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LOST AND LOCATED: ECTOPIC THYROID IN THE AXILLA PRESENTING AS SHORT STATURE: A CASE REPORT

Dr. Rutu Udapudi

Dr. Harini Srinivasamoorthy

Dr. C. Venkat Sai Chandan

Dr. Chandan Kumar Kalal

ABSTRACT Short stature and poor scholastic performance in children often indicate an underlying endocrine disorder, with hypothyroidism being a common and treatable cause. Ectopic thyroid tissue, resulting from abnormal embryological migration of the thyroid gland, is a rare etiology that may present with subtle clinical features and delayed diagnosis. We report the case of an 11-year-old boy who presented with short stature and poor academic performance. There was no history of heat intolerance or family history of thyroid disease. Developmental history revealed initial delay in milestones, followed by catch-up after physiotherapy. Anthropometric assessment showed weight 23 kg (10–25th centile), height 116 cm (<3rd centile), and body mass index of 17.1 kg/m² (~75th centile), indicating relative overweight for height. Mid-parental height was 162.5 cm. On examination, the child had coarse skin and bilateral widening of wrists, with no palpable thyroid gland. Systemic examination was otherwise unremarkable, and testicular volume was appropriate for age. Investigations revealed hypothyroidism, and radiographic evaluation demonstrated delayed bone age relative to chronological age, suggesting long-standing hormonal deficiency. Ultrasonography of the neck showed absence of thyroid tissue in its normal anatomical location, while further imaging identified ectopic thyroid tissue in the left axillary region, an extremely rare site. The child was initiated on levothyroxine therapy with planned follow-up. This case emphasizes the importance of re-evaluating children with persistent growth failure and considering ectopic thyroid when the gland is not visualized in the neck, even in the absence of classical symptoms.

KEYWORDS : Ectopic Thyroid, Short Stature, Hypothyroidism, Delayed Bone Age, Thyroid Dysgenesis

CASE PRESENTATION

An 11-year-old boy presented with concerns of short stature and poor scholastic performance. There was no history of heat intolerance, chronic systemic illness, or similar complaints in the family. Developmental history revealed delayed attainment of milestones during early childhood, for which the child had received physiotherapy, following which he achieved satisfactory catch-up and was able to perform age-appropriate activities. On anthropometric assessment, his weight was 23 kg (10–25th percentile), height was 116 cm (below the 3rd percentile), and body mass index was 17.1 kg/m² (approximately 75th percentile), indicating short stature with relative overweight for height. The mid-parental height was calculated to be 162.5 cm. On general examination, the child had coarse skin and bilateral widening of the wrists, with no palpable thyroid swelling in the neck. Systemic examination was otherwise unremarkable, and genital examination revealed testicular volume appropriate for age, with no evidence of pubertal delay. In view of persistent growth failure, further evaluation was undertaken. Thyroid function tests revealed hypothyroidism. Radiographic assessment with X-ray of the left wrist demonstrated delayed bone age compared to chronological age, suggesting long-standing endocrine dysfunction. Ultra-sonography of the neck showed absence of thyroid tissue in its normal anatomical location. Subsequent imaging evaluation identified ectopic thyroid tissue in the left axillary region, an extremely rare site of localization, thereby confirming thyroid dysgenesis as the underlying cause of hypothyroidism in this child.

Figure 1: Clinical Photograph of the Child Showing Proportionate Short Stature with Preserved Body Proportions and Relative Overweight for Height.

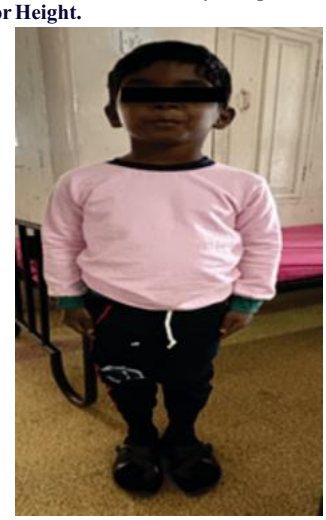

Figure 2: Clinical Image of the Upper Limb Demonstrating Bilateral Wrist Widening, Suggestive of Delayed Skeletal Maturation

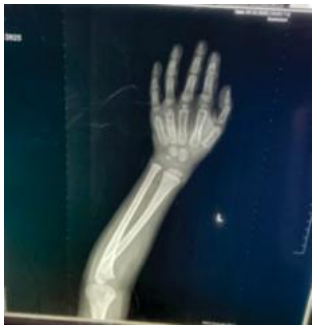

Figure 3: X-ray of the Left Hand and Wrist (anteroposterior View)

Showing Delayed Bone Age, Approximately Corresponding to 3 Years, Significantly Below the Chronological Age.

