

Case Report

The Missing Half: a Congenital Thyroid Mystery a Case Report

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Abstract

Thyroid hemiagenesis is a rare congenital anomaly characterized by the absence of one thyroid lobe, with or without agenesis of the isthmus. We report a 33-year-old woman who presented with a painless right-sided neck swelling without compressive symptoms or clinical features of thyroid dysfunction. Ultrasonography demonstrated a normal right thyroid lobe with nonvisualization of the left lobe and isthmus. Plain and contrast-enhanced CT of the neck confirmed complete agenesis of the left thyroid lobe and isthmus, with homogeneous enhancement of the normally positioned right lobe. No ectopic thyroid tissue was identified in the head and neck region. Based on these findings, congenital left thyroid hemiagenesis with isthmus agenesis was diagnosed. This case highlights the importance of imaging in recognizing this rare developmental anomaly, excluding ectopic thyroid tissue, and preventing unnecessary diagnostic or surgical intervention.

INTRODUCTION

Thyroid hemiagenesis is a rare congenital developmental anomaly characterized by the absence of one thyroid lobe, with or without agenesis of the isthmus [1]. Its true prevalence remains uncertain because most affected individuals are asymptomatic and the condition is usually detected incidentally during imaging performed for unrelated neck complaints [1,2]. The left thyroid lobe is more commonly absent than the right, and agenesis of the isthmus may coexist with lobar agenesis [1,3]. Although most patients are euthyroid, the remaining thyroid lobe may develop compensatory enlargement or other thyroid disorders, including nodular goitre, thyroiditis, hypothyroidism, and hyperthyroidism [1,4]. Ultrasonography is the primary imaging modality for diagnosis, while computed tomography and thyroid scintigraphy may provide additional anatomical and functional confirmation and help identify ectopic thyroid tissue [3,5]. We report a rare case of congenital left thyroid hemiagenesis with agenesis of the isthmus in a 33-year-old woman presenting with a painless right-sided neck swelling.

Case Presentation

A 33-year-old woman presented with a vague swelling over the right side of the neck. The swelling was painless and was not associated with any recent increase in size or local discomfort. There were no compressive symptoms such as dysphagia, dyspnoea, choking sensation, or change in voice. She did not report symptoms suggestive of thyroid dysfunction, including palpitations, tremors, excessive sweating, heat or cold intolerance, unexplained weight gain or weight loss, altered bowel habits, or menstrual disturbances. There was no significant family history of thyroid disease.

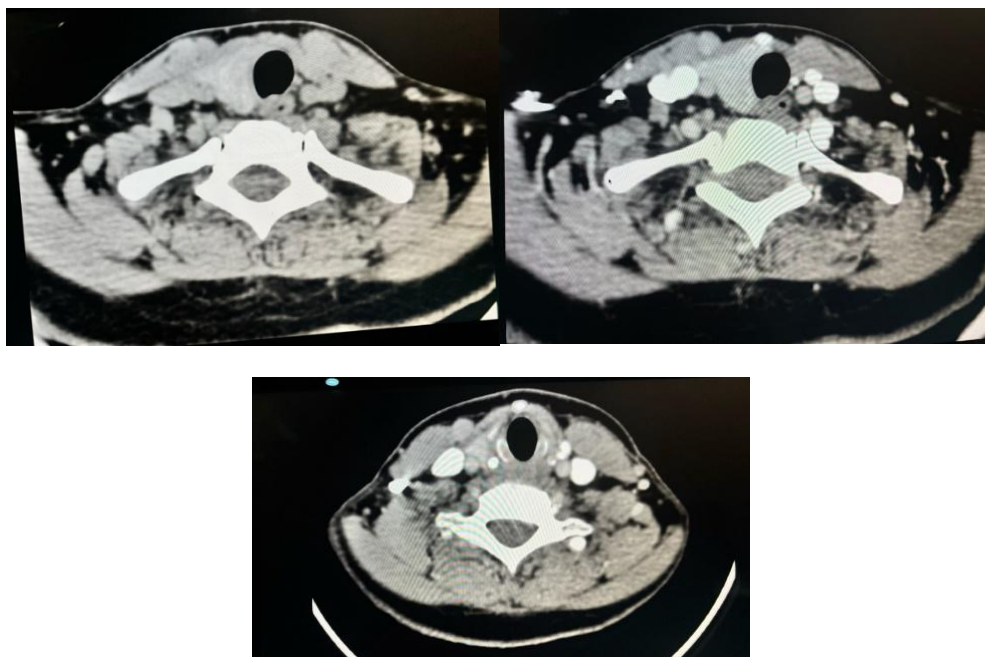
On general examination, the patient was clinically stable, with no obvious features of hyperthyroidism or hypothyroidism. Local examination of the neck revealed a soft, mobile swelling over the right thyroid region that moved with deglutition, suggesting a thyroid origin. The overlying skin appeared normal, with no erythema, dilated veins, or other cutaneous changes. No palpable cervical lymphadenopathy was noted. There were no ocular or cutaneous manifestations suggestive of thyroid dysfunction.



