A RARE CASE OF PAPILLARY THYROID CARCINOMA (PTC) ARISING FROM THYROGLOSSAL DUCT CYST

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Abstract

Thyroid gland is the first endocrine organ to develop during intrauterine life (3rd to 4th week of gestation). Ectopic thyroid is a rare developmental defect involving the aberrant embryogenesis of the thyroid gland during it's migration from the floor of the primitive foregut to it's final pretracheal location. Ectopic thyroid tissue can be found in many sites, the most common being the base of the tongue (lingual thyroid) but it can also be found anywhere along it's embryonic course - the thyroglossal tract. Other less common sites include trachea, palatine tonsils, carotid bifurcation, iris of the eye, pituitary gland, axilla, heart and ascending aorta, thymus, esophagus, duodenum, gall bladder, stomach bed, pancreas, mesentery, porta hepatis, adrenal gland, ovary, fallopian tube, uterus, and vagina. It's prevalence is about 1 per 100000–300000 people. However postmortem studies have shown a prevalence ranging between 7 to 10%. One in three infants who present with hypothyroidism have ectopic thyroid tissue. And it is most commonly seen in females, especially of Asian descent.

Conventionally, the etiology of ectopic thyroid has been considered to be multifactorial. However, recent genetic studies have indicated that mutations in the genes TITF-1(Nkx2-1), Foxe1(TITF-2) or PAX-8 may be etiologic. Majority of the cases are asymptomatic and some present as hypothyroidism. But rarely, some cases of ectopic thyroid may turn malignant with or without the involvement of the primary orthotopic thyroid gland leading to diagnostic and therapeutic dilemmas. We report one such rare case of papillary carcinoma arising from the ectopic thyroid located in the thyroglossal duct in a 21 year old Indian female with a normal orthotopic thyroid gland.

Anatomically and functionally normal orthotopic thyroid does not exclude the possibility of pathologic changes in the ectopic thyroid tissue as is seen in this case. Though rare, the possibility of an ectopic thyroid carcinoma must always be considered in cases of cystic midline neck swellings.

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1. Introduction

Papillary thyroid carcinoma (PTC) is the commonest type of thyroid neoplasm. It constitutes of about 80-85% of all thyroid carcinomas and carries the best overall prognosis. Histopathologically, it is an epithelial malignancy showing evidence of follicular differentiation. It is generally sporadic but some cases may be associated with familial adenomatous polyposis (Gardner syndrome, Werner syndrome, Carney complex type etc.). Somatic rearrangement of the RET protooncogene is the most frequent genetic abnormality seen in PTC.

Ectopic thyroid is defined as the presence of thyroid tissue at a site other than the pretracheal area i.e., thyroid tissue localised outside of the level of second to fourth tracheal cartilages. Even though, PTC is a fairly common entity, the presence of ectopic thyroid tissue in thyroglossal duct is an uncommon condition. And malignant transformation of the ectopic thyroid located in a thyroglossal duct cyst makes it even rarer (accounting for less than 1% of all cases of thyroglossal duct cysts). There are less than 300 such cases reported in literature till date.

The uniqueness of the case presented here strives in the rarity that the orthotopic thyroid gland was free of malignancy, despite ectopic tissue located in the thyroglossal duct being positive for thyroid carcinoma (PTC).

Case Report :

A 21 year old Indian lady presented to the surgical OPD of Meenakshi Medical College Hospital & Research Institute, Kanchipuram, India with the complaints of a painless swelling in the anterior aspect of the neck since 4 years. She gave no history of dysphagia/ dyspnoea/ weight gain or loss/ palpitations/ heat or cold intolerance/altered bowel movements/ tremors. However, she did give a history of menstrual irregularities. She had been diagnosed as a case of polycystic ovarian disease on work up for the same and was on sustained medication and regular follow up at the time of presentation.

On local examination, a solitary oval swelling measuring ~3x2cm was palpable in the suprahyoid region. The swelling was firm in consistency, non-tender and moved with the protrusion of tongue but not with deglutition.

She was evaluated for the same with USG Neck which showed an irregular heteroechoic mass lesion in the suprahyoid region with a cystic component and increased vascularity. Findings were suggestive of ?Ectopic thyroid malignancy vs ? Infected thyroglossal duct cyst. On further evaluation with CECT Neck, a well-defined heterogeneous solid and cystic lesion measuring 3x2.8x2.5cm was noted anterior to the hyoid bone. Enlarged lymph nodes were noted in Ib and II levels. Other remarkable radiological findings were loss of fat plane in the superior margin and geno-hyoid muscle infiltration. Orthotopic thyroid showed no abnormality radiologically. The above findings were indicative of malignant transformation of the ectopic thyroid located in the thyroglossal duct cyst. Thyroid function tests of the patient were within normal limits and thyroid scan showed a normal distribution of radiotracer in the thyroid gland. USG guided FNAC of the suprahyoid swelling showed monolayered sheets of papillary fragments with nuclear crowding and overlapping along with powdery chromatin and occasional intranuclear cytoplasmic inclusions and nuclear grooving (Fig. 1 & 2). Was reported as Category III FLUS (Follicular lesion of undetermined significance).

