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Thyroid Hemiagenesis: A Rare Finding

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Abstract:

Thyroid hemiagenesis is a rare embryological condition characterized by absence of one of the lobes of the thyroid gland, with the left lobe being absent in 80% of cases and right lobe absent in 20% of cases (a left to right hemi-agenesis ratio of 4:1). We report a case of 25 year old female who presented with right neck mass gradually increasing in size over a period of 2 months. Patient had the history of weight loss and fatigability. The patient work up included an ultrasound neck followed by plain CT neck and contrast CT neck. Ultrasound neck showed absence of left lobe and part of isthmus of the thyroid gland with the right lobe showing features of thyroiditis. These findings were supported by plain and contrast CT neck findings.

Key words: Female, Neck, Thyroiditis, Tomography, Weight Loss.

Introduction

Thyroid hemiagenesis is a rare congenital condition characterised by the absence of one of the thyroid lobes with its prevalence rate being 0.02% [1]. Embryological development of the thyroid gland starts in the primitive pharynx in form of an invagination of the endoderm. This invagination grows ventrally while remaining attached to the pharyngeal floor. This thyroid rudiment will then migrate to its actual anatomical position that is anterior to the pharynx and after that it begins to grow laterally to create the bilobed thyroid gland [2]. Congenital thyroid anomalies may be caused either by abnormal descent of the gland or by incomplete genesis of a lobe. However, the etiology of the hemiagenesis still remains unclear. There may be a genetic component to the etiology as this rare condition has been documented in monozygotic twins [3].

The first case of hemithyroid agenesis was reported in 1866 [3]. We report a patient who presented to our department with a right neck mass and an incidental finding of a hemiagenesis of the left thyroid lobe and part of the isthmus. We also present a relevant review of literature with emphasis on the clinical presentation.

Case Report

A 25 year old female patient reported to the hospital with right sided neck swelling. The swelling gradually increased in size over the preceding two months. This was associated with minimal weight loss and fatigability. She confirmed an uneventful pre-illness medical history. The patient underwent biochemical screening for thyroid dysfunction which

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revealed elevated levels of TSH and reduced levels of T3. T4 was found to be within normal limits.

An ultrasound scan of the neck was performed which showed absent left thyroid lobe and part of the isthmus. Right thyroid lobe and remnant isthmus was enlarged in size and showed heterogeneous echotexture, multiple small hypoechoic nodules and coarse septations [Fig.1,2]. On colour Doppler examination, increased vascularity was noted in the right thyroid lobe and visualized part of the isthmus [Fig.3]. Few adjacent enlarged lymph nodes were also observed.

Following this, plain CT neck and contrast CT neck were performed which confirmed the findings of absent left thyroid lobe and part of the isthmus. Right lobe was enlarged with a homogeneous intense post-contrast enhancement (47-215 HU) [Fig.4-7]. CT scan failed to reveal ectopic thyroid tissue in the head and neck region. The ultrasound and CT features in correlation with FNAC of the right thyroid lobe [Fig.8] led us to the diagnosis of thyroid hemiagenesis with features of chronic thyroiditis in the remnant right thyroid lobe.

Discussion

Hemithyroid agenesis is a rare congenital anomaly of the thyroid and been has found to be three time more common in females as compared to males [4]. In 80% cases the left thyroid lobe is absent and in about 50% of patients with left hemithyroid agenesis will also have an absent thyroid isthmus [5]. In most of the cases it is an incidental finding with associated thyroid disorders in the remaining lobe. The diagnosis of thyroid hemiagenesis should be considered in any patients where, on physical examination, no apparent thyroid tissue is noted on one side of the neck. The coexisting disorders of the remnant thyroid lobe may be hyperthyroidism, hypothyroidism, multinodular goiter, chronic thyroiditis, adenocarcinoma or

Fig.1: Ultrasound demonstrating absent left thyroid lobe.

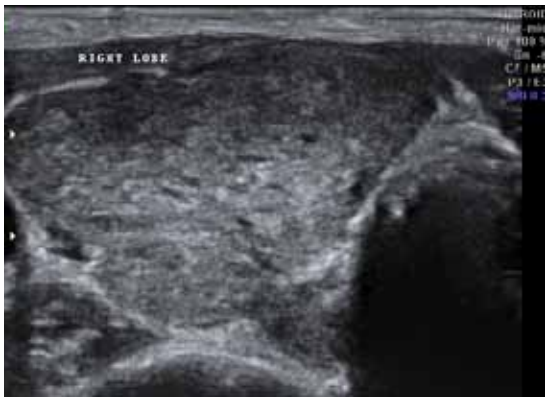

Fig.2: Ultrasound demonstrating enlarged right thyroid lobe with heterogenous echotexture and multiple small hypoechoic nodules and coarse echogenic septations.

