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Thyroglossal Duct Cyst in an Adult Male: The Risk of Papillary Thyroid Carcinoma

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Abstract
One of the most common benign congenital malformations in the central neck area is a thyroglossal duct cyst, which occurs rarely in adults. Failure of degeneration during embryogenesis leads to the formation of a redundant tract. This tract later forms an expansile cyst with an epithelial lining and the capacity to secrete fluid, resulting from repeated infection or inflammation. The Sistrunk procedure remained the mainstay of treatment to prevent recurrence. However, there is 1% chance the cyst will evolve into carcinoma and influence the extent of the treatment provided to the patient. Here, we presented a case of a 25-year-old gentleman who presented with a painless midline neck mass for one year and underwent the Sistrunk procedure after diagnosis was confirmed after work-up. Nonetheless, histopathology revealed a well-differentiated papillary thyroid carcinoma in a previously normal fine needle aspiration cytology. The patient later underwent total thyroidectomy and received radioactive iodine therapy. The patient's prognosis was excellent postoperatively. Despite its rarity, every patient with neck swelling should include malignancy risk stratification by a full and comprehensive examination by the attending clinician.

Keywords: Thyroglossal duct cyst; Sistrunk procedure; Thyroglossal duct cyst carcinoma; papillary thyroid carcinoma

Introduction

The thyroglossal duct cyst (TGDC) arises from the failure of involution of the thyroglossal duct, runs from the foramen caecum through the hyoid bone to the thyroid gland. Normally, involution occurs between the 8th and 10th week of gestation[1]. Failure of this process can lead to the formation of a TGDC, which contains an epithelial lining and is capable of producing secretions after repeated inflammation and infection[2]. In

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children, TGDC is regarded as the most common benign congenital neck deformity, accounting for about 75% of midline neck masses[1].

It is rare about 7% of the adult population but common in childhood[1,2]. Adult TGDC is generally indistinguishable from its carcinoma variant, which develops in about 1% of cases[1,2].

Diagnosis is made after the Sistrunk procedure which entails the complete removal of the cyst, tract, and hyoid bone [3]. We present a case of an adult thyroglossal duct cyst with negative preoperative fine-needle aspiration cytology, confirmed by final histopathology following the Sistrunk procedure.

Case presentation

A 25-year-old male with no co-morbidities presented with a midline neck swelling for a year. It presented initially as a small swelling with occasional discharge of yellowish fluid. The swelling gradually increased in size and became painful when infected. He has no other complaints, such as obstructive or constitutional symptoms. Physical examination revealed a non-tender, firm, smooth-surfaced, solitary 2 x 2 cm midline neck swelling that moves with deglutition and tongue protrusion. There was a punctum, but no discharge was seen. No other palpable cervical lymph nodes were found. Flexible nasopharyngoscopy showed normal findings. Ultrasound (USG) neck reported as the presence of a well-defined hypo-echoic lesion measuring 1x1 cm, located at the infrahyoid, as well as normal thyroid tissues with no nodules seen. Contrast-enhanced computed tomography (CECT) of the neck was conducted, revealing an infrahyoid midline cyst measuring 1.6 x 1.3 x 1.9 cm, consisting of a central heterogeneous solid component and foci of calcification (Figure 1).

