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Case Report

Thyroid isthmus agenesis coinciding with papillary thyroid carcinoma: a case report

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ABSTRACT

Thyroid isthmus agenesis is a rare congenital anomaly characterized by the absence of the thyroid isthmus. Papillary thyroid carcinoma (PTC) is the most common type of thyroid malignancy. The coexistence of thyroid isthmus agenesis and PTC is extremely rare, with limited cases reported in the literature. This case report aims to describe a rare presentation of thyroid isthmus agenesis coinciding with PTC, detailing the clinical presentation, diagnostic approach, and management. Here, we discuss a case of a 49-year-old female presented with neck pain attributed to a cervical disc issue. An MRI scan incidentally identified a left thyroid lesion measuring 5×2.5 cm. Neck ultrasound confirmed a 2.4×2.7×4.2 cm lesion in the left lobe with no suspicious lymph node enlargement. Fine-needle aspiration (FNA) biopsy indicated PTC, classified as Bethesda VI. The patient underwent total thyroidectomy. Intraoperatively, the absence of the thyroid isthmus was noted. Histopathology confirmed a unifocal, 3.9×1.8 cm PTC of the classic subtype, staged as PT2pNx, with no neurovascular invasion and a tumor-free left thyroid lobe. The patient had an uneventful recovery and was discharged two days postoperatively. The absence of the thyroid isthmus can complicate surgical procedures. Preoperative imaging and careful intraoperative management are crucial. This case report highlights the rarity of thyroid isthmus agenesis coinciding with PTC, and emphasizes the importance of recognizing anatomical variations and their implications for surgical management and prognosis. Although it is hypothesized that abnormal development may predispose to malignancy, further studies are warranted to explore the potential association between congenital thyroid anomalies and thyroid malignancy.

Keywords: Case report, Thyroidectomy, Congenital anomaly, PTC, Thyroid isthmus agenesis

INTRODUCTION

The thyroid gland is typically composed of two lateral lobes connected by a thin band of tissue known as the isthmus, which is anterior to the second and third tracheal rings. However, variations in the anatomy of the thyroid gland can occur, with thyroid isthmus agenesis being one of the rarest anomalies. This condition, characterized by the congenital absence of the thyroid isthmus, has an estimated incidence ranging from 0.05% to 0.2% in the general population. Despite being a benign congenital anomaly, the presence of thyroid isthmus agenesis can pose unique challenges in clinical practice, particularly in surgical procedures involving the thyroid gland.[1]

PTC is the most prevalent type of thyroid malignancy, accounting for approximately 80% of all thyroid cancers.[2] PTC is generally associated with an excellent prognosis, especially when detected early and appropriately treated.[3] The occurrence of PTC in patients with congenital thyroid anomalies, such as thyroid isthmus agenesis, is exceedingly rare, with only a few cases documented in the

literature. This co-occurrence presents a unique diagnostic and therapeutic challenge, necessitating careful preoperative planning and intraoperative management.[4]

The embryological development of the thyroid gland involves the descent of the thyroid primordium from the floor of the primitive pharynx to its final position in the neck. Any disruption in this process can result in anatomical variations, including the agenesis of the thyroid isthmus. The etiology of PTC in the context of such congenital anomalies remains unclear, though it is hypothesized that abnormal thyroid development may predispose the gland to malignant transformation.[5]

This case report describes a rare presentation of thyroid isthmus agenesis coinciding with PTC in a 45-year-old female. We aim to highlight the clinical presentation, diagnostic approach, and management of this unusual case, along with a review of the relevant literature.

CASE REPORT

A 49-year-old female presented with complaints of neck pain attributed to a cervical disc issue. An MRI scan of the neck incidentally revealed a left thyroid lesion measuring 5×2.5 cm (Figure 1).

