Hemiagenesis of the thyroid gland

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Abstract

Thyroid hemiagenesis is a rare developmental anomaly. This congenital abnormality is usually discovered upon investigating thyroid disorders primarily through thyroid imaging studies. Coupled with its sensitivity, ultrasonography remains as the non-invasive investigation of choice in establishing the diagnosis. A thyroid-stimulating hormone test may similarly provide confirmatory evidence of an absent thyroid lobe which may occasionally involve the isthmus of the gland as well. Surgical exploration performed in relation to a pathological entity affecting the remaining thyroid gland as illustrated in our patient is another means of establishing the diagnosis. Various other abnormalities have been described in relation to thyroid hemiagenesis. We report a case of hemiagenesis of the thyroid gland in a young female who presented with a benign nodule affecting the thyroid isthmus. We believe this to be the first reported case of thyroid hemiagenesis in Malaysia and this congenital anomaly is discussed with a brief review of the literature.

Key words: thyroid, hemiagenesis, agenesis, congenital

Introduction

Developmental abnormalities of the thyroid gland are rare. The first documented case of thyroid hemiagenesis was reported in 1866 in Europe (Melnick & Stemkowski, 1981). Melnick & Stemkowski in 1981 reported 4 cases out of 8,000 patients over a 15-year period, while Hamburger & Hamburger (1970) found only 4 cases of 7,000 patients who underwent thyroid imaging. Even though the number of reported cases of thyroid hemiagenesis has steadily increased over the last four decades, a detailed literature search failed to reveal any reported cases of thyroid hemiagenesis in Malaysia. We present this rare abnormality in a young female patient who presented with a thyroid nodule and discuss this entity based on a review of the literature on thyroid hemiagenesis.

Case report

A 25 year-old female patient presented to the outpatients department complaining of a 6 months history of a painless midline neck swelling. She had neither compressive symptoms nor symptoms suggestive of a hyper or hypothyroidism and her primary concern related to the cosmetic appearance of the swelling and the need to rule out a sinister pathology. Her past medical history was unremarkable. Examination confirmed a fit patient who was clinically euthyroid with the presence of a solitary thyroid nodule palpable affecting the isthmus of the thyroid gland measuring 3 cm in diameter. The rest of the examination was unremarkable.

Thyroid function test was normal and a fine needle aspirate of the lump demonstrated benign features compatible with a colloid nodule. The patient was reassured and following a discussion of the options available, she opted for surgical intervention primarily due to the cosmetic appearance of the swelling. On surgical exploration, a solitary nodule was evident affecting the thyroid isthmus. Exploration of both thyroid lobes lobe revealed absence of the left-sided gland and a normal looking right thyroid lobe. The nodule together with the thyroid isthmus was excised leaving the normal looking right-sided thyroid gland intact.

She made an uncomplicated postoperative recovery and histological analysis of the excised lump confirmed features of a benign colloid goitre. An ultrasound scan postoperatively confirmed an absent left thyroid lobe. She remained clinically euthyroid following her surgery with a normal thyroid function test.

Discussion

Hemiagenesis of the thyroid gland is a rare anomaly in which one of the thyroid lobes fails to develop. The thyroid is the earliest glandular structure to appear in the embryo. It develops from an ectodermal outpouching at

the ventral surface of the foregut. As the developing thyroid sac "descends" and becomes solid, it remains connected to the pharyngeal floor via the thyroglossal duct. This duct becomes a solid stalk that usually disintegrates by the sixth embryologic week. The gland is bi-lobed at an early stage in development. This bi-lobed structure normally spreads out laterally at the inferior extent of the gland to develop into a normal thyroid gland (Skandalakis & Grey, 1994).

A number of theories have been put forward in relation to the development of thyroid gland hemiagenesis. It has been postulated that failure of the cells to either migrate laterally or failure of the median anlage or lateral anlage of the embryologic thyroid to develop leads to this anomaly (Bough *et al.*, 1994; Skandalakis & Grey, 1994). The two lobes of the thyroid are not always symmetrical, a feature that is evident upon surgical exploration or on findings during an autopsy. The asymmetry does not necessarily denote pathology even though further investigations may be warranted upon discovery of this entity especially in the paediatric age group. In a study in the last century of the morphology of the thyroid in children, 7% of the cadavers were found to have one lobe larger than the other, and in 7% the isthmus was entirely absent. (Skandalakis & Grey, 1994). A few authors have even suggested that thyroid hemiagenesis is simply an extreme degree of asymmetry commonly seen in all bilateral symmetric organs rather than actual absence of a gland (Mortimer *et al.*, 1981; Hamburger & Hamburger, 1970).

The prevalence of hemiagenesis is not known. Since the initial discovery of the abnormality in 1866, only 18 further cases were reported over the next 50 years (Melnick & Stemkowski, 1981). A recent population study in Austria by Mikosch *et al.* (1999), in which 71,500 people were screened over a period of 9 years, found 16 patients with the condition as compared with 4 reported cases each by Melnick & Stemkowski (1981) and Hamburger & Hamburger (1970), of 8000 and 7000 subjects screened respectively. It must however be stressed that the Austrian study (Mikosch *et al.*, 1999) which suggests a lower prevalence of thyroid hemiagenesis was performed on a normal population with no thyroid diseases while the latter 2 studies were conducted on patients who already had underlying thyroid disorders.