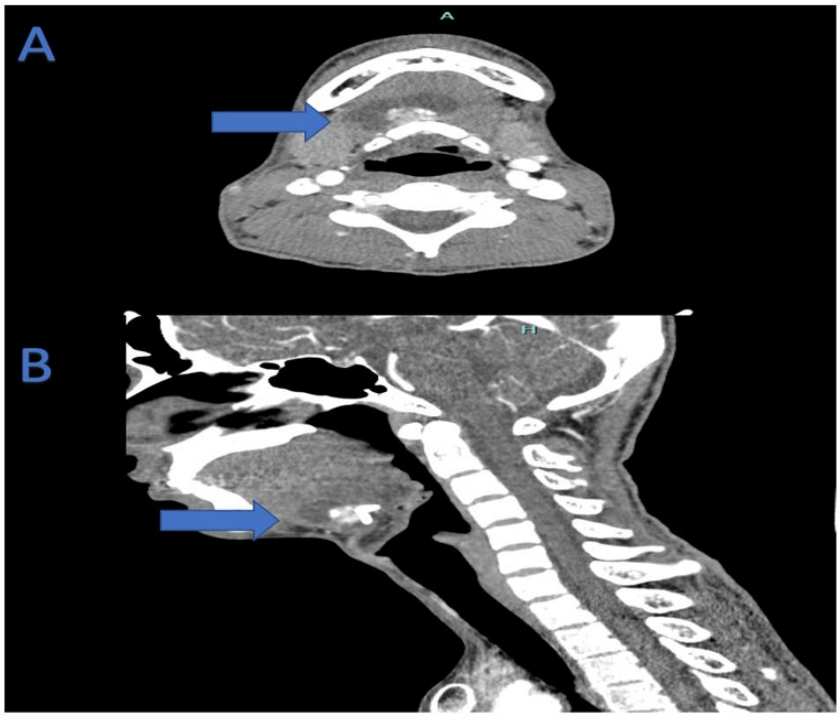

Figure 1. Contrast-enhanced Computed Tomography of the neck;

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(A) Arrow shows the presence of a well-defined hypodense thyroglossal duct cyst with central foci of calcification anterior to the body of the hyoid bone, no evidence of hyoid bone erosion, and involvement of adjacent muscles.
(B) Arrow showed the relation of a well-defined hypodense thyroglossal duct cyst with foci of calcifications to the tongue base at the level of the hyoid bone.

He proceeded with fine-needle aspiration cytology (FNAC) as part of the workup. However, the specimen consisted of acellular, yellowish, cystic material, with no malignant cells detected. Thyroid function tests were normal.

He underwent the Sistrunk procedure, during which an intraoperative note stated a firm, 2 x 1 cm, yellowish, solitary cyst with its tract attached to the body of the hyoid bone, which was removed in toto. The thyroid glands appear normal without any suspicious lesions or infiltration into the surrounding structures. Histopathological examination confirmed the diagnosis of papillary thyroid carcinoma (PTC) with TGDC, showing thyroid tissue with foci of neoplastic cells arranged in papillary architecture, exhibiting enlarged, overlapping nuclei with irregular nuclear membranes, nuclear grooving, and chromatin clearing (Figure 2A-C).

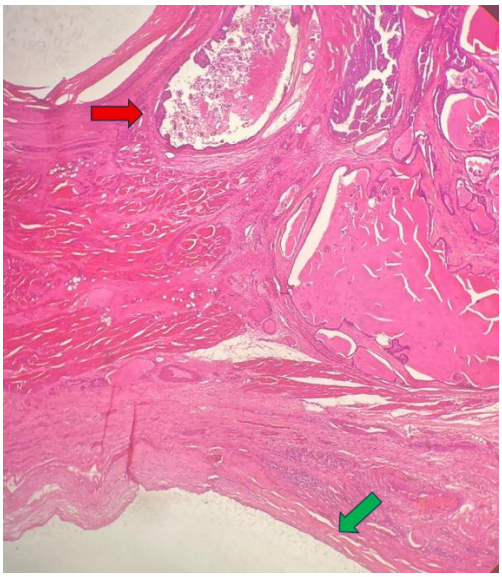

Figure 2A. Thyroglossal duct cyst (green arrow) with malignant papillary thyroid carcinoma cluster (red arrow) in H&E, X 4 magnification.

