Case Report

A case of hemiagenesis of thyroid with double ectopic thyroid tissue

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ABSTRACT

Developmental abnormalities of the thyroid gland are very rare. The most common abnormalities include ectopic thyroid tissues that are commonly seen in lingual or sublingual location, agenesis, and hemiagenesis of the thyroid gland. These developmental defects may or may not be associated with thyroid dysfunction. Our case is an 18-year-old male who presented with swelling in the neck of 4-year duration. Clinical examination revealed an oval-shape swelling in the left side of the thyroid gland. The ultrasound and the nuclear scan report revealed the presence of thyroid hemiagenesis of the right lobe with isthmus along with double ectopic thyroid tissue at suprahyoid and infrahyoid region. His thyroid function test showed elevated thyroid-stimulating hormone (TSH) and normal free T4. We report a very rare case of thyroid hemiagenesis with double ectopic thyroid tissue; and to the best of our knowledge, this is the first report in the world literature.

Key words: Ectopic thyroid, hemiagenesis, hypothyroidism

Introduction

Thyroid gland develops from the median endodermal thickening in the floor of the pharynx that later form the median diverticulum. This grows caudally as bifurcating tubular ducts to form the lateral lobes and isthmus.[1] Abnormalities in the development may result in defective organogenesis or descent, complete or partial absence of the gland with or without ectopic thyroid tissue. Ectopic thyroid tissue is an entity that is characterized by the presence of thyroid tissue in locations other than anterolateral region of second and fourth tracheal cartilages. It is commonly situated in the base of the tongue but can be present in the midline, anywhere between the foramen caecum and the normal location of the thyroid gland. It has been very rarely found in the mediastinum.[2,3] Thyroid hemiagenesis is a rarer congenital abnormality that is characterized by the absence of one lobe of the thyroid gland with or without the absence of the isthmus.[4] We report here a patient with both the above congenital abnormalities.

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Case Report

An 18-year-old male presented with gradual onset swelling in front of the neck for 4 years. It was painless and was not associated with dysphagia, dysphonia, or dyspnea. There was no history of tremors, palpitations, or change in weight or bowel habits. There was no history of neck surgery or irradiation. Clinical examination of the neck revealed a soft swelling in the region of the left lobe of thyroid which moved freely with deglutition. There was no cervical lymphadenopathy and no eye or skin changes suggestive of autoimmune thyroid disease. Systemic examination was normal with no dextrocardia. Investigations revealed the following: (i) Thyroid function test was suggestive of subclinical hypothyroidism [free T4 1.1 ng/ml (normal 0.8-1.7), TSH 8.8 mIU/L (normal 0.4-4.2)]; (ii) antithyroperoxidase

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antibody was negative; (iii) ultrasound neck showed absent isthmus and right lobe with normal echogenecity and vascularity of the left lobe suggestive of hemiagenesis of thyroid; and (iv) technetium uptake nuclear scan of the thyroid gland revealed hemiagenesis of right lobe of the thyroid gland and also showed uptake in the suprahyoid and infrahyoid position suggestive of double ectopic thyroid gland [Figure 1].

