Case Report

Accidentally Detected Thyroid Hemiagenesis: A Rare Case & Literature Review

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Abstract

Thyroid gland hemiagenesis is a rarely reported congenital anomaly wherein developmental failure of one of the thyroid lobe with or without isthmus can be seen. Its prevalence ranges from 0.05%-0.2%. From these very few reported cases, a female preponderance has been noted. The left lobe appears to be more commonly affected than the right. Frequently these patients are hyperthyroid, but euthyroid and hypothyroid cases have also been reported. Our objective here is to report a case, review the literature and discuss the management of these cases. The idea of this study started from an accidentally detected case of left Hemithyroid agenesis when the literature review was done in PubMed using key words like “congenital absence of thyroid lobe”, “Hemithyroid agenesis”, “thyroid hemiagenesis” and “absent thyroid gland”. We report a case of congenital absence of left lobe of thyroid and isthmus in a 65 year old lady, who presented with history of dyspnea and cough with expectoration. A contrast enhanced computed tomography (CECT) scan of chest showed an incidental finding of hemiagenesis of left thyroid gland. Further evaluation with neck Ultrasonography and Doppler study reconfirmed the diagnosis. The thyroid function tests were essentially within normal limits. She was diagnosed to have Pericardial effusion and treated accordingly under cardiology department. Our management included proper counseling of the patient and family regarding the known pathological entities associated with these conditions like adenocarcinoma, chronic thyroiditis, adenoma and multinodular goiter (MNG). This case being an incidental finding with negative work up was kept under observation.

ABBREVIATIONS

MNG: Multi Nodular Goiter; OPD: Out Patient Department; CECT: Contrast Enhanced Computer Tomography; USG: Ultrasonography

INTRODUCTION

Hemiagenesis of thyroid gland is an extremely rare congenital anomaly. The first ever reported case was by Handfield-Jones in 1866, while till date almost 300 cases have been reported worldwide [1]. Literature review shows prevalence ranging between 0.05%-0.2% without any geographical predominance [2,3]. During the embryogenesis of thyroid gland, there is endodermal invagination in primitive pharynx, which grows ventrally while the upper end remains attached to floor of the pharynx. The exact pathogenesis is not known, however the abnormal descent or incomplete genesis of a thyroid lobe is the most accepted one. The occurrence of this entity in monozygotic twins may point towards the role of genetics. The left lobe is the more commonly (80%) affected than the right. The isthmus may also be absent in 40-50% of the cases [4,5]. This anomaly is seen to be three times more common in females when compared to males [6].

We report here a case of incidentally detected hemiagenesis of the left lobe and isthmus of the thyroid in a 65year old lady who was having cough and dyspnea later diagnosed to have pericardial effusion and managed accordingly.

CASE PRESENTATION

A 65-year old lady with complaints of dyspnea on exertion and cough with expectoration for 3 months was referred to radiology department for a CECT scan of thorax. The scan revealed presence of pericardial effusion. An incidental finding noted was absence of left lobe and isthmus of thyroid gland (Figure 1). Further history taking revealed no previous history of any neck surgery. The patient was then taken for ultrasonography (USG) and Doppler scans of neck (Figure 2), which showed absence of the left lobe of thyroid and isthmus with decreased vascularity in right lobe. However, No nodule/lesion could be seen in the present lobe.
No further ectopic thyroid tissues could be noted on any of the radiological tests. T3, T4 and TSH levels were found to be within normal limit. Thus, an incidental diagnosis of euthyroid patient with left thyroid hemiagenesis was confirmed and possibility of thyroid hypoplasia was excluded. The patient was later treated for her cardiac pathology. The patient and family members were counseled regarding the entity and kept under observation.

DISCUSSION

Thyroid agenesis is a rarely reported entity. This agenesis may be complete or partial. The partial group includes unilateral and isthmic agenesis. During embryogenesis, the origin of thyroid gland is from foramen cecum between the first and second pharyngeal pouches, which grows ventrally in the form of a thyroid diverticulum. The exact etiology of hemithyroid agenesis is not known, but postulations ranges from role of genetics, failure of descent of diverticulum towards the trachea, or defective lobulation [7].

Reported literature has shown presence of hypothyroidism, hyperthyroidism, MNG, thyroiditis, adenocarcinoma and papillary thyroid carcinoma associated with thyroid agenesis [8]. Our case was euthyroid but more commonly the patients of hemithyroid agenesis are hyperthyroid. In 2010, a large cohort case-control study by ruchala et al., on increased risk of thyroid pathology in patients with hemithyroid agenesis proposed that these patients are more likely to develop functional, morphological and autoimmune pathologies of thyroid gland like Hashimoto’s thyroiditis, nodular variant and nontoxic nodular goiter [9].

A number of imaging modalities have been found helpful in diagnosing these cases of hemithyroid agenesis such as USG, magnetic resonance imaging (MRI), and radioisotopes thyroid scintigraphy [10]. USG easily demonstrates anomaly of thyroid gland and is a very helpful tool during follows up to detect any new pathology. Although, diagnostic radioisotope scintigraphy is rarely used, it has more roles to play in locating ectopic thyroid. Multiple case reports have proven this role of radioiscntigraphy in identifying ectopic thyroid tissues in lingual thyroid and pre laryngeal area [11,12].

To conclude, thyroid gland hemiagenesis is a rare developmental anomaly of unknown etiology, found more commonly in female patients and on the left side. It is important to counsel and educate these cases for further workup, as the gland may get involved by a whole spectrum of thyroid diseases ranging from benign to malignant. Incidental hemithyroid agenesis with a negative workup can be kept on regular follow up using serial ultrasonography.

REFERENCES

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