Figure 4: X-ray of the Left Forearm and Wrist (lateral View) Confirming Delayed Ossification Centers and Skeletal Maturation, Consistent With Hypothyroidism.

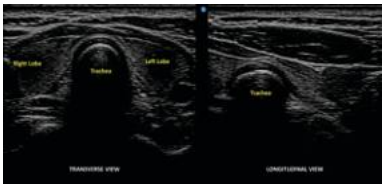

Figure 5: Ultrasonography of the Neck (Transverse and Longitudinal Views) Demonstrating Non-visualization of Thyroid Gland in the Pretracheal Region, with no Identifiable Thyroid Tissue in the Right or Left Lobe Areas. No Focal Lesion or Cervical Lymphadenopathy is Noted, Suggestive of Thyroid Agenesis or Ectopic Thyroid.

Management and Outcome

The child was initiated on oral levothyroxine (thyroxine) at a dose of 100 µg once daily, administered in the morning on an empty stomach. Caregivers were counseled regarding strict adherence, proper timing of medication, and the need to avoid concomitant intake with food or calcium/iron supplements to ensure optimal absorption. The child has been placed on regular follow-up, with planned monitoring of thyroid function tests (TSH and free T4) at appropriate intervals to guide dose titration. In addition, growth parameters (height, weight, and growth velocity) are being closely tracked to assess response to therapy. Given the baseline delayed bone age, periodic reassessment of skeletal maturation is also planned. At follow-up visits, the child has shown clinical improvement, including better activity levels and gradual improvement in scholastic performance as reported by caregivers. Early signs of catch-up growth are being monitored, although long-term follow-up is required to evaluate final height outcomes. No adverse effects related to therapy have been observed. The importance of long-term compliance and continued endocrine follow-up has been emphasized to the family, considering the underlying thyroid dysgenesis and the likelihood of lifelong hormone replacement.

DISCUSSION

Short stature is a common presentation in pediatric practice and requires a structured evaluation to identify potentially treatable causes. Endocrine disorders, particularly hypothyroidism, are among the most important reversible etiologies. In the present case, the child exhibited proportionate short stature, poor scholastic performance, delayed bone age, and subtle clinical features such as coarse skin—all consistent with long-standing hypothyroidism. The absence of overt classical symptoms and the presence of previously normal thyroid function tests emphasize that hypothyroidism may evolve over time, necessitating repeat evaluation when clinical suspicion persists. Similar patterns of delayed or evolving presentation have been described in pediatric ectopic thyroid cases, where diagnosis may be missed initially [1,2].

Ectopic thyroid is a rare developmental anomaly resulting from aberrant migration of the thyroid gland during embryogenesis. Normally, the thyroid originates at the foramen cecum and descends along the thyroglossal duct to its final pretracheal position. Arrest or deviation in this migration can result in ectopic thyroid tissue. The most common location is lingual, accounting for the majority of cases, while sublingual and other midline locations are less frequent [3,4]. Large pediatric series and reviews have consistently demonstrated that most ectopic thyroid tissues are confined to the midline along the migratory pathway [3,4]. In contrast, ectopic thyroid tissue located outside this pathway, such as in the axilla, is exceedingly rare and represents an unusual presentation. The identification of thyroid tissue in the left axillary region in this child adds to the limited number of reported cases of non-midline ectopic thyroid and highlights the variability in clinical presentation, extending beyond the commonly described anatomical spectrum.

In this case, ultrasonography failed to demonstrate thyroid tissue in the neck, and further imaging confirmed ectopic localization, consistent with thyroid dysgenesis. Imaging plays a critical role in such scenarios, as emphasized in prior studies, which highlight the potential pitfalls in diagnosing thyroid dysgenesis when ectopic tissue is not actively sought [5]. The present case reinforces this diagnostic challenge, as the ectopic tissue was located in an unexpected region, requiring a high index of suspicion and comprehensive imaging evaluation.