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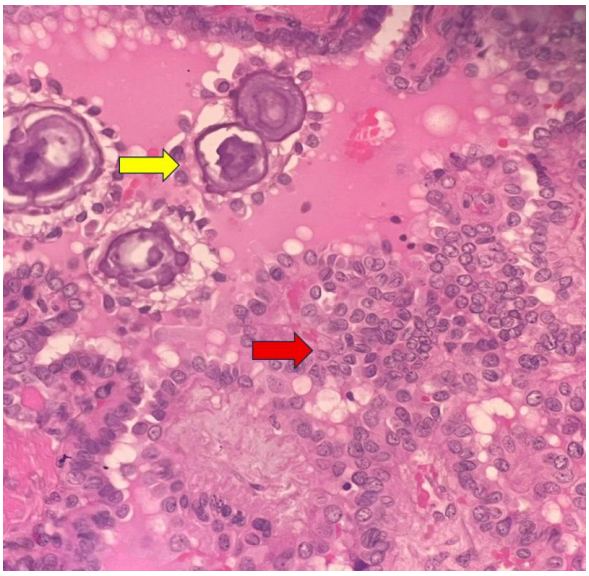

Figure 2B. The psammomatous calcification of papillary thyroid carcinoma (yellow arrow) in H&E, x 40 magnification. The malignant cells show overlapping nuclei, irregular nuclear membrane, nuclear grooving, and chromatin clearing (red arrow).

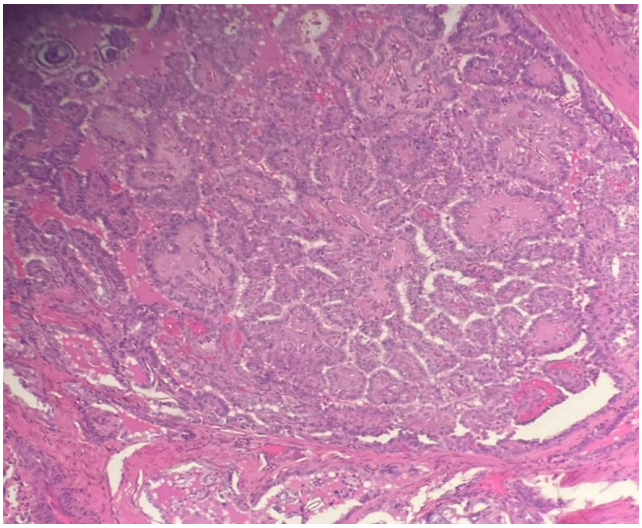

Figure 2C. showed malignant cells arranged in a papillary architecture in H&E, x10 magnification.

There was no lymphovascular or perineural invasion. Neoplastic cells were thyroglobulin positive. Total thyroidectomy performed 4 weeks later revealed normal thyroid tissue. He was well postoperatively during follow-up without recurrence of neck swelling. He received radioactive iodine (RAI) ablation and was prescribed lifelong thyroid hormones.

Discussion

Adult-onset thyroglossal duct cysts (TGDC) can raise suspicion for malignancy, as they have the potential to undergo malignant transformation[3]. The incidence of PTC in TGDC is more frequent in adults with the

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prevalence of 2:1 female to male ratio and mean age of 40-year-old [4]. PTC has been reported as the most frequently identified cancer in TGDC carcinoma (85%) along with other types of carcinomas such as mixed, papillary/follicular (7%), squamous cell carcinoma (SCC) (5%), and a few other rare types (2%); Hurtle cell, follicular, and anaplastic carcinoma[5].

The presentation of TGDC carcinoma may resemble its benign counterpart [4]. It appears as a mobile midline neck mass. Additional symptoms—such as pain, dysphagia, odynophagia, dyspnoea, and dysphonia—often signal complications and may indicate cancer spread [4,5].

The aetiology of the entity has been proposed as either a primary neoplasm arising from de novo ectopic thyroid tissue within the cyst or direct metastasis of PTC from the thyroid gland [6]. Hence, the preoperative identification will be difficult. The limitation of FNAC to exclude the malignancy has been reported in the literature [4–6]. The diagnostic rate is only 47-53%, which may indicate false results of benign tumour [4–6]. This occurs due to poor cytological sample dilution within the cyst itself with the relatively small size of the malignant elements [2].