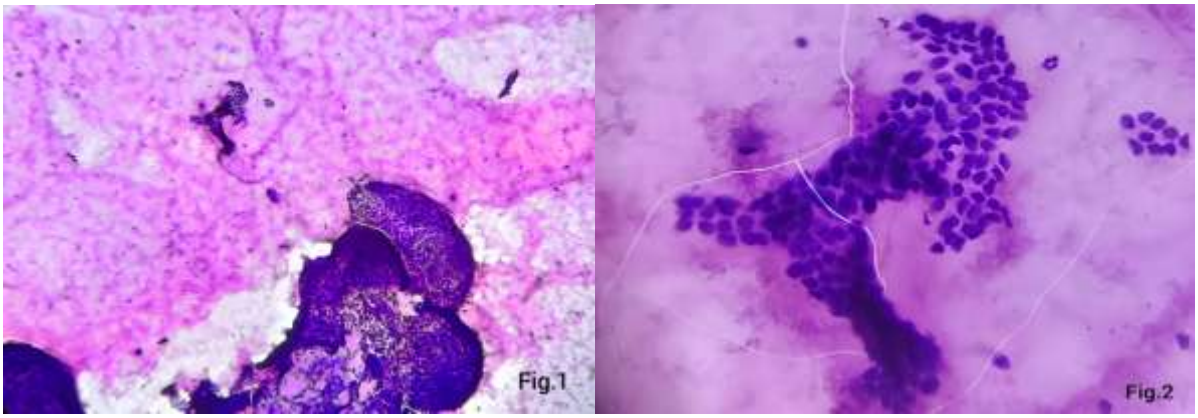


Fig.1 & Fig.2 - H&E Staining (10X, 40X), Monolayered sheets of papillary fragments on FNAC.

In view of the above findings, she underwent 'Sistrunk procedure' which involves excision of the thyroglossal duct cyst, the middle part of the hyoid bone and the surrounding tissue around the thyroglossal duct (Fig. 3). Intra-operative and post-operative period were uneventful. She is under regular follow-up and has been asymptomatic till the publication of this case report.

