Agenesis of the isthmus of the thyroid gland

The thyroid is an endocrine gland composed of two lobes connected by the isthmus tissue. Thyroid isthmus agenesis is a rare condition, and only a few cases have been reported in the literature. Here, we discuss the case of a 56-year-old female patient in whom agenesis of the thyroid isthmus was discovered incidentally during surgery for a multinodular goitre. When agenesis of the isthmus is found, associated thyroid lobe agenesis and the presence of ectopic thyroid tissue must be considered. In addition, associated autoimmune thyroid nodule, thyroiditis, primary thyroid carcinoma, metastasis, and amyloidosis must be considered in the differential diagnosis. Preoperative awareness of potential agenesis of the isthmus and its associated thyroid anomalies in patients with planned thyroid surgery will significantly contribute to safety during surgical procedures and result in fewer surgery-related complications.

**Keywords:** Agenesis, isthmus, thyroid

INTRODUCTION

The thyroid gland is an endocrine gland composed of two lobes connected by the isthmus tissue (1). The isthmus is localized at the level of the second and third tracheal rings (2). A wide range of morphological variations of the isthmus, such as hypoplasia, ectopy, hemiagenesis, and agenesis as well as developmental anomalies have been described in the literature. Thyroid isthmus agenesis is rare, and only a few cases have been identified (3). Here, we discuss the case of a patient in whom agenesis of the thyroid isthmus was discovered incidentally during total thyroidectomy for multinodular goitre.

CASE PRESENTATION

The patient was a 56-year-old female patient who provided her consent for the publication of this case report. She underwent surgery for multinodular goitre, and agenesis of the isthmus was determined intraoperatively. Her past medical history was significant for hypertension and total abdominal hysterectomy with bilateral salpingo-oophorectomy. Preoperatively, physical examination revealed palpable nodules in both thyroid lobes, and neck ultrasonography (USG) revealed multiple hypoechoic thyroid nodules, the largest being 4 cm and located in the right lobe. Total thyroidectomy was planned. Laboratory analyses were normal. Total thyroidectomy was performed under general anaesthesia with neuromuscular monitoring. It was discovered that the patient had no thyroid isthmus tissue during intraoperative surgical exploration. Intra-operative images of the patient and images of both thyroid lobes after surgical excision are shown in Figures 1 and 2, respectively.

DISCUSSION

Although the incidence of thyroid isthmus agenesis has been reported to range between 0.5% and 10%, the precise incidence rate is unknown (4, 5). Because a few cases have been reported, data regarding the incidence of thyroid agenesis are mainly based on cadaver series. Dixit et al. (1) reported the rate of isthmus agenesis to be 14.6% in a series of 41 cadavers, whereas Ranade et al. (6) 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 etiology 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, 7, 8).

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 USG, computed tomography (CT), and magnetic resonance imaging (MRI) or during thyroid surgery (1). 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 (5). In our case, thyroid function was normal.
When agenesis of the isthmus is determined, diseases such as autoimmune thyroid nodule, thyroiditis, primary thyroid carcinoma, metastasis, and amyloidosis must be considered in the differential diagnosis. Agenesis of the isthmus can be associated with agenesis of the thyroid lobe, ectopic thyroid tissue, or parathyroid hyperplasia (1, 5, 9). Bearing these associations in mind may contribute to safer surgical procedures and fewer surgery-related complications. Therefore, the importance of diagnosing 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.

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.

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REFERENCES