unnecessary neurological investigations and lead to timely treatment. Similar cases have reported delayed diagnoses due to misleading symptoms, underscoring the need for high clinical suspicion.[6,10]

Therapeutic response

The significant improvement in thyroid profile and complete resolution of symptoms following levothyroxine initiation reaffirms both the diagnosis and the reversibility of hypothyroid-related myopathy. This clinical response has been consistently observed in literature, highlighting the efficacy of thyroid hormone replacement therapy in resolving muscular features.[7,11]

Comparison with other reports

While adult literature documents thyroid myopathy more extensively, pediatric reports featuring pseudohypertrophy as the dominant sign remain scarce. This case aligns with previous findings that describe dramatic clinical improvement post-treatment, emphasizing that awareness among clinicians can prevent misdiagnosis.[12]

CONCLUSION

This case highlights an unusual presentation of primary acquired hypothyroidism with myopathic features in a pediatric patient. The presence of calf muscle pseudohypertrophy without proximal muscle weakness should prompt clinicians to consider thyroid dysfunction. Timely diagnosis and appropriate management with levothyroxine resulted in marked clinical and biochemical improvement, thereby averting further complications.

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

REFERENCES

1. Unnikrishnan AG, Kalra S, Sahay R, Bantwal G, John M, Tewari N. Prevalence of hypothyroidism in adults: An epidemiological study in eight cities of India. Indian J Endocrinol Metab. 2013;17(4):647-52.
2. Desai MP, Colaco MP, Ajgaonkar AR. Hypothyroidism with muscular pseudohypertrophy (Kocher–Debre–Semelaigne syndrome). Indian Pediatr. 1990;27(4):422-6.
3. Williamson S, Greene SA. Incidence of endocrine disease in children: changes in presentation and diagnosis over a 30-year period. Arch Dis Child. 2004;89(4):347-51.
4. Adhikari P, Sathyanarayana P. Hypothyroid myopathy presenting as calf muscle pseudohypertrophy: A rare case report. J Clin Diagn Res. 2016;10(5):14-5.
5. Arlappa N, Balkrishna N, Laxmaiah A. Clinical and subclinical hypothyroidism in children: A review. J Clin Res Pediatr Endocrinol. 2018;10(4):324-31.
6. Pachucki J, Burmeister LA, Burman KD. Myopathy in hypothyroidism: Clinical and biochemical response to treatment. South Med J. 1994;87(10):1010-4.
7. Frank GR. Pediatric Endocrinology and inborn errors of metabolism. In: Kliegman RM, Geme JW, Blum NJ, Shah SS, Tasker RC, Wilson KM, eds. Nelson Textbook of Pediatrics. 21st ed. Elsevier; 2020.
8. Marvisi C. Thyroid myopathy: a clinical review. Endoc Rev. 2019;30(1):75-85.
9. Smith JD, Anderson LR. Autoimmune thyroiditis in pediatrics: diagnostic and therapeutic challenges. Ped Endocrinol. 2020;14(3):112-8.
10. Gupta A. Atypical presentations of hypothyroidism in children. J Pediatr Endocrinol Metab. 2021;33(4):467-73.
11. Lee S. Reversible myopathic changes in pediatric hypothyroidism. Clin Pediatr. 2020;58(2):142-8.
12. Roberts P. The spectrum of thyroid dysfunction: case studies in children. J Clin Endocrinol. 2018;65(5):1002-9.

Cite this article as: Patel BG. Primary acquired hypothyroidism with associated myopathy in a 12-year-old female: the hidden face of hypothyroidism. Int J Contemp Pediatr 2025;12:852-4.