Ultrasonography of the neck was performed as the initial imaging investigation. It demonstrated a normally positioned right thyroid lobe with preserved size, contour, and parenchymal echotexture. Vascularity of the right lobe was also maintained. However, the left thyroid lobe and isthmus could not be visualized in their expected anatomical locations. These findings raised the possibility of a congenital developmental anomaly of the thyroid gland, particularly left thyroid hemiagenesis with associated agenesis of the isthmus.

To further delineate the thyroid anatomy and confirm the ultrasonographic findings, plain and

contrast-enhanced computed tomography (CT) of the neck was performed. CT demonstrated complete absence of thyroid tissue in the expected location of the left thyroid lobe and isthmus. The right thyroid lobe was normally located and showed homogeneous attenuation on plain CT with uniform and intense enhancement following intravenous contrast administration. There was no evidence of a focal lesion within the visualized right thyroid lobe. The surrounding neck structures were unremarkable, and no significant cervical lymphadenopathy was identified.



The CT images were also carefully evaluated for the presence of ectopic thyroid tissue. No ectopic thyroid tissue was identified within the visualized head and neck region. The combined ultrasonographic and CT findings therefore confirmed the anatomical absence of the left thyroid lobe and isthmus rather than displacement of thyroid tissue to an ectopic location. Based on the clinical presentation and

concordant ultrasonographic and CT findings, a final diagnosis of congenital left thyroid hemiagenesis with associated agenesis of the isthmus was established. The presence of a normally located and morphologically preserved right thyroid lobe, together with the absence of identifiable ectopic thyroid tissue, supported the diagnosis of this rare congenital thyroid developmental anomaly.

DISCUSSION

Thyroid hemiagenesis is a rare congenital developmental anomaly resulting from failure of formation or migration of one thyroid lobe, with or without absence of the isthmus. The exact embryological mechanism remains uncertain, although abnormalities in thyroid migration and lobulation have been proposed. Most cases are sporadic, but familial occurrence and reports in monozygotic twins suggest a possible genetic contribution [6]. A characteristic feature of thyroid hemiagenesis is the predominance of left-sided involvement. Lee et al. reported left-lobe agenesis in 72.7% of cases compared with 27.3% involving the right lobe; notably, absence of the isthmus was observed in all patients with left-sided hemiagenesis in their series [7]. Our case demonstrated the same anatomical pattern, with complete absence of the left thyroid lobe and isthmus and preservation of the right lobe. Most patients remain asymptomatic because the remaining thyroid lobe generally provides adequate hormone production. However, compensatory stimulation may result in enlargement of the remaining lobe [6]. Various associated thyroid abnormalities have been described. Wu et al. reported associations with compensatory hypertrophy, hypo- and hyperthyroidism, simple or multinodular goitre, and thyroid carcinoma [8]. Graves' disease has been reported by Cansu et al. [9], while Nsame et al. described thyroid hemiagenesis associated with Hashimoto's thyroiditis [10]. More recently, Wang et al. reported thyroid hemiagenesis associated with both papillary thyroid carcinoma and Hashimoto's thyroiditis [11]. These observations highlight the importance of evaluating the remaining thyroid lobe for structural and functional abnormalities. Imaging is fundamental for establishing the diagnosis. Ultrasonography is generally the initial investigation because it can demonstrate the absence of a thyroid lobe and simultaneously assess the morphology of the remaining thyroid tissue [12]. CT provides more detailed anatomical evaluation and can help differentiate congenital hemiagenesis from previous hemithyroidectomy or other acquired causes of unilateral absence. Lee et al. demonstrated characteristic CT findings that may assist in this distinction [7]. In our patient, ultrasonography initially demonstrated nonvisualization of the left lobe and isthmus, while CT subsequently confirmed their complete absence and demonstrated a normally enhancing right thyroid lobe. Thyroid

scintigraphy provides complementary functional information and may be particularly useful for detecting ectopic functioning thyroid tissue. However, absence of radionuclide uptake on one side is not specific for hemiagenesis and may also occur in severe hypofunction, thyroiditis, or infiltrative disease; therefore, correlation with anatomical imaging is important [12,13]. In the present case, no ectopic thyroid tissue was identified within the evaluated head and neck region on CT. Recognition of this anomaly is clinically important because an absent lobe may otherwise be mistaken for an acquired abnormality. McHenry et al. emphasized that accurate preoperative recognition of thyroid hemiagenesis may prevent unnecessary neck exploration [14]. Moreover, because all functioning orthotopic thyroid tissue is confined to a single lobe, careful evaluation of any disease affecting the remaining lobe is essential. In the absence of associated pathology, treatment is generally not required, although clinical assessment, thyroid function testing, and appropriate imaging follow-up may be considered [6]. The present case emphasizes the complementary role of ultrasonography and CT in identifying this rare congenital thyroid anomaly.

CONCLUSION

Thyroid hemiagenesis is a rare congenital anomaly that is often detected incidentally in otherwise asymptomatic individuals. This case highlights congenital absence of the left thyroid lobe and isthmus with preservation of the right lobe. Ultrasonography, supplemented by CT, enables accurate anatomical diagnosis and helps exclude ectopic thyroid tissue and other causes of an apparently absent thyroid lobe. Recognition of this condition is important to avoid diagnostic errors and unnecessary intervention. Careful assessment and appropriate follow-up of the remaining thyroid lobe are advisable because associated functional and structural thyroid abnormalities may develop.

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