Thyroid Hemiagenesis with Ipsilateral Parathyroid Adenoma; Case report with Review of the Literature

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ABSTRACT

Thyroid hemiagenesis (THA) is a rare congenital anomaly in which one lobe of thyroid gland fails to develop during embryological stage. Agenesis may be unilateral, total or isthmic. Left thyroid lobe is more commonly involved than right lobe in hemiagenesis. Agenesis of the isthmus was seen in 50% of cases. Left sided hemiagenesis is more common than right sided hemiagenesis with a Left to right ratio of 4:1. Clinically patients can be euthyroid, hypothyroid or hyperthyroid. Often it is diagnosed as an incidental finding during ultrasonography (USG) study of neck, which can easily diagnose this condition.

Actual incidence of THA is unknown; most cases are diagnosed in patients admitted for thyroid scan or thyroid surgery because of suspicion of other thyroid abnormalities. This explains high frequency of association of hemiagenesis with other thyroid abnormalities such as multinodular goiter, adenoma, hyperthyroidism, hypothyroidism, chronic thyroiditis, and carcinoma.

Khaladkar, et al. emphasized that surgeon when planning a thyroidectomy must be aware of rare anatomical variations because such anomalies are not noticed in the differential diagnosis, investigations and treatment.

We report a rare case of 46 years old female patient who presented with nontoxic nodular goiter and intraoperatively we found a right sided multinodular goiter including the isthmus with complete absence of the left thyroid lobe with ipsilateral inferior parathyroid adenoma which is according to the available studies is the sixth case recorded worldwide till now.

Key words: thyroid hemiagenesis, parathyroid adenoma

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INTRODUCTION

Thyroid gland is the first endocrine gland to start developing in the embryo [(1)]. It is well known for its developmental anomalies such as persistence of pyramidal lobe, thyroglossal duct, agenesis of isthmus, agenesis or hemiagenesis of thyroid gland, or aberrant thyroid glands [(2)].

These anomalies may cause clinical functional disorders and should be kept in mind while doing surgery on the gland [(3)].

Thyroid gland develops as a median thickening of endoderm on the floor of the pharynx between the first and second pharyngeal pouches. Later, this area invaginates to form a median diverticulum, which appears in the later half of 4th week [(4)]. The median diverticulum grows as a bifurcating tubular duct from which both isthmus and both lateral lobes of the thyroid are developed [(5)].

Though the cause of unilateral agenesis of thyroid gland is unknown, there are two proposed theories related with etiology are a descent defect from the floor of primitive pharynx to trachea, and also it is believed to arise from the failure of original anlage to become bi-lobed and spread out laterally to both sides [(2,3)].

Embryologically, parathyroid glands develop from the dorsal epithelium of the third and fourth pharyngeal pouches [(6)]. The inferior parathyroid gland, from the third pharyngeal pouch, migrates with the thymus caudally to lie on the dorsal surface of the inferior aspect of the thyroid gland. The superior parathyroid gland, from the fourth pharyngeal pouch, migrates very little before it rests on the dorsal and superior aspect of the thyroid gland [(7)] . This explains why parathyroid adenomas can be found in association with thyroid hemiagenesis in an anatomically normal position [(8)].

Handsfield John first described thyroid hemiagenesis in 1866 [(9-10)]. The functioning lobe of thyroid gland can be a site of pathological changes similar to normally developed gland and may present with a spectrum of diseases like

multinodular goiter, colloid goiter, follicular adenoma, thyroiditis, hypothyroidism and hyperthyroidism. Other associated anomalies with THA include unilateral absence of superior and inferior thyroid artery and right sided aortic arch [(10)].

Melnick and Stenkowski reported four patients with THA and described hockey stick sign by imaging studies in case of THA. They reviewed the literature that revealed a total 94 cases of THA in 1981. Until 1986, thirteen more reported cases of THA were reported. Thus bringing the total to just over 100. Between 1986 and 1999, most of the studies were designed as case reports [(1)]. In 2003, a THA with multinodular goiter was reported from Turkey and their review of literature revealed 270 cases with THA. After this study, five more case reports with THA were reported in literature thus bringing total to over 270 [(4)].

Thyroid agenesis should be suspected clinically when there is no palpable thyroid tissue felt on one or either side of the neck. Radiological investigations hold a key role in detecting THA. USG, CT and thyroid scintigraphy are useful in detecting this condition. USG is noninvasive technique, which is easily available, and easy to perform with no radiation hazards. Thyroid scintigraphy is useful in THA from a suppressed lobe. It will show total absence of uptake while THA activity will reappear in suppressed lobe [(10)]. Thyroidal hemiagenesis with presence of isthmus has the appearance of a hockey stick on 99 m technetium pertechnetate thyroid scan [(1)].

Most cases of THA are diagnosed when patients present with a lesion in functioning lobe. Clinically hemiagenesis of thyroid lobe and isthmic agenesis can be suspected if tracheal rings are easily palpated and edge of sternomastoid on the affected side is nearer the midline overlying the trachea [(7)].

The incidence rate of hemiagenesis varies in different studies; researchers have estimated an occurrence rate of less than 0.2 % with female population suffer more than male [(11)]. In hospital-based studies done in adults, the disorder is seen more common in females with a female: male ratio of 1:3 [(2)]. However, in a prevalence study conducted in healthy children, the male to female ratio was 1.4:1[(12)].

