Case Report

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Colloid goitre in submental ectopic thyroid: a rare entity posing a diagnostic and therapeutic challenge

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ABSTRACT

Ectopic thyroid is a rare entity in clinical practice. It results from defects at early stages of thyroid gland embryogenesis, during its passage from the base of the tongue to its final pre-tracheal position, anterolateral to second, third, and fourth tracheal rings. It is usually found along the course of the thyroglossal duct or laterally in the neck, also in distant places such as the mediastinum or very rarely in the sub-diaphragmatic organs. In our case, an 18-year old girl presented with a sub-mental swelling. There are various differentials for such swelling. A careful history, scrupulous clinical examination and investigations comprising ultrasonography of neck, MRI scan, Tc-99m pertechnetate thyroid scan and Fine needle aspiration cytology (FNAC) from the swelling revealed that it was a case of colloid goitre in the ectopic sub-mental thyroid and there was no thyroid tissue in its normal anatomic location. We initially treated the patient with supplemental levothyroxine trial for 6-months followed by surgical excision. We report this case as it is a rare phenomenon and also because diagnostic and therapeutic dilemma exists in its management.

Keywords: Sub-mental swelling, Tc-99m pertechnetate thyroid scan, Ectopic thyroid, Colloid goitre

INTRODUCTION

Thyroid ectopia is a congenital disease due to abnormal migration of thyroid gland in the embryonic stage, which usually manifests later in life especially during increased physiological demand of thyroid hormones. Prevalence of this entity is about 1 per 1,00000-3,00000 people and 1 per 4,000–8,000 patients with thyroid disease. 1,2 However, in autopsy studies, the prevalence ranges from 7 to 10% in asymptomatic general population. Ectopic thyroid tissue (ETT) most commonly seen in females (65 to 80% of cases). It may occur at any age but most common at younger ages. It is extremely rare to have dual or triple ectopic thyroid tissue with or without presence of a normally located pre-tracheal thyroid gland. In >70% of cases of ETT, the eutopic thyroid gland is

absent.^{5,6} This article emphasizes the role of scrupulous clinical examination in a case of neck swelling and describes current knowledge about the presentations of ETT at different locations. In addition, we also try to optimise the clinician's diagnostic approach and treatment in a case of ETT.

CASE REPORT

An 18-year-old Bengali girl presented to our surgery unit with a 1-year history of a painless swelling below the chin in august 2013. She first noticed a small marble-ball sized swelling but was gradually increased in size over next one year. She did not report any difficulty in swallowing and breathing, or any history of fever or local trauma. There was no abnormality in her birth history,

developmental milestones, and menstrual history and she did not have any history of neck swelling in her family members. On clinical examination, a 5x4 cms, nontender, smooth and firm swelling was found just below the chin but it was not moving with deglutition or protrusion of the tongue (Figure 1a, 1b).



Figure 1: (a) Swelling in submental region (b) No movement of the swelling in protrusion of the tongue.

There was no other swelling in the neck and the oral cavity. On ultrasound of the neck, we suspected ectopic thyroid gland in sub-mental (supra-hyoid) region in absence of eutopic thyroid. FNAC of the swelling revealed adenomatous hyperplasia of the thyroid gland. T2-weighted MRI images showed a hyper-intense lesion below the tongue (Figure 2a). A technetium-99 thyroid scan showed marked uptake in the submental mass but no uptake in the normal location of the thyroid gland (Figure 2b).

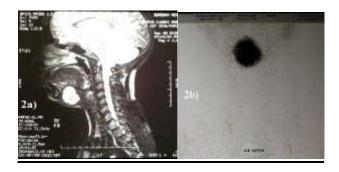


Figure 2: (a) T2-weighted MRI images shows a lesion below the tongue with high signal intensity (b) Tc-99m pertechnetate thyroid scan shows uptake in the region of the submental mass but no uptake in the usual area of the thyroid gland.

On biochemical examination she was found to be hypothyroid (serum TSH 16.4 mIU/L) but without clinical symptoms of hypothyroidism. We initially treated her with 100 mcg/day levothyroxine trial for 6- months but the swelling did not regress in size. We explained her about the surgical consequences and the need for lifelong thyroid hormone replacement therapy but she was very much concerned about the cosmetic deformity and also worried about future chances of malignant

transformation. Surgical extirpation done (Figure 3a) on March 2014 and the histopathological features were suggestive of colloid goitre (Figure 3b). There was no recurrence in last 1.5-year follow-up.