Histopathological findings usually confirm the diagnosis [7]. Microscopic evaluation revealed the presence of papillary carcinoma (finger-like projection overgrowth) surrounded by a background of benign thyroid parenchyma and TGDC lined by epithelium [3]. Our FNAC was unable to yield malignant cells, and the diagnosis of PTC was made later after our in-house pathologist reviewed the post-operative specimen. The workup of TGDC should also include radiological assessment to examine the cyst and confirm the presence of the thyroid gland [8]. Thus, USG neck is recommended as the initial assessment for paediatric cases, while CECT neck is preferred for adults to aid surgical planning, particularly in determining lesion size, extent, and content [7,8]. In our patient, the ultrasound revealed a normal thyroid without nodules or suspicious lymph nodes. This occurs because the lesion was small at the time of assessment. Due to high suspicion of malignancy, we proceeded with a CECT neck to rule out local infiltration or lymphadenopathy. This imaging can detect cyst calcifications, suggestive of malignancy, as these have not been reported in benign TGDC [7,8].

The diagnosis of PTC within TGDC is challenging because it is difficult to distinguish from its benign counterpart. Histopathological evaluation after excision can aid in reaching a definitive diagnosis. As it's rare, there is no consensus on standardised treatment [9]. The Sistrunk procedure is the initial surgical option to remove TGDC carcinoma [6–9]. This procedure offers a promising cure rate of up to 95% for PTC in TGDC with an excellent prognosis [6–9].

Sistrunk procedure is warranted in all TGDC cases. The removal of the cyst and its duct around the foramen caecum at the tongue base along with partial hyoid bone resection can reduce as low as 3% recurrence rates [1,5]. However, in the cases of TGDC carcinoma, the role of total thyroidectomy is debated and become controversial [9]. Some authors highlight the high risk of unsuspected PTC infiltration, the incidence of 'occult' thyroid carcinoma and the risk of secondary malignancies in the thyroid, even when preoperative findings are unremarkable [9]. In contrast, others recommend total thyroidectomy only if the patient meets specific criteria: 1) over 45 years of age, 2) aggressive disease, 3) childhood neck irradiation, 4) large tumours>1.5 cm, or 5) histological findings such as positive tumoral margin, cyst invasion, or lymph node metastases [1,3–5].

Most tumours were identified in patients aged 45 years or older (67%). while the remaining 37% had synchronous PTC after total thyroidectomy [6,9].

The adjuvant RAI ablation is given to those who have undergone total thyroidectomy to enhance therapeutic outcomes, according to some authors [1,4]. Despite that, some other authors did not recommend RAI for the patient and suggested annual thyroid-stimulating hormone levels with ultrasound neck for the patient's surveillance [3]. Our case had a second surgery, which was a total thyroidectomy, as it was a large tumour> 1.5cm, followed by RAI aiming to ablate the possibility of occult extrathyroidal extension. If neck metastasis is present, therapeutic neck dissection is encouraged to eradicate the involved nodes for a better prognostic approach [6–8].

The overall prognosis of patients with thyroglossal carcinoma is good, especially for the common PTC subtype after the Sistrunk procedure, with a reported 10-year survival rate of 95–100% [9]. In contrast, SCC have a poorer prognosis, requiring more aggressive treatment, with 36% reported mortality [4].

Conclusion

PTC within TGDC is usually diagnosed after total excision by histopathological evaluation. Preoperative diagnosis is difficult due to its varied aetiologies. Even though treatment is controversial, the Sistrunk procedure is the preferred initial surgery to eradicate the TGDC carcinoma. Once the diagnosis is confirmed, second-stage surgery-such as total thyroidectomy and neck dissection-can be tailored according to the patient's prognosis and potential outcomes, if indicated. Another approach, such as RAI, can be used to complement post-total thyroidectomy for low-risk patients; a more aggressive approach is needed for high-risk patients, as mentioned earlier, like PTC. The prognosis, however, is excellent.

Patient Consent Statement

Written informed consent was obtained from the patient for the publication of this case report and any accompanying images.

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Conflict of Interest Statement

The authors declare no conflicts of interest.

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