Rare disease

Left thyroid hemiagenesis in a patient with primary hyperparathyroidism

Siddek Isreb,¹ Faisal Alem,² David Smith²

¹ Department of General Surgery, Northumbria Health Care, North Shields, UK

² Department of General Surgery, Ninewells Hospital and Medical School, Dundee, UK

Correspondence to Siddek Isreb, drisreb@yahoo.com

Summary

The case of a patient with primary hyperparathyroidism with the incidental finding of left thyroid hemiagenesis with isthmus involvement is presented. Thyroid hemiagenesis is a rare congenital abnormality. There have been approximately 310 cases reported in the literature to date. It has been reported as an incidental finding with a wide range of associated pathological conditions. It is important to consider this in patients being prepared for thyroid lobectomy due to the inevitability of postoperative hypothyroidism. Due to the female predominance of thyroid disease, hemiagenesis was first thought to be more common in women. Left lobe cases accounts for the majority of instances and isthmus is absent in half the cases. Ultrasonography is the diagnostic tool of choice.

BACKGROUND

Thyroid hemiagenesis was first reported by Handfield-Jones in 1866.¹ There have been approximately 310 cases reported in the literature to date.² It is usually found incidentally during other thyroid or parathyroid abnormality investigations. Due to the female predominance of thyroid disease, hemiagenesis was first thought to be more common in women. Left lobe cases account for the majority of instances and isthmus is absent in half the cases.

CASE PRESENTATION

A 75-year-old patient presented with a history of several years of progressive depression, tiredness, thirst, constipation, poor appetite and recurrent urinary tract infections. She denied any history of fractures or renal calculi. She had had an episode of renal colicky pain treated conservatively. Investigation revealed a corrected calcium level of 2.94 mmol/litre, with an elevated parathormone (PTH) level of 7.6 pmol/L (normal 1–6.9). Her thyrotrophin level was normal.

INVESTIGATIONS

Abdominal ultrasound ruled out renal calculi. A dual energy x-ray absorptiometry (DEXA) scan showed osteopaenia. A parathyroid sestamibi scan showed a solitary focus of abnormal sestamibi uptake consistent with the inferior left parathyroid gland position with absence of the left thyroid lobe and the isthmus (figure 1). The patient had had no previous thyroid surgery.

OUTCOME AND FOLLOW-UP

Primary hyperparathyroidism was suspected and the patient was referred for surgery. The neck was explored through a standard thyroid incision. Operative findings confirmed the absence of the left thyroid lobe and the isthmus. The two left parathyroid glands were adjacent to each other with an adenoma of the inferior gland, which was excised.

Pathology confirmed parathyroid adenoma with no evidence of malignancy. Postoperative calcium and PTH levels were normal and the patient was discharged the following day.

DISCUSSION

Thyroid hemiagenesis is a rare congenital abnormality. It is difficult to find a precise prevalence rate, but it is estimated to be between 1 in 1900 and 1 in 2675.³ A study from Belgium on 2845 normal schoolchildren established a prevalence of 0.2%.⁴ A large prospective study on 24 032 unselected 11–14-year-old schoolchildren from Southeastern Sicily reported a prevalence of 0.05%.⁵

Previous studies based on symptomatic patients and the Belgian study reported a female predominance of 3:1.^{3 5–7} This may be due to the fact that thyroid diseases are more frequent in women than in men. A large screening study on normal children showed slight male predominance (1.4:1) although the difference between men and women was not statistically significant.⁵

The left lobe is most commonly affected and accounts for 80% of cases.^{6–8} Association with isthmus agenesis was found in 50% of cases.⁸ The thyroid gland develops from invagination of the endoderm in the primitive pharynx between the first and second pharyngeal pouches immediately dorsal to the aortic sac. This duct shaped invagination takes place at 16–17 days of gestation. Although it expands ventrally with the most rapid proliferation at its distal tip, it remains attached by a stalk (thyroglossal duct) to the pharyngeal floor. It begins to expand laterally at the end of the second month, forming the characteristic bilobar structure.⁹ Environmental or genetic factors could affect this bilobar formation and hence result in hemiagenesis. The reported familial occurrence of thyroid hemiagenesis, the occurrence among monozygotic twins and the association with other familial thyroid malformation support a genetic predisposition.^{10–12} A higher prevalence in areas of iodine deficiency reported by Maiorana *et al*⁶ was of no statistical

Figure 1 Sestamibi scan results.

significance. A higher prevalence of hemiagenesis (10.7%) in early childhood subclinical hypothyroidism was also reported.¹³ Maiorana *et al*⁶ also reported a subtle thyroid function abnormality. As a result they advise a follow-up for possible development of hypothyroidism. Further studies are needed to confirm such recommendations.⁵

Thyroid hemiagenesis has been reported as an incidental finding with a wide range of associated pathological conditions, including hypothyroidism,¹³ multinodular goiter,¹⁴ Graves' disease,¹⁵ thyroiditis,¹⁶ papillary carcinoma,¹⁷ ectopic thyroid¹⁸ and primary hyperparathyroidism,¹⁹ as in our case.

Ultrasonography is the diagnostic tool of choice for detecting thyroid hemiagenesis.³ Differential diagnoses of absent lobe images on thyroid scintiscan include solitary toxic adenoma, primary or secondary neoplasms, infiltrative diseases, unilateral inflammations and true hemiagenesis. As a result, ultrasonography is not considered the best imaging modality for hemiagenesis.¹¹ In our case surgery was already indicated and hemiagenesis confirmed at the time of operation.

Learning points

- ▶ Thyroid hemiagenesis is a rare congenital abnormality.
- ▶ The left lobe is most commonly affected.
- ▶ Ultrasonography is the diagnostic tool of choice.
- ▶ Lobectomy, for possible cancer, becomes total thyroidectomy in such cases.
- ▶ Thyroid hemiagenesis prepared for thyroid lobectomy should be warned about the inevitable postoperative hypothyroidism.

Competing interests None.

Patient consent Obtained.

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