Thyroid Isthmus Agenesis: A Rare Phenomenon

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ABSTRACT: The thyroid gland is composed of two lobes connected by an isthmus that traverses the midline of the upper trachea between the second and third tracheal rings. The most common manifestation of dysgenesis of the thyroid gland is hemi agenesis or ectopic thyroid gland tissue. Specifically, the incidence rate of agenesis of the thyroid isthmus ranges between 0.5 and 10%. Remember that the identification of agenesis of the isthmus and other thyroid anomalies during preoperative evaluation of patients scheduled for thyroid surgery would significantly contribute to safer surgical procedures and fewer complications. We present a patient with an enlarged right thyroid nodule on USG who was scheduled for a right hemithyroidectomy but was ultimately diagnosed intraoperatively with thyroid isthmus agenesis.

KEYWORDS: Agenesis, Dysgenesis, Thyroid Gland.

INTRODUCTION

The thyroid gland is divided into two lobes that are connected by the isthmus, which crosses the midline of the upper trachea at the second and third tracheal rings. There are a large number of morphological variations of the thyroid gland. The position of the thyroid gland and its close relationship with various structures brings about several surgical considerations with clinical relevance. Dysgenesis of the thyroid isthmus is however, poorly reported with an incidence rate ranging between 0.5 and 10% in literature (1).

CASE SUMMARY

A 34-year-old female reported to the surgical outpatient department with chief complaints of neck swelling that moved with deglutition but not on tongue protrusion. She recalled discovering this swelling inadvertently two years ago; it was painless and gradually grew to double its original size. She does not have a history of diabetes or hypertension and is not currently on any drugs. Her scan revealed nodules on her right thyroid lobe, the largest measuring 3 centimetres in diameter. Her standard blood tests, which included a thyroid panel, indicated that she was euthyroid. There are no clinical manifestations of hyperthyroidism or hypothyroidism. She was planned to undergo a right hemithyroidectomy; however, during surgery, an isolated right thyroid lobe without connection to the left enlarged thyroid nodule was observed, demonstrating thyroid isthmus agenesis. The nodule's histopathology confirmed the preoperative fine needle aspiration cytology (FNAC) diagnosis of colloid goitre.
DISCUSSION
Normal development of the thyroid gland begins with a median thickening of endoderm on the floor of the pharynx between the first and second pharyngeal pouches. During the 4th week of gestation, invagination of this region leads to the formation of the median diverticulum. The thyroid diverticulum which forms into the thyroglossal duct, bifurcates to give rise to the thyroid lobes and the isthmus. The cephalic end of the thyroglossal duct degenerates during this process. (2) Developmental abnormalities of the thyroid can be divided into three major categories: A. agenesis of thyroid gland; B. dysgenesis of the thyroid; C. abnormalities due to persistence of the thyroglossal duct. Dysgenesis of the thyroid gland most commonly presents with hemi agenesis or ectopic thyroid gland tissue. In particular the agenesis of the thyroid isthmus has an incidence rate ranging between 0.5 and 10%. (3) Because a few cases have been reported, data regarding the incidence of thyroid agenesis are mainly based on cadaver series. Dixit et al. reported the rate of isthmus agenesis to be 14.6% in a series of 41 cadavers, whereas Ranade et al. reported a rate of 33% in a series of 105 cadavers. Therefore, with such a limited number of cadaver series available in the literature, the data on the incidence of isthmus agenesis is considered to be insufficient. The aetiology of isthmus agenesis is also not completely known. It has been reported that it may be related to genetic factors, developmental anomalies, and mutations of chromosome 22 and thyroid transcription factor (TITF) 1–2 genes. (4) Clinically, scintigraphy with an overload of thyroid stimulating hormone (TSH) can be used to diagnose agenesis of the isthmus. It can also be diagnosed using ultrasonography (USG), computed tomography (CT), and magnetic resonance imaging (MRI) or during thyroid surgery (5). In this case report, agenesis of the isthmus was determined during thyroid surgery. In our patient, the reason for not determining agenesis of the isthmus by preoperative neck USG may be because of the expansion of the thyroid lobe into the midline by the large nodule in the right lobe. Patients with isthmus agenesis are generally euthyroid but hypothyroidism or hyperthyroidism may be seen (6). In our case, thyroid function was normal. Agenesis of isthmus can be associated with, absence of a lobe or the presence of ectopic thyroid tissue and hence in clinical practice when such a condition is diagnosed, it is necessary to perform a differential diagnosis against other pathologies such as autonomous thyroid nodule, thyroiditis, and so on. While planning for thyroidectomy one should be prepared to find variations like ectopic thyroid nodules around the normally-located thyroid gland and also has to be precise in dissection as important nerves and vessels lies in the vicinity of thyroid gland

CONCLUSION
Agenesis of the thyroid gland is rare. When agenesis of the isthmus is determined, its association with agenesis of the thyroid lobe or the presence of ectopic thyroid tissue must be kept in mind. In addition, diseases such as autoimmune thyroid nodule, thyroiditis, primary thyroid carcinoma, metastasis, and amyloidosis must be considered in the differential diagnosis. It should be remembered
that the determination of agenesis of the isthmus and other thyroid anomalies during preoperative assessment in the patients for whom thyroid surgery is planned would contribute significantly to safer surgical procedures and fewer surgery-related complications.

REFERENCES