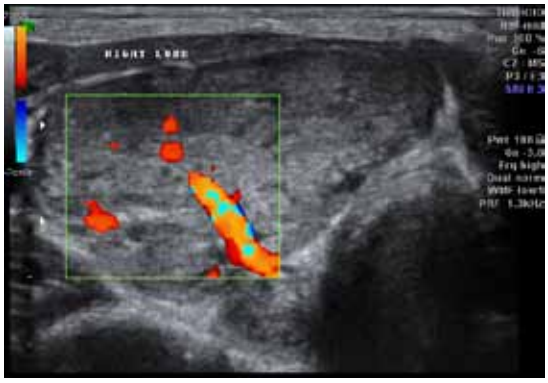

Fig.3: Increased vascularity noticed on Doppler examination.

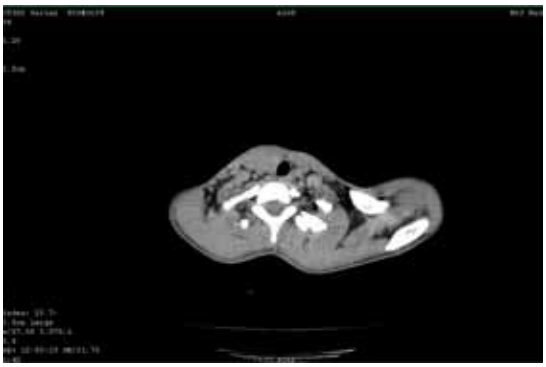

Fig.4: Plain CT study axial section showing absent left thyroid lobe.

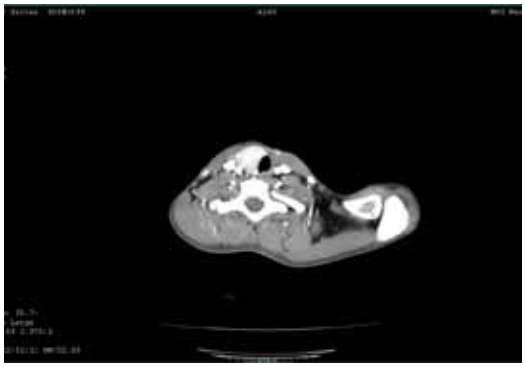

Fig.5: Contrast CT study at the same level as above showing homogenously enhancing enlarged right thyroid lobe (arrow) and absent left thyroid lobe.

papillary thyroid carcinoma [3,6]. Most common of these being hyperthyroidism, associated with thyroid hemiagenesis. However, as seen in our patient, hemithyroid agenesis can also be associated with a hypothyroid or euthyroid states as well [3,4,6]. A recent large cohort case study revealed that patients with thyroid hemiagenesis were more likely to develop thyroid pathology in the remnant thyroid lobe, including functional, morphological, and autoimmune diseases with Hashimoto's thyroiditis being the most common of the pathologies encountered [7]. The differential diagnosis would include unilateral inflammatory

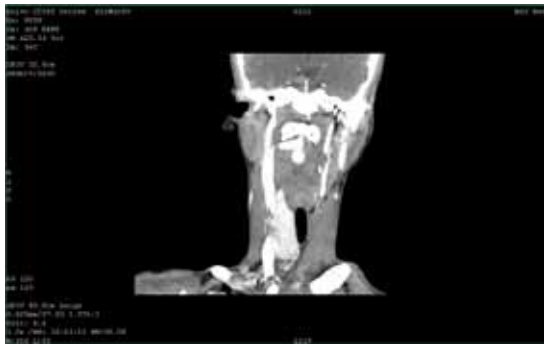

Fig.6: Coronal reconstruction showing right thyroid lobe and absent left thyroid lobe.

Fig.7: Part of the isthmus along with right thyroid lobe (arrow) is visualized on this coronal reconstructed image.

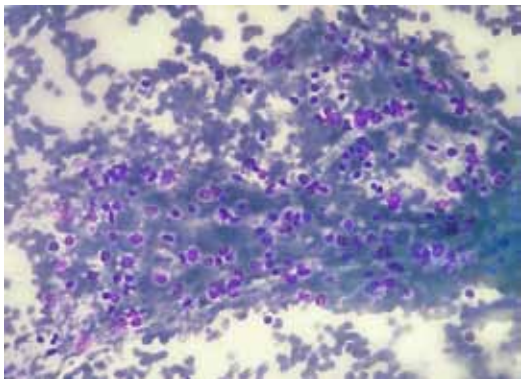

Fig.8: FNAC of the right thyroid lobe showing follicular epithelial cells and lymphocytes giving impression of lymphocytic thyroiditis (MGG x 200).

disease and infiltrative disease, such as amyloidosis. In our case, we used ultrasound neck and computerized tomography scan of the head and neck region to diagnose the thyroid hemi-agenesis. Other diagnostic methods which can be used are magnetic resonance imaging (MRI), and thyroid scintigraphy using radioisotopes. Ultrasonography can demonstrate an absent lobe and is also helpful in follow up of euthyroid patients [8]. Scintigraphy has also been used but is not necessarily required to make the diagnosis of an absent thyroid lobe. However, scintigraphy has been helpful in identifying and locating ectopic thyroid tissue [5].

Conclusion

The purpose of this presentation is to discuss and review the literature on thyroid hemi-agenesis and present a rare case of absent left thyroid lobe with thyroiditis in the remaining right thyroid lobe in order to create awareness in the masses regarding the associated conditions and offer appropriate work up if indicated.

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