There is greater female sex preponderance amongst patients with thyroid hemiagenesis (3:1 ratio) and the condition affects the left lobe more commonly (80% of cases) than the right lobe. Concomitant absence of the isthmus may be obvious in 50% of cases (Hamburger & Hamburger, 1970). Since hemiagenesis of the thyroid is a clinically silent entity, it is usually thyroid disorders that manifests in the remaining functioning lobe that draw the attention of the clinician to this developmental anomaly. The patient more commonly than not are euthyroid and thyroid function tests need to be supplemented by further investigations such as thyroid imaging studies to confirm the diagnosis. A significant number of cases are diagnosed during surgical exploration, such as in our patient, as dictated by the clinical presentation and preoperative diagnosis (Russotto & Boyar, 1971; Tashima *et al.*, 1973).

The diagnosis of thyroid hemiagenesis may be entertained by the clinician in any patient who presents, on examination no palpable thyroid tissue on one side of the neck. Together with the finding on physical examination, thyroid imaging and not uncommonly a thyroid stimulating hormone (TSH) study coupled with a thyroid scan should fail to demonstrate thyroid tissue on the affected side (Melnick & Stemkowski, 1981). Other than the methods stated above, other modalities that can be used to diagnose thyroid hemiagenesis include computed tomography (CT), magnetic resonance imaging (MRI) and a TSH test. A TSH test may be a useful supplement to imaging when surgical exploration has not been performed in confirming the diagnosis. A TSH stimulation test will in cases of congenital hemiagenesis show total absence of uptake in an entire lobe in contrast to other pathological entities, which may give rise to a reduction in functional activity. This investigation is also helpful in ruling out the presence of an ectopic gland (Bough *et al.*, 1994). Ultrasonography, even though operator dependant, remains as the non-invasive investigation of choice. Other investigations such as fine needle aspiration cytology and scintigraphic studies are useful in diagnosing other diseases within the remaining gland (Mikosch *et al.*, 1999).

Thyroid hemiagenesis is usually discovered in conjunction with an associated pathology in the thyroid gland including hyperthyroidism, multinodular goiter, benign adenoma, primary and secondary hypothyroidism, chronic thyroiditis, hyperparathyroidism and thyroid cancer (Harada *et al.*, 1972; Melnick & Stemkowski, 1981; Piera *et al.*, 1986). Other conditions that can mimic thyroid hemiagenesis include unilateral inflammatory disease of the thyroid gland, autonomously functioning nodule with suppression of extra nodular tissue, primary neoplasia, secondary deposits from neoplasms elsewhere in the body, or infiltrative diseases, such as amyloidosis (Melnick & Stemkowski, 1981). These conditions can produce a similar feature on isotope scanning of the thyroid by rendering the affected gland non-functional even though the tissue on the side with absent function can always be palpated (Hamburger & Hamburger, 1970). Absence of the gland secondary to surgery would be readily evident from the history together with the presence of a surgical scar.

There has been an increase in the number of reported cases of thyroid gland hemiagenesis. A comprehensive literature review confirmed the 130 reported (Duh *et al.*, 1994) cases of thyroid hemiagenesis in 1994, compared to

94 cases reported in 1980 (Melnick & Stemkowski. 1981). Our review of the literature to date increases the number of cases reported to a total of 275 including our patient. The increase in detecting this anomaly is most likely due to the increased utilisation and improvement in imaging studies in investigating the thyroid gland.

Hemiagenesis of the thyroid gland by itself does not require surgical treatment unless it is complicated by concurrent thyroid disease. In a rare variant of the condition in which there is complete agenesis of the gland, hypothyroidism is inevitable.

References

Bough D, Greco T, Romanczuk B. Imaging case study of the month: thyroid hemiagenesis. *Ann Otol Rhinol Laryngol* 1994; 103: 328-330.

Duh QY, Ciulla TA, Clark OH. Primary parathyroid hyperplasia associated with thyroid hemiagenesis and agenesis of the isthmus. *Surgery* 1994; 115: 257-63.

Hamburger J, Hamburger S. Thyroid hemiagenesis. *Arch Surg* 1970; 100: 319-20.

Harada T, Nishikawa Y, Ito K. Aplasia of one thyroid lobe. *Am J Surg* 1972; 124: 617-9.

Melnick J, Stemkowski P. Thyroid hemiagenesis (hockey stick sign) a review of the world literature and a report of four cases. *J Clin Endocrinol Metab* 1981; 52: 247-51.

Mikosch P, Gallowitsch HJ, Kresnik E, Molnar M, Gomez I, Lind P. Thyroid hemiagenesis in an endemic goiter area diagnosed by ultrasonography: report of sixteen patients. *Thyroid* 1999; 9 (11): 1075-84.

Mortimer PS, Tomlinson IW, Rosenthal FD. Hemiaplasia of the thyroid with thyrotoxicosis. *J Clin Endocrinol Metab* 1981; 52: 152-5.

Piera J, Garriga J, Calabuig R, Bargallo D. Thyroidal hemiagenesis. *Am J Surg* 1986; 151: 419-21.

Russotto JA, Boyar RM Thyroid hemiagenesis. *J Nucl. Med* 1971; 12: 186-7.

Skandalakis JE, Gray SW. *Embryology for Surgeons 2nd Ed.* 1994; Williams and Wilkins page 50.

Tashima CK, Lee WY, Leong A. Agenesis of the thyroid. *JAMA* 1973; 224: 1761-2.