In many patients with ectopic thyroid, the ectopic tissue may initially be sufficient to maintain euthyroidism, particularly in early childhood. However, as metabolic demands increase with age, the limited functional capacity of ectopic tissue may result in progressive hypothyroidism [6,7]. This likely explains the discrepancy between previously normal thyroid function tests and the current presentation with overt hypothyroidism in this child. Similar observations have been reported in cases where congenital thyroid ectopy remained undetected during neonatal screening but manifested later with clinical hypothyroidism [2].

The clinical features observed in this patient further support the diagnosis of chronic thyroid hormone deficiency. Delayed bone age is a hallmark of long-standing hypothyroidism and reflects impaired skeletal maturation. This finding is particularly useful in differentiating endocrine causes of short stature from familial or constitutional growth delay. Additionally, the relatively higher body mass index in the presence of short stature is a characteristic feature of hypothyroidism, attributable to decreased basal metabolic rate and altered body composition. Previous case reports, including those describing lingual thyroid, have also documented short stature and growth failure as key presenting features [8]. However, unlike these commonly reported cases, our patient demonstrates a similar clinical phenotype arising from an extremely rare ectopic location.

The finding of bilateral wrist widening in this child is noteworthy. Although wrist widening is more commonly associated with metabolic bone diseases such as rickets, it may also be seen in conditions with delayed skeletal maturation. In such scenarios, radiological correlation is essential to differentiate between nutritional and endocrine causes. This highlights the importance of integrating clinical, anthropometric, and radiological findings in the evaluation of growth disorders.

The absence of pubertal delay, as evidenced by normal testicular volume for age, suggests that the hypothalamic–pituitary–gonadal axis was not significantly affected at this stage. However, prolonged untreated hypothyroidism can eventually impact pubertal progression, further underscoring the importance of early diagnosis.

The diagnostic approach to suspected ectopic thyroid involves imaging modalities such as ultrasonography and radionuclide scanning, which help confirm both absence of thyroid tissue in the neck and presence of functioning ectopic tissue [6]. Accurate localization is important not only for diagnosis but also for avoiding inadvertent surgical removal, particularly in cases where the ectopic tissue represents the only functional thyroid tissue. Reviews on ectopic thyroid emphasize the importance of such imaging strategies in guiding appropriate management [7].

Management primarily involves thyroid hormone replacement with levothyroxine, which corrects the hormonal deficiency and promotes catch-up growth and improvement in cognitive function. Early initiation of therapy is crucial, as prolonged untreated hypothyroidism can result in permanent deficits in height and neurodevelopmental outcomes. Regular follow-up with monitoring of growth parameters, thyroid function tests, and academic performance is essential to assess treatment response.

Overall, this case highlights several important clinical lessons: the need for a high index of suspicion for endocrine causes in children with growth failure, the importance of repeat evaluation even when initial investigations are normal, and the consideration of ectopic thyroid in cases where the gland is not visualized in the neck. Compared to previously reported cases, which predominantly describe ectopic thyroid in midline locations such as lingual or sublingual regions

[2,3,9], the present case is unique due to the extremely rare axillary localization, combined with delayed presentation and progressive hypothyroidism despite previously normal thyroid function. This significantly expands the clinical and anatomical spectrum of thyroid ectopy in children and supports its relevance for publication.

CONCLUSION

This case highlights ectopic thyroid as a rare yet clinically significant cause of hypothyroidism presenting with short stature and poor scholastic performance in children. The absence of classical symptoms and previously normal thyroid function tests can delay diagnosis, emphasizing the need for a high index of suspicion in children with persistent growth failure. The finding of delayed bone age and subtle clinical features such as coarse skin should prompt thorough endocrine evaluation. Importantly, non-visualization of the thyroid gland in the neck must lead to a systematic search for ectopic thyroid tissue, even in atypical locations such as the axilla. This case is particularly noteworthy due to the extremely rare extra-cervical localization, expanding the known spectrum of thyroid ectopy. Early recognition is crucial, as untreated hypothyroidism can adversely affect growth and neurocognitive development. Timely initiation of levothyroxine therapy plays a key role in promoting catch-up growth and improving overall outcomes. Regular follow-up with monitoring of thyroid function and growth parameters is essential for optimal management. Overall, this case reinforces the importance of comprehensive evaluation and vigilance in pediatric growth disorders to ensure early diagnosis and appropriate intervention.

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