Discussion

Developmental anomalies of the thyroid gland are rare and are usually due to the result of abnormal descent of the thyroid gland. Hemiagenesis is a form of thyroid dysgenesis, in which one thyroid lobe fails to develop, with or without agenesis of the isthmus. It was first described in 1866 by Handsfield-Jones.[5] The absence of one thyroid lobe usually goes unnoticed as they do not cause clinical symptoms *per se* and the true prevalence is not exactly known. Studies on adults based on autopsy, or perioperative findings may have a pathological bias such as postinflammatory sclerosis of one lobe and may not reflect the true prevalence. Large studies with ultrasound screening in children show varying prevalence of hemiagenesis from 0.05% to 0.2%.[6-8] The left lobe of the thyroid gland is involved in most of the cases of thyroid hemiagenesis.[8,9] The isthmus may also be absent in up to half of them.[10] In hospital-based studies done in adults, the disorder is seen more common in females with a male:female ratio of 1:3.[6] However, in a prevalence study conducted in healthy children, the male to female ratio was 1.4:1.[8] The exact etiology of unilateral agenesis of the thyroid gland is not known, but several genetic mechanisms, a descent defect from the floor of primitive pharynx to trachea, or a lobulation defect has been postulated to explain thyroid hemiagenesis.[11,12] It is unclear if the disturbance of the lobulation process is due to environmental factors or inherited genetic defect. The prevalence of ectopic thyroid tissue from postmortem studies suggests that ectopic thyroid tissue may be seen in as many as 7-10% of adults, along the path of the thyroglossal duct.[13] There has been a case report of thyroid hemiagenesis with a single lingual ectopic thyroid tissue.[14]

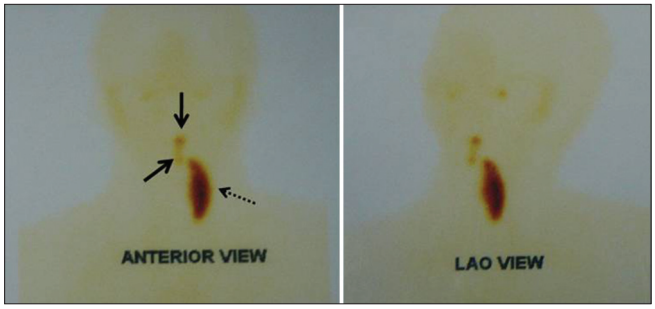

Figure 1: Technetium uptake nuclear imaging of the thyroid tissue (anterior and left antero-oblique view). The image shows the absent right lobe and isthmus of the thyroid gland with double ectopic thyroid tissue as shown by black arrow. The dashed arrow shows the presence of normal left lobe of the thyroid gland

The clinical presentation of thyroid hemiagenesis is highly variable. Although patients may have a normal thyroid lobe with euthyroidism, both hypothyroidism and hyperthyroidism are known to occur.[15] Thyroid function is abnormal in 38-47% of patients.[10] TSH was observed to be elevated in children with thyroid hemiagenesis when compared with normal children, presumed to be due to overstimulation of the normal lobe and it is suggested that this may not truly represent subclinical hypothyroidism.[8] The index case also had elevated TSH and he had recent increase in size of the gland and hence we had treated the patient with thyroxine therapy. Other anomalies such as thyroiditis, adenoma, multinodular goiter, and papillary carcinoma have also been reported.[16,17]

Most cases of thyroid hemiagenesis are discovered when patients present with a lesion in the functioning lobe or are diagnosed incidentally.[6] Clinical examination is of limited diagnostic value, but tracheal rings may be easily palpable in patients with absent isthmus. Our patient presented with swelling of the existing thyroid lobe and diagnosis was made incidentally with ultrasonogram. Thyroid scintigraphy using technetium or iodine is helpful in these situations but could be misleading on its own. The reasons for nonvisualization of one thyroid lobe include neoplasm, contralateral autonomous solitary thyroid nodule that is suppressing normal tissue, inflammatory, and infiltrative diseases of the thyroid such as amyloidosis.[18] Therefore, it is reasonable to confirm scintigraphy findings with ultrasound, CT, or MRI. Ultrasonography is a better diagnostic tool as it is widely available and cost-effective with no radiation exposure to the patient.[19] Nuclear scans can also identify small ectopic thyroid tissue that can be missed by other imaging techniques.

Conclusion

We report this case for its rare and interesting features: (i) Hemiagenesis of the right lobe with isthmus is very uncommon compared with left thyroid hemiagenesis. (ii) the presence of thyroid hemiagenesis with double ectopic thyroid tissue is extremely rare to been seen in the same patient. To the best of our knowledge, there are no previous cases reported in the literature.

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