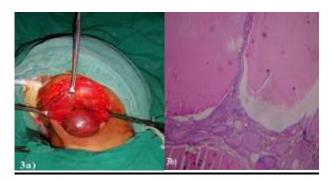


Figure 3: (a) Dissection of the mass in the neck (b) Histopathological slide showing colloid filled follicles (haematoxylin and eosin stain x 100).

DISCUSSION

During fetal development thyroid gland is the first endocrine gland to develop and developmental abnormalities of the thyroid gland can be classified into three groups:

- A. Agenesis of thyroid gland, causing of neonatal hypothyroidism
- B. Dysgenesis (hemiagenesis /ectopia) of the gland
- C. Persistence of the thyroglossal duct and related abnormalities.

First ectopic thyroid case was reported by Hickman as a lingual thyroid in 1869 where a newborn presented with an upper airway obstruction 16 hours after birth. ETT in the sub-mental region is relatively rarely reported and is usually associated with a thyroglossal duct cyst. we encountered a rare case of colloid goitre in a sub-mental ectopic thyroid presenting as sub-mental swelling which was not associated with thyroglossal duct cyst.

Embryology

From embryological point of view, an endodermal diverticulum developed from the median plate of the floor of the pharyngeal gut, during the 3rd or 4th week of intrauterine life. This diverticulum descends in the midline, from the foramen cecum (located between the anterior two-thirds and posterior third of the tongue) to the final location of the gland, anteriorly to the pretrachea and larynx. This migration begins at embryonic day 24 and as a result, a narrow channel is created (thyroglossal duct) which undergoes atrophy prior to the definitive thyroid gland formation. Ectopic thyroid tissue is the result of a failure of migration of thyroid, not only along the route of thyroglossal duct but also rarely in subdiaphragmatic organs.⁸

Pathology & Genetics

The cause of ectopic thyroid tissue remains unclear in most of the cases. Genetic research has shown that the transcription factors TITF1 (NKX2-1); FOXE1 (TITF2) and PAX8 are essential for thyroid morphogenesis and

differentiation. Mutation in these genes may be involved in abnormal migration of the thyroid. It has been shown in gene-targeting experiments that FOXE1 is required for thyroid migration. Mice homozygous for FOXE1 mutations found to have a sublingual thyroid.⁹

Table 1: Published case reports on ectopic thyroid with their different locations & presentations.

Author	Year of publication	Age & Sex	Location of Ectopic thyroid	Presentation	Management
EyüboĞlu E et al ¹¹	1999	50[F]	Pancreas	Deodenal ulcer pain	Excision of mass
Fumarola A et al ¹²	2006	36[F]	PTC arising in neck branchial cyst	Lt. sided neck mass	Total thyroidectomy; Central & Lat. compartment LNs dissection.
Guimarães MJAC et al13	2009	40[F]	Ant. mediastinal mass	cough; dyspnea	Excision
Wu Z et al ¹⁴	2009	33[F]	Rt. ventricle	Exertional dyspnea	Open heart surgery
Choudhury BK et al ⁶	2010	17[F]	Base of tongue	Dysphagia	Daily 100mcg levothyroxine
Keles E et al ¹⁵	2012	32[F]	Rt. submandibular region	Swelling under Rt jaw.	Excision of mass with Rt. hemithyroidectomy
Khediri Z et al ¹⁶	2012	17,31, 45[F]	Three cases of ovarian cystic mass (Struma ovarii)	Menstrual irregularities	Laparotomy
Kim SY et al ¹⁷	2013	67[M]	Rt. paratracheal ectopic thyroid	Detected on follow up CT chest in an asymptomatic post- treatment colon cancer pt. mimicking metastatic lesion.	Surgical excision
Deshmukh SD et al ¹⁸	2014	24[F]	Submental ectopic thyroid	Submental swelling	Excision
Wang J et al ¹⁹	2014	45[F]	Cervical & Ant. mediastinal ectopic thyroid	Chest pain	VATS
Hebbar AK et al ²⁰	2014	25[F]	PTC in Median Aberrant Thyroid (Ectopic)	Neck swelling	Excision of med. aberrant thyroid
Adelchi C et al ²¹	2014	33[M]