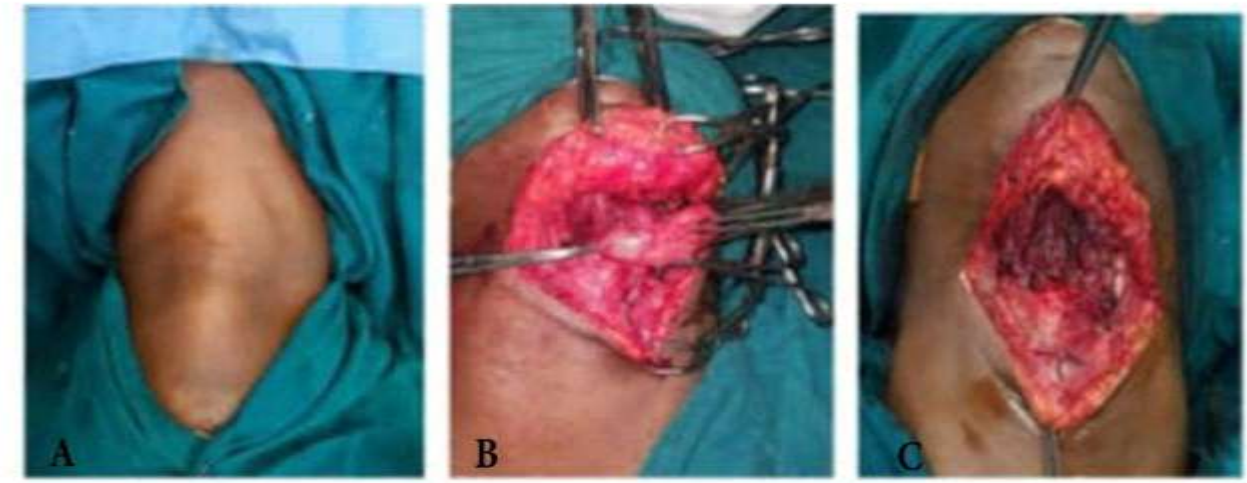


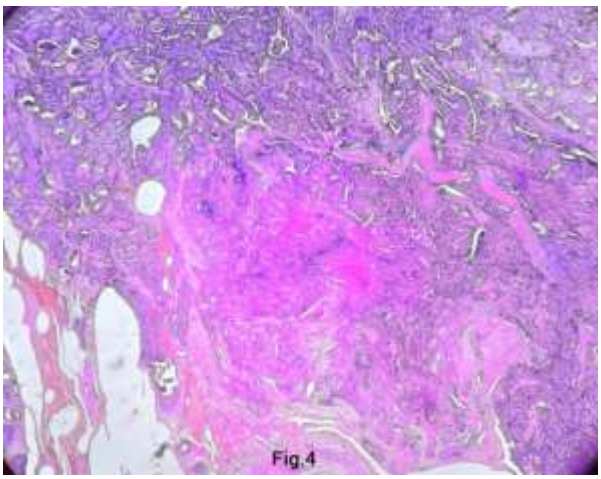
Fig.3 A) Suprahyoid swelling. B) Intra-op image of the thyroglossal duct cyst. C) View of the surgical site after excision of the lesion in toto.

Gross Examination :

We received an excised grey brown specimen of thyroglossal duct cyst measuring 3.5x3.2x1.5 cm. Cut section showed an ill-defined grey white area measuring~1.5x1.3 cm infero-laterally.

Microscopic Features :

Histopathological examination of the material received showed a cyst lined by stratified squamous epithelium with an adjacent malignant neoplasm arranged in complex papillary architecture and in follicles. The papillae and the follicles were lined by cuboidal cells with nuclear enlargement, nuclear overlapping and crowding and optically clear nuclei (Fig.4 & 5). Occasional nuclear pseudo-inclusions and nuclear grooves were seen. Psamommatous calcification and perineural invasion were also noted. The surrounding fibromuscular tissue showed involvement. However, margins were free of tumour invasion. Findings were consistent with Papillary carcinoma thyroid arising from thyroglossal duct cyst.



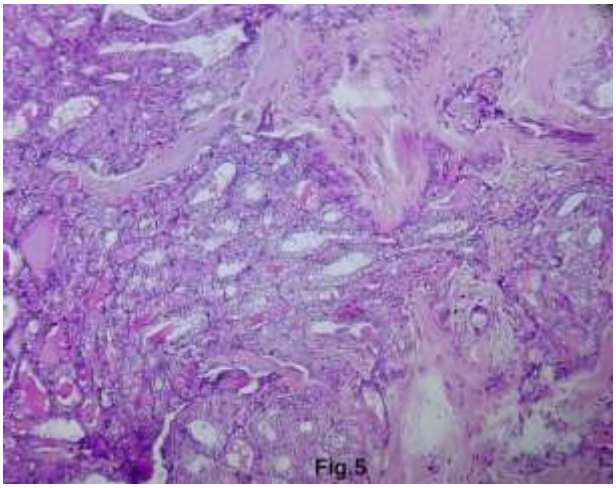


Fig.4 and Fig.5 - H&E Staining (10X)- Papillae and follicles lined by cuboidal cells with nuclear enlargement, nuclear overlapping, crowding and optical clearing.

