

Is Thyroid Hemiagenesis Really Seen Seldomly?: Two Case Reports and Review of the Literature - Case Report

Hülya Ilıksu Gözü, Haluk Sargın, Mehmet Sargın, Hatice Gündüz, Mustafa Tekçe, Cem Gezen, Ali Yayla

Summary

Thyroid hemiagenesis is a rare embryological anomaly. It is diagnosed incidentally during examination for other thyroid gland diseases. Two patients with hemiagenesis of the thyroid gland were presented. Case Reports: The first patient was a 36-year-old woman with thyroid hemiagenesis presented with euthyroidism. Thyroid peroxide antibody and thyroglobulin levels were normal. Tc-99m pertechnetate scan and ultrasonography (US) findings demonstrated absence of the left lobe of the thyroid gland. A fine needle aspiration biopsy of the right lobe showed benign follicular cells. The second patient, a 31-year-old woman presented with subclinical hypothyroidism. Thyroid peroxide antibody and thyroglobulin levels were very high. Tc-99m pertechnetate scan revealed absence of the right lobe and isthmus of the thyroid gland and minimal hyperplastic left lobe. Thyroid ultrasonography (US) demonstrated a normal sized, pseudonodular left lobe of the thyroid gland with no visualization of the right lobe and isthmus. A fine needle aspiration biopsy of the left lobe showed lymphocytes. Conclusion: There are more than 270 thyroid hemiagenesis cases reported in the literature. But this may not reflect the true incidence of thyroid hemiagenesis.

Keywords: Thyroid hemiagenesis, goiter, thyroiditis

Introduction

Thyroid hemiagenesis is a rare congenital anomaly in which one lobe of the thyroid fails to develop. Agenesis may be total, unilateral or isthmic [1]. The true incidence of the thyroid hemiagenesis is estimated to be 0.02 % in normal children [2]. Although the cause of the defect is unknown, there are two proposed theories related with the etiology: a descent defect from the floor of the primitive pharynx to trachea or failure of the original anlage to become bilobed and spread out laterally to both sides [1,3]. Most of the patients remain unknown. It is usually diagnosed incidentally during examination for other thyroid gland diseases [2]. Here, we present two cases of thyroid hemiagenesis referred to our clinic within a 4 month of period.

Case Presentation

Case 1. A 36-year-old woman referred to our clinic because of asymptomatic swelling in the right side of the neck. She had goiter since her childhood, but she had no treatment from the beginning. She had history of diabetes mellites diagnosed recently and no significant family history of a thyroid disorder. Examination of neck revealed an enlarged, palpable right lobe of the thyroid gland. The left lobe of the thyroid gland was nonpalpable. Laboratory studies revealed that her serum free triiodothyronine level (F T3) was 7.09 pmol/l (normal: 2.8-7.1), free thyroxine level (F T4) was 21.01 pmol/l (normal: 12-22) and thyroid-stimulating hormone (TSH) level was 1.02 uIU/mL (0.27-4.20). The antithyroid peroxidase antibody level was 10.01 IU/mL (normal: 0-35) and antithyroglobulin level was 20 IU/mL (0-40). Tc-99 m pertechnenate thyroid scintigraphy showed that the right lobe of the thyroid gland was hyperplastic and the distribution of the activity is homogenous. The left lobe of the thyroid gland was not visualised (Fig. 1). Thyroid ultrasonography revealed enlarged isthmus (5 mm) and right lobe (72x16x20 mm), but absence of the left lobe. There are three hypoechoic nodules measured by 8 mm, 10 mm and 4mm in the right lobe of the thyroid gland. Thorax CT also revealed a hyperplastic right lobe and absence of the left lobe of the thyroid gland. There was no sign of compression to the trachea (Fig. 2). The US-guided fine needle aspiration biopsy of the nodule found in the right lobe of the thyroid gland showed benign follicular cells. Because there was no sign of compression to the trachea and biopsy revealed no malignancy, patient has been followed-up periodically without any medication. Figure 1: Tc-99m thyroid scintigraphy (case 1); showing hyperplastic right lobe in which the distribution of the activity is homogenous and the left lobe of the thyroid gland is absent. Figure 2: Thorax CT(case 1); showing a