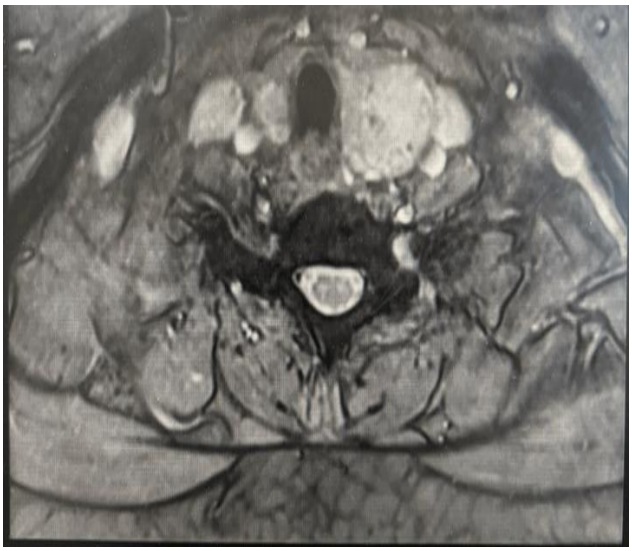

Figure 1: MRI of the neck revealing incidentally discovered left thyroid lesion in a 49-year-old female with papillary thyroid carcinoma.

The patient was subsequently referred for a general surgery consultation. A neck ultrasound was performed, which confirmed the presence of a thyroid lesion measuring 2.4×2.7×4.2 cm in the left lobe with no suspicious lymph node enlargement. FNA biopsy of the thyroid nodule revealed PTC classified as Bethesda VI.

Given the diagnosis, the patient was counseled, and after obtaining informed consent and surgical consensus, a total thyroidectomy was planned. During the surgical procedure, the absence of the thyroid isthmus was noted, confirming thyroid isthmus agenesis. Histopathological examination of the excised thyroid tissue confirmed a unifocal, 3.9×1.8 cm PTC of the classic subtype, staged as PT2pNx. There was no evidence of neurovascular invasion, and the left thyroid lobe was free of tumor, in addition to the confirmed absence of the isthmus.

Postoperative recovery was uneventful, and the patient was discharged two days after surgery with no complications (Figure 2).

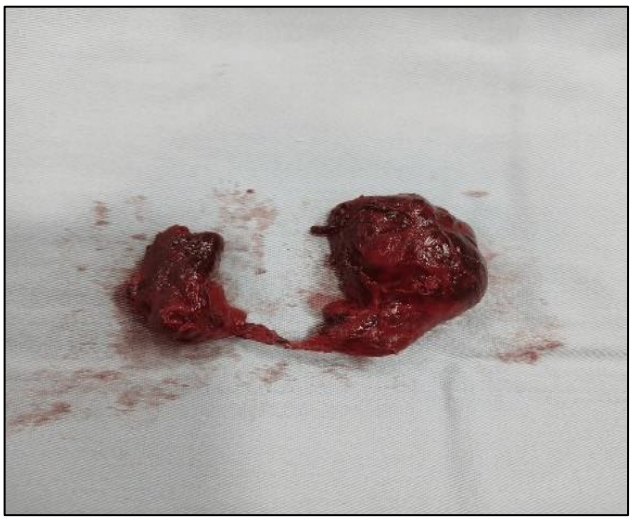

Figure 2: Thyroidectomy specimen demonstrating agenesis of the thyroid isthmus with papillary thyroid carcinoma.

The image shows a thyroid gland specimen following a total thyroidectomy. The specimen is placed on a sterile surgical drape, highlighting the absence of the thyroid isthmus, which is normally situated between the left and right lobes of the thyroid gland. The lobes appear separate and distinct, with a clear gap where the isthmus would typically be located. The tissue is slightly pinkish, typical of thyroid tissue. The texture of the lobes is consistent with thyroid morphology, and small nodules may be visible, indicating the presence of papillary thyroid carcinoma. The image provides a clear visual representation of thyroid isthmus agenesis in conjunction with the carcinoma.

DISCUSSION

Thyroid isthmus agenesis is a rare congenital anomaly characterized by the absence of the thyroid isthmus, which typically connects the two lobes of the thyroid gland. The incidence of this anomaly ranges from 0.05% to 0.2% in the general population.[1] While thyroid isthmus agenesis does not generally cause symptoms, its presence can complicate diagnosing and managing thyroid pathologies.