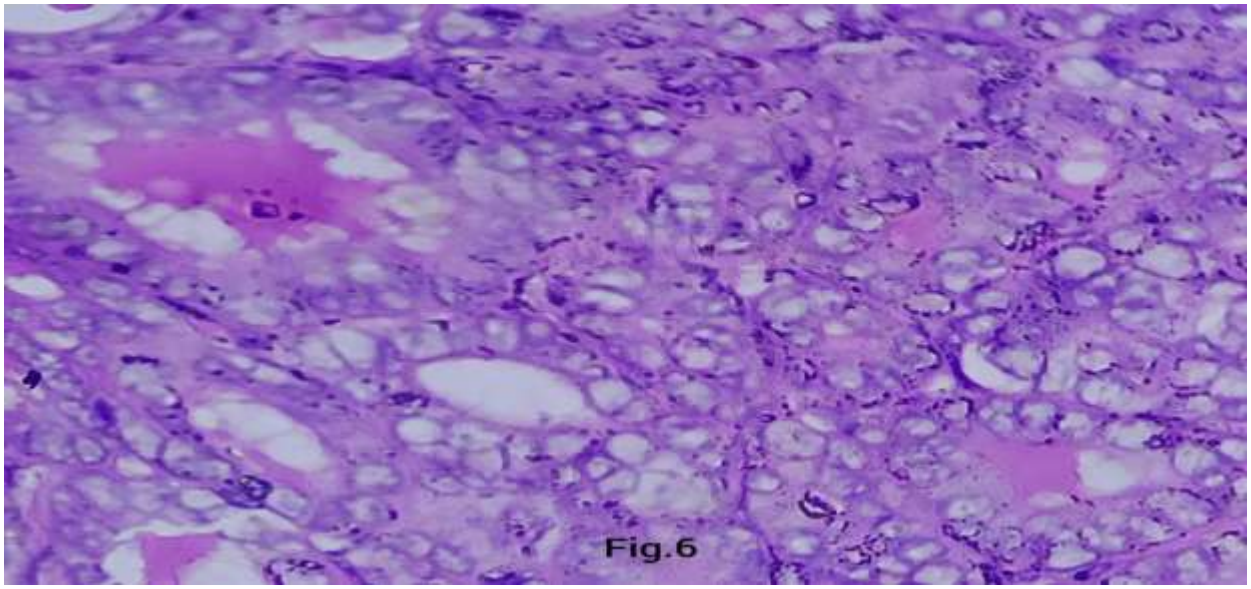


Fig. 6 - H&E Staining (40X) - Neoplastic cells with prominent optically clear nuclei/Orphan Annie nuclei, nuclear grooving and psammoma bodies.

2. Discussion

Thyroglossal duct cysts are congenital fibrous cysts that form as a result of the persistence of the thyroglossal duct which is an embryologic remnant extending from the foramen cecum in the tongue to the thyroid's final location (level of 2nd to 4th tracheal cartilage). The thyroid begins to develop around the third week of gestation as a median outgrowth from the primitive pharynx. The thyroid primordium which is derived from the pharyngeal endoderm originates at foramen cecum at the junction of the anterior two thirds and posterior one third of the tongue. From there, the thyroid migrates caudally passing anterior to the developing hyoid bone and laryngeal cartilage. It reaches it's final position in the pretracheal neck around the seventh week. The thyroglossal duct is the narrow tubular structure left from the thyroid's descent which involutes by tenth week of gestation. If any part of the duct persists, secretions from the epithelial lining can lead to cyst formation.

The most common primary thyroglossal duct cyst carcinoma is papillary carcinoma (75–80%), but other types of primary tumour do occur, such as mixed papillary-follicular carcinomas (7%), squamous cell carcinoma (5%), follicular carcinoma (1.7%); Hürthle cell and anaplastic carcinoma (0.9%). And these de novo tumours often exhibit biological behavior similar to their primary gland counterparts.

There is considerable dissent between clinicians on the appropriate line of management of PTC. The extent of surgery (lobectomy vs total thyroidectomy), timing of central neck dissection (prophylactic vs therapeutic) and indications for radioactive iodine ablation, targeted oncotherapy and TSH suppressive therapy are widely debated. However, there has been a shift in treatment paradigm from a radical to a risk stratified management approach in the recent years. Risk stratification is done based on the following parameters.

Low risk patients are considered to be the ones that have a unifocal, well-encapsulated tumor without extrathyroidal extension, without local or distant metastases, without aggressive histology and without lymphoid or vascular invasion.

Intermediate risk patients are the ones with microscopic extrathyroidal invasion, cervical lymph node metastases, I-131 uptake outside the thyroid bed, having aggressive histology (tall cell, columnar cell, insular carcinoma) or if vascular invasion is present.

High risk patients are considered to be the ones who have multifocal lesions, have extrathyroidal extension, have vascular invasion, are incompletely resected, have lymph node metastases, especially if multiple nodes are involved and the nodes are > 3cm in largest dimension or if extra-nodal tumour extension is present.

Management of a thyroglossal duct PTC includes Sistrunk procedure as a stand-alone surgery in low-risk patients. It consists of the excision of the thyroglossal duct cyst, the middle part of the hyoid bone and the surrounding tissue around the thyroglossal tract.

However high-risk patients need to undergo an additional total thyroidectomy and postoperative radioactive iodine ablation therapy with or without neck dissection.

Targeted therapy drugs such as Lenvatinib, Sorafenib or Cabozantinib may be tried if the cancer has metastasised and/or other treatments are not helpful.

3. Conclusion

This case demonstrates the essential role of diagnostic cytology in the evaluation of a midline neck mass. A cytological evaluation becomes imperative in a presumed thyroglossal duct cyst (cystic midline neck mass) to rule out/confirm the presence of ectopic thyroid tissue with/without neoplastic or secondary changes. Also a preoperative sonographic identification of an orthotopic thyroid gland in patients diagnosed with thyroglossal duct cyst is a must so as to confirm the presence of a normally positioned and normally functioning primary thyroid gland. The case reported here holds the rare distinction of being a case of primary papillary carcinoma in a thyroglossal duct cyst in the absence of concomitant involvement of the orthotopic thyroid gland.

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