hyperplastic right lobe and absence of the left lobe of the thyroid gland. There were no compression signs to the trachea. Case 2. A 31-year-old woman presented to our clinic because of weakness, weight gain and dyspnea for 2 months. She reported no significant family history of a thyroid disorder. Her thyroid was not palpable on physical examination. Her serum free triiodothyronine level (FT3) was 4.09 pmol/l (normal: 2.8-7.1), free thyroxine level (FT4) was 10.3 pmol/l (normal: 12-22) and thyroid-stimulating hormone (TSH) level was 4.49 uIU/mL (0.27-4.20). The antithyroid peroxidase antibody level was measured as 1000 IU/mL (normal: 0-35). Thyroid scintigraphy with Tc-99m pertechnenate demonstrated absence of the right lobe and isthmus of the thyroid gland and minimal hyperplastic left lobe (Fig. 3). Thyroid ultrasonography showed a normal sized, pseudonodular left lobe of the thyroid gland with no visualization of the right lobe and isthmus (Fig. 4). US-guided fine needle aspiration biopsy of the left lobe of the thyroid gland was performed, and cytological examination revealed lymphocytes. On the follow-up of the patient, L-T4 therapy was begun. Figure 3: Tc-99m thyroid scintigraphy (case 2); demonstrated absence of the right lobe and isthmus of the thyroid gland and minimal hyperplastic left lobe Figure 4: Thyroid US (case 2); showed a normal sized, pseudonodular left lobe of the thyroid gland with no visualization of the right lobe and isthmus.



Figure 1. Tc-99m thyroid scintigraphy (case 1); showing hyperplastic right lobe in which the distribution of the activity is homogenous and the left lobe of the thyroid gland is absent.

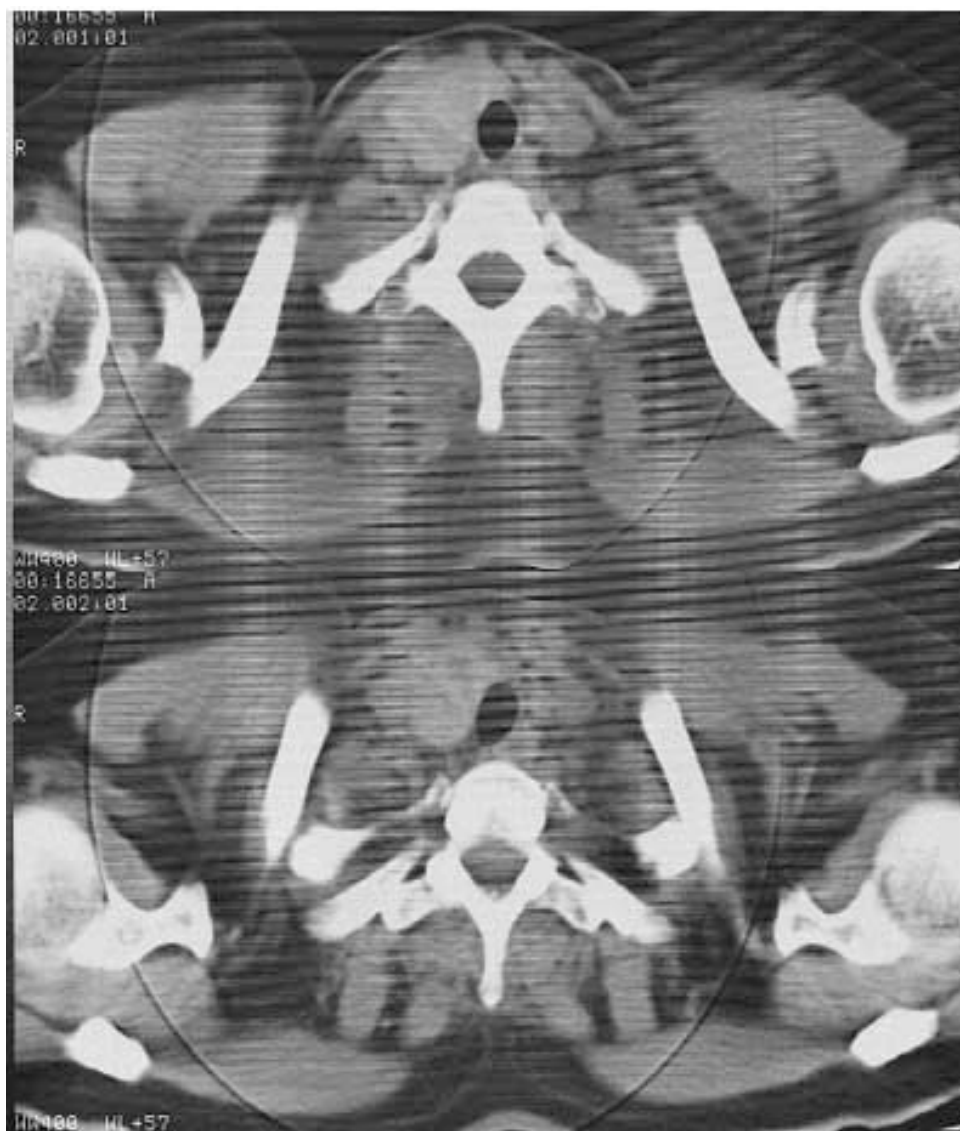


Figure 2. Thorax CT(case 1); showing a hyperplastic right lobe and absence of the left lobe of the thyroid gland. There were no compression signs to the trachea.



Figure 3. Tc-99m thyroid scintigraphy (case 2); demonstrated absence of the right lobe and isthmus of the thyroid gland and minimal hyperplastic left lobe

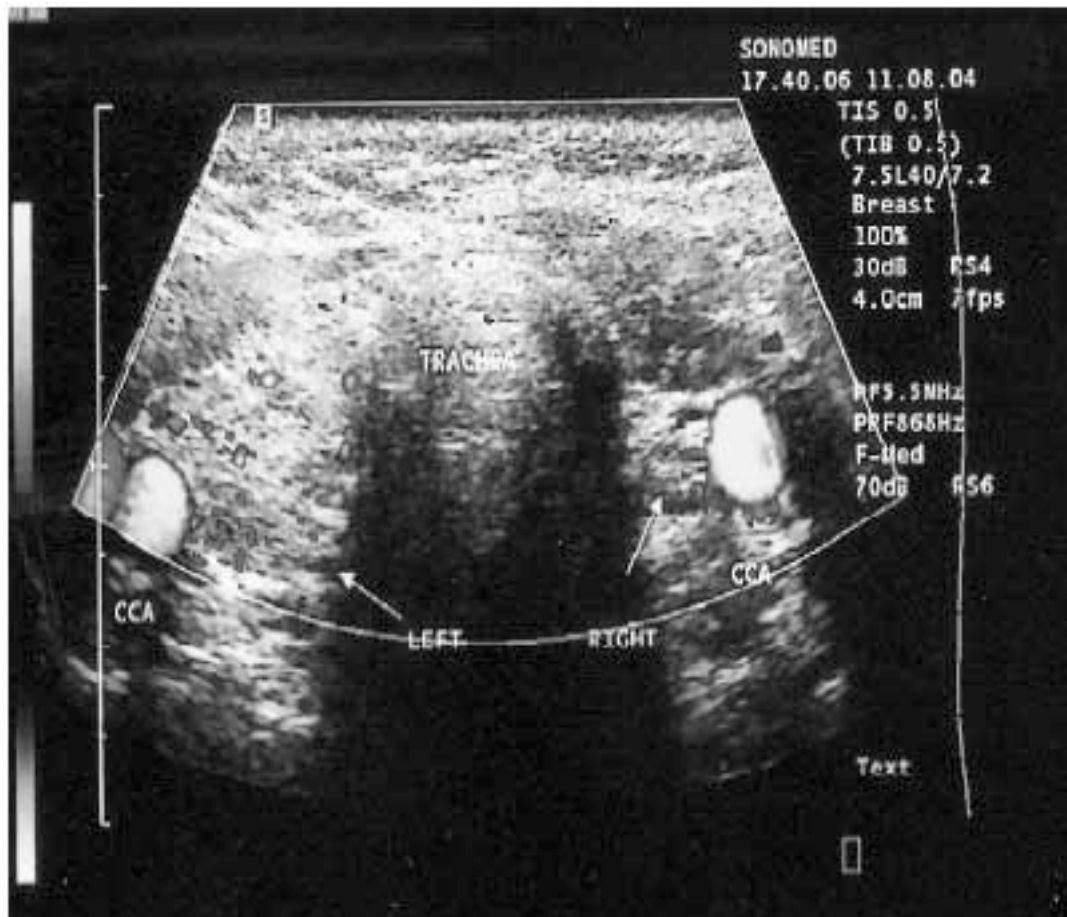


Figure 4. Thyroid US (case 2); showed a normal sized, pseudonodular left lobe of the thyroid gland with no visualization of the right lobe and isthmus

Discussion

Because it is usually diagnosed incidentally during examination for other thyroid gland disease, the true incidence of thyroid hemiagenesis is uncertain. In the literature two group of the studies have been documented. Majority of them have been documented as case reports. The others being a small part described on the basis of scale postmortem and imaging studies. Thyroid hemiagenesis was first described by Marshall in 1895. Marshall found one case in 60 autopsies of children [4]. The discovery rate of thyroid hemiagenesis by scanning has been reported by Andreew to be 7 cases in 708 scan and by Hamburger and Hamburger to be 4 in 7000 patients [5,6]. In 1972 a large study was conducted in which 12456 patients who underwent surgery were investigated and seven thyroid hemiagenesis were detected [7]. In 1979, Friedman et al. found six cases with thyroid hemiagenesis in 12000 scans [8]. After these large scale studies, the

studies reported as case history have been declared between 1979-1981. Melnick and Stemkowski reported four patients with thyroid hemiagenesis and described the hockey stick sign by imaging studies in patients with thyroid hemiagenesis. They also reviewed the literature, which revealed a total 94 cases of thyroid hemiagenesis in 1981 [9]. Thirteen more reported cases with thyroid hemiagenesis were added till 1986 [10,11,12,13,14,15,16]. Thus bringing the total to just over 100. Between 1986-1999 the studies designed as case reports have been documented. Bandao et.al described a case of thyroid hemiagenesis with Graves hyperthyroidism developed from hypo-thyroidism and revealed a total of 196 cases of thyroid hemiagenesis from the literature in 1999 [17]. In 2003, a thyroid hemiagenesis with multinodular goiter was reported from Turkey and their review of the literature revealed a total number of 270 cases with thyroid hemiagenesis [2]. After this study, five more case reports with thyroid hemiagenesis have been documented in the literature [3,18,19,20,21], thus bringing the total to over 270. We present two more cases with thyroid hemiagenesis. Thyroid hemiagenesis is associated with a female predominance of 3:1 [1,2,8,19,21-26]. The subjects ranged in age from infancy to 85 years [8]. In our study both of the patients were female consistent with the literature. Agenesis may be total, unilateral or isthmic [22]. It primarily involves the left lobe [1,2,19,22-27]. The absence of the left lobe and right lobe is encountered in 80% and in 20% of patients. The isthmus is absent in 50% of cases [25]. In this study, the left lobe was absent in the first patient. The right lobe and isthmus were absent in the second patient. Any type of thyroid disorders might have been found in conjunction with thyroid hemiagenesis; including carcinoma [1,22,28-30], thyrotoxicosis [10,11,17-19,29,31-37], goiter [2,9,22,26,29,38], thyroiditis [17,21,23,39] or thyroglossal duct cyst [25]. Diagnosis was established as goiter in the first patient and lymphocytic thyroiditis in the second patient in this study. Although the etiology of thyroid hemiagenesis is not known, one theory seems to be a genetic one [2]. Several target genes, including thyroid transcription factors (TTF-1 and TTF-2), PAX-8 and sonic hedgehog have been found to be involved in thyroid morphogenesis [2,40] Thyroid hemiagenesis is mostly asymptomatic. It does not manifest itself in clinical symptoms and the diagnosis is often established incidentally during evaluation of other thyroid disorders [41]. Our review of the literature brings the total number of thyroid hemiagenesis to over 270. But it is very likely that the total number of reported cases in the literature reflect a marked underestimation of true incidence of thyroid hemiagenesis since diagnosis was made coincidentally in most of the cases [42]. So a diagnosis of thyroid

hemigenesis should always be considered when unilateral thyroid enlargement is encountered. We also need a group of large studies described on the basis of scale postmortem and imaging techniques. So, under this circumstances, can we ask the question "Is Thyroid Hemigenesis Really Seen Seldomly?"

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