Thyroid hemiagenesis could be found in some families suggesting a genetic cause [(13)]. GCMB gene is important for normal synthesis of parathyroid hormone in humans and could be involved in parathyroid adenoma genesis [(14)]. Disorders that are associated with THA have been largely related to the thyroid gland, but some cases of superimposed parathyroid abnormalities have also been reported. These are mostly coexisting parathyroid adenomas on the ipsilateral side of THA and have been single or even double [(15)].

Case Report

We are presenting a case of 46-year-old female patient presented to the clinic with a nodular goiter for six months duration, without any other apparent neck swelling or cervical lymphadenopathy. She was clinically and biochemically euthyroid, with no other comorbid disease.

An ultrasonography (USG) of the neck showed a mildly enlarged thyroid gland with inhomogeneous echotexture well-defined 3*1.7*2.4 cm solid mass lesion involving right thyroid lobe suggestive of thyroid adenoma. In addition, small, 7mm diameter cystic nodules seen in both lobes. A further review of the ultrasonography did not reveal any tracheal compression or retrosternal goiter extension or cervical lymphadenopathy also without any fluid collection. Ultrasonography of the neck did not mention any signs of left thyroid hemiagenesis, or the presence of left parathyroid adenoma.

Fine needle aspiration Cytology of the prominent right thyroid nodule was performed and revealed features of follicular lesion with a possibility of follicular neoplasm or hyperplastic nodule. Therefore, the decision for surgery was taken and preoperative investigations were done including triiodothyronine (T3), thyroxine, (T4), thyroid stimulating hormone (TSH), serum calcium, serum parathormone level, vitamin D3 with examination of the vocal cords, which all revealed normal results.

Right hemithyroidectomy (right lobectomy with isthmus) was performed, we found that the left lobe of the thyroid gland was completely absent but there was a big inferior parathyroid adenoma on the ipsilateral side of hemiagenesis and it was excised after we identified the other parathyroid gland which were all normal in size and preserved. Then the entire specimens were sent

for histopathology. Postoperatively the patient ran uneventful recovery.

The histopathology report, The histological picture is that of benign thyroid simple nontoxic colloid multinodular goiter with associate marked retrogressive changes in the form of variable sizes colloid nodules, minimal lymphoid aggregate with absence of active germinal center with no evidence of hyperplastic thyroid follicular tissues or malignancy is seen.

Regarding the Left inferior parathyroid swelling (excisional biopsy specimen), the gross examination detected a single parathyroid gland measure 2.0 cm with central cystic degenerative changes contain hemorrhagic fluid thickened fibrotic capsule all taken through the surgery. The histopathological picture is that of benign parathyroid nonfunctioning oxyphil adenoma with characteristics abundant eosinophilic granular cytoplasmic cytomorphological changes.

DISCUSSION

Thyroid hemiagenesis is a rare abnormality in which one thyroid lobe fails to develop. A careful clinical assessment, including thyroid function tests, thyroid ultrasonography and scintigraphy, plays an important role in diagnosis of thyroid hemiagenesis [(16)].

The absence of one thyroid lobe is usually asymptomatic and is often being diagnosed incidentally or during assessment for thyroid related or non-related conditions [(17)]. Clinical and biochemical presentation of THA are highly variable. Although patients with THA may have a normal thyroid lobe with euthyroidism, both hypothyroidism and hyperthyroidism are known to occur. [(18)]. Thyroid function may be altered in 38% to 47% of all THA cases [(19)].

The diagnosis of THA was established based on sonographic assessment combined with scintiscan, which were performed to exclude the presence of functional thyroid tissue on the contralateral side to the lobe demonstrated in ultrasonography, and to visualize potential accessory or probable ectopic thyroid tissue located ectopically [(20)].

Marshall first described thyroid hemiagenesis in 1895. He found one case in 60 autopsies of children [(1)]. Discovery rate of THA by scanning was reported by Andrew to be seven cases in 708 scans and Hamburger to be four in 7000 patients. A large study conducted in 1972 on 12456 patients undergoing surgery revealed seven cases of THA. Friedman et al. in 1979 found six cases of THA in 12000 scans [(21)].

Review of literature in year 2000 documented 256 cases of hemiagenesis. Fifty-one new cases were published between 2000 and 2007 in English literature [(3)]. 310 cases of THA have been reported in world literature to date [(9)].

Regarding parathyroid adenoma, review of the literature showed five other cases of parathyroid adenoma with thyroid hemiagenesis on the same patient. Maganini and Narendran were the first to report a left inferior parathyroid adenoma in a patient with left thyroid hemiagenesis in 1977 [(22)]. In 1992, Woods and Loury reported a left superior parathyroid adenoma in a patient with left thyroid hemiagenesis [(23)].

Sakurai et al. reported one case of an adenoma on the contralateral side of thyroid hemiagenesis in 2007. In addition, they described the absence of parathyroid glands on the side of hemiagenesis [(24)]. Mydlarz et al. published in 2010.case report of ipsilateral double parathyroid adenoma and left thyoroid hemiagenesia [(25)]. Teresa Kroeker published the case report of left lobe hemiagenesia and ipsilateral parathyroid adenoma [(26)].

In the review of the previous literatures, we found that the coincidental finding of left hemiagenesis of the thyroid gland with ipsilateral parathyroid adenoma in our current case is very rare and exceptional.

Our patient was presented with non toxic nodular goiter,the ultrasound did not mention anything regarding the absence of the left lobe or to give any hint about the presence of ipsilateral parathyroid adenoma.

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