In this case, the patient initially presented with neck pain due to a cervical disc issue, and an MRI scan incidentally identified a significant thyroid lesion. The subsequent

ultrasound and FNA confirmed the diagnosis of PTC, classified as Bethesda VI. PTC is the most prevalent type of thyroid malignancy, accounting for approximately 80% of all thyroid cancers.[2] It is known for its excellent prognosis, particularly when detected early and treated appropriately.[3]

The decision to perform a total thyroidectomy was made following thorough counseling and consent from the patient. Intraoperatively, the absence of the thyroid isthmus was discovered, confirming the diagnosis of thyroid isthmus agenesis. This anatomical variation necessitated careful intraoperative navigation and reinforced the importance of thorough preoperative imaging to anticipate and plan for such anomalies.[1,4] Histopathological analysis confirmed a unifocal, 3.9×1.8 cm PTC of the classic subtype, staged as PT2pNx. The absence of neurovascular invasion and the lack of tumor in the left thyroid lobe were reassuring findings.

The successful surgical outcome in this patient, who was discharged without complications, underscores the importance of recognizing anatomical variations like thyroid isthmus agenesis. Such knowledge is crucial for adapting surgical techniques to ensure complete resection of the thyroid tissue while preserving surrounding structures.[5] This case also highlights the significance of incidental findings in imaging studies, which can lead to the early detection and treatment of otherwise asymptomatic malignancies.

As a comparison with previous literature thyroid isthmus agenesis is a rare congenital anomaly, with limited cases reported in the literature. A study by Kim et al reviewed 69 patients with thyroid isthmus agenesis and found that 5.8% of these patients also had PTC, underscoring the rarity of this association. In our case, similar to the cases reported, the patient presented with PTC confirmed incidentally, highlighting the importance of imaging in diagnosing asymptomatic cases.[6]

Furthermore, Gandla et al reported a case of PTC in a patient with thyroid hemiagenesis, an anomaly similar in significance to isthmus agenesis, which also complicates surgical approaches. Their case required a modified surgical strategy due to the absence of thyroid tissue typically found in the isthmus region, which aligns with our findings where careful intraoperative management was necessary to navigate the anatomical variation.[7]

Another study by Szczepanek-Parulska et al emphasized the importance of preoperative imaging to identify such anomalies, as the absence of the isthmus could influence surgical planning and outcomes. This resonates with our experience, where detailed imaging allowed for appropriate surgical intervention without complications.[8]

The literature suggests that while thyroid isthmus agenesis itself is benign, its coexistence with PTC necessitates careful consideration during surgical planning. The cases reviewed illustrate that, despite the rarity, the presence of congenital thyroid anomalies like isthmus agenesis does not appear to worsen the prognosis of PTC, but it does require a more tailored surgical approach.

Overall, this case emphasizes the need for heightened awareness among clinicians and surgeons regarding congenital thyroid anomalies and their potential implications. Further research is necessary to explore the relationship between such anomalies and the development of thyroid malignancies, and to refine surgical techniques to accommodate these variations.

CONCLUSION

This case report presents an extremely rare case of thyroid isthmus agenesis associated with papillary thyroid carcinoma in a middle-aged female. Although the absence of the thyroid isthmus is a benign congenital anomaly by itself, it brings some special difficulties into the surgical management of thyroid malignancies. This case demonstrates the necessity for careful preoperative imaging to identify anatomic variations that can impact surgical strategy and performance. Successful management of such patients is therefore illustrative of the importance of interdisciplinary collaboration in sorting out such complex cases for optimal surgical outcomes while reducing postoperative complications.

Incidental detection of PTC in patients emphasizes an important role of extensive imaging studies that may often lead to the discovery of malignant diseases at an asymptomatic stage. Clinicians should therefore maintain a high index of suspicion for congenital thyroid anomalies and consider their possible implications in thyroid cancer. Clearly, further studies are required to resolve the relationship between congenital thyroid developmental anomalies and malignant transformation, which might result in further insights into pathogenesis and adequate management of these rare cases.

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