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Thyroid hemiagenesis: a report of three cases and review of the literature

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Thyroid hemiagenesis resulting from the failure of embryologic development of one thyroidal lobe is a very rare anomaly. It is usually incidentally discovered during the investigation of accompanying thyroid disorders. Here we report three cases with right lobe agenesis in two patients and left lobe agenesis in one patient. Two of them were hyperthyroid, while the other euthyroid patient had a thyroid mass.

Key words: thyroid hemiagenesis, multinodular goiter, hyperthyroidism

INTRODUCTION

THYROID HEMIAGENESIS is a rare congenital abnormality, in which one of the thyroidal lobes fails to develop. In the literature, most of the patients reported to have thyroid hemiagenesis had accompanying thyroid disorders because the absence of one thyroid lobe usually does not cause clinical symptoms by itself. Therefore, the true prevalence of thyroid hemiagenesis is difficult to determine. However, a recently reported study on normal school children showed a 0.05% prevalence.[1] Thyroidal hemiagenesis is predominantly seen in females with the left lobe being absent.[2–5] We report here three cases of thyroid hemiagenesis involving the right lobe in two patients and the left lobe in one patient. Our purpose is to describe these patients who were diagnosed incidentally on thyroid scintigrams and to briefly review the relevant literature.

CASE REPORTS

Case 1

A 59-year-old man with a 3-year history of hyperthyroidism was referred to our department for I-131 therapy. He had been taking propylthiouracil and a β-blocker for 3 years. On physical examination, blood pressure was 140/80 mmHg and pulse rate 102/min and regular. The patient appeared somewhat anxious, his skin was warm and moist, and there was a fine tremor of the hands. There was no lid lag or proptosis. Total T_3 was 2.52 ng/m*l* (normal range: 0.8–2), total T_4; 8.87 μg/d*l* (normal range: 4.6–12), free T_3; 6.31 pg/m*l* (normal range: 1.8–4.6), free T_4; 1.33 ng/d*l* (normal range: 0.93–1.7), TSH; 0.005 μIU/m*l* (normal range: 0.27–4.2). As shown in Figure 1A, a technetium image of the thyroid gland revealed no radioactivity in the left lobe or isthmus. There was an enlarged right lobe with hyperactive nodules. Ultrasonography confirmed agenesis of the left lobe and isthmus (Fig. 1B). There were multiple nodules in the right lobe. The dominant nodule, measuring 46 × 31 mm, was a solid nodule in the lower pole of right lobe.

Case 2

This 47-year-old man had toxic multinodular goiter. He was referred to our department for thyroid scan and ultrasonography. He presented with a long-standing, painless, palpable left thyroid mass. His blood pressure was 160/100 mmHg and pulse rate 96/min and regular. TSH was 1.1 μIU/m*l* (normal range: 0.28–4), free T_3; 3.2 pg/m*l* (normal range: 1.8–4.6), free T_4; 1.3 ng/d*l* (normal range: 0.9–1.7). A technetium image revealed an enlarged left lobe with a hyperactive nodule in the mid-zone but no visible radioactivity where the right lobe is normally located (Fig. 2A). Ultrasonography confirmed the agenesis of right lobe and isthmus (Fig. 2B). USG also showed two solid nodules in the left lobe measuring 27 × 17 mm and 12 × 10 mm.

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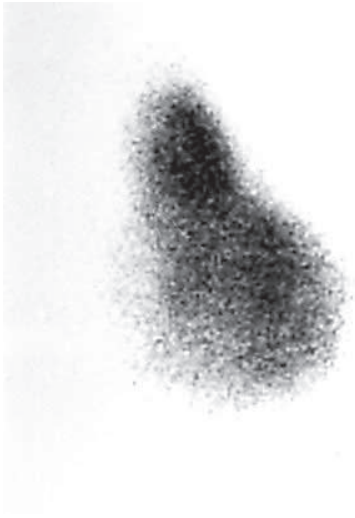


A

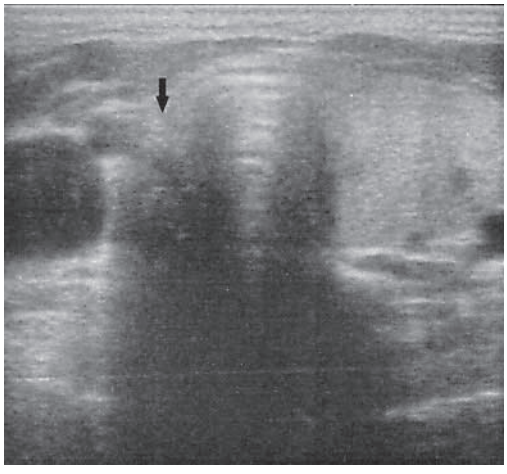
B

Fig. 1 Case 1; (A) Thyroid scintigraphy reveals absence of the left lobe and isthmus. There is an enlarged right lobe with hyperactive nodules. (B) Ultrasonography in transverse section confirms the nonvisualization of the thyroid tissue on the left side (*arrow*) and isthmus.

Case 3

The patient was a 41-year-old woman who complained of a mass in the left side of her neck. She was clinically euthyroid. A thyroid scan, performed with Tc-99m pertechnetate, showed absence of radiotracer accumulation in the region of the right lobe and isthmus and a hypoactive nodule in the upper pole of the left lobe (Fig. 3). Thyroid ultrasonography confirmed the absence of the right lobe and isthmus. There was a large left lobe with multiple nodules. The dominant nodule was a degenerated cystic nodule in the upper lobe of the left lobe (23 × 15 mm).

A



B

Fig. 2 Case 2; (A) Thyroid scan shows absent right lobe and isthmus. A hyperactive nodule is seen in the mid-zone of the left lobe. (B) Thyroid ultrasonography in transverse section: no thyroid tissue is present on the right lobe (*arrow*) or isthmus.

DISCUSSION

Thyroid hemiagenesis is a rare congenital anomaly with absence of a lobe or a lobe and the isthmus. Embryologically, the thyroid gland develops in the midline from the pharyngeal floor at the foramen cecum.[6] The thyroid is then displaced inferiorly until it comes to its final location anterior to the trachea by the seventh week.[6] Failure of descent of the thyroid results in functioning thyroid at any point from the foramen cecum at the base of the tongue to low in the neck. The cause of hemiagenesis is not clear. It is thought to result from failure of the cells to migrate laterally resulting in agenesis of a part of the thyroid.[7] It is unknown whether disturbance of the lobulation process is due to the interference of environmental factors or to some genetic abnormality. A genetic one is suggested by

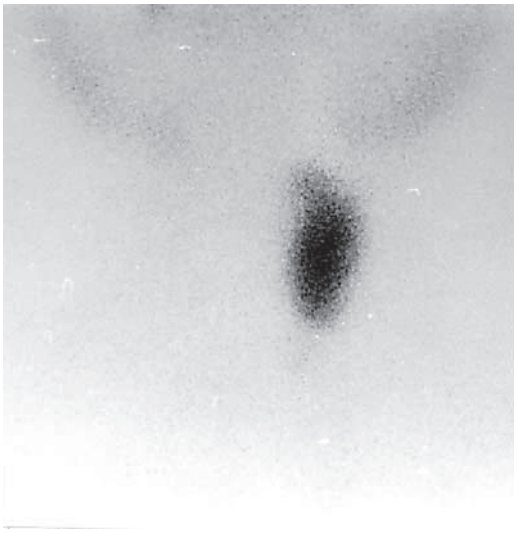


Fig. 3 Case 3; Absence of technetium-99m pertechnetate uptake in the right lobe. A hypoactive nodule is seen in the upper pole of the left lobe.

the occurrence of thyroid hemiagenesis among monozygotic twins,[8] among sisters,[9] or together with other thyroid malformations within one family.[10,11] Several genes have been found to control thyroid descent, development and morphogenesis. Three thyroid transcription factors, TTF1, TTF2, and Pax-8 are reported to be candidates in thyroid development defects but these genes have not been investigated in thyroid hemiagenesis.[12,13]

The total number of reported cases in the literature probably underestimates the true incidence of thyroid hemiagenesis because it is usually incidentally discovered during the investigation of accompanying thyroid disease. Thyroid disease in the remaining thyroid lobe includes multinodular goiter,[14] hyperthyroidism,[15] hypothyroidism,[3] adenoma,[16] adenocarcinoma,[17] chronic thyroiditis and subacute thyroiditis.[18] The most common disease of the remaining lobe was hyperthyroidism.[14,17] Some patients were found to be in euthyroid state without any abnormalities.[3] All our three cases had multinodular goiters. Two of them had a history of hyperthyroidism (cases 1 and 2) and the other one (case 3) was euthyroid. The first case of thyroidal hemiagenesis was described in 1866,[1,16] since which approximately 285 cases have been reported in the literature.[19] Due to the fact that reported cases are still few in number and thyroid hemiagenesis is usually diagnosed coincidentally, the real incidence in the general population is unknown and its determination is difficult. The diagnosis of thyroid hemiagenesis by imaging has been reported to be 4 in 7,000 scans in one study.[20] However, it is probably rarer because these patients underwent clinically indicated scanning. Mikosch et al.[5] evaluated 71,500 patients who underwent thyroid investigations during a period of 9 years. Sixteen patients with thyroid hemiagenesis were seen during this period. Its prevalence was estimated at between 1 : 1900 and 1 : 2675. Shabana et al.[21] evaluated 2,845 normal school children in a systematic ultrasound study of thyroid gland volume for the evaluation of iodine deficiency and found left lobe agenesis in 4 girls and 2 boys. Their study showed that the estimated prevalence of thyroid hemiagenesis is 0.02%. Maiorana et al.[1] recently studied thyroid hemiagenesis prevalence by neck ultrasound examination in 24,032 unselected 11 to 14-yr-old children. In this study, 12 cases of left lobe agenesis were identified, with a prevalence of 0.05%.

The prevalence of this developmental defect is greater in females (75% versus 25%) with a left lobe being absent.[2–5] Absence of the left lobe occurs in 80% of cases and agenesis of the isthmus occurs in 50% of cases. Two of our patients (case 2 and case 3) had both right lobe and isthmus agenesis which is much rarer than left lobe agenesis.

Thyroid hemiagenesis is commonly diagnosed on thyroid scintigraphy but there are some conditions which can be confused with it. Hyperfunctioning nodule with marked suppression of all other thyroid tissue, neoplasms, inflammations and infiltrative diseases such as amyloidosis can mimic thyroid hemiagenesis.[2,14,22,23] For this reason, it is reasonable to make a confirmation of thyroid scans with ultrasound, CT or MRI. Tc-99m methoxyisobutyl isonitrile (MIBI) scintigraphy is also an alternative and practical way to show the suppressed thyroid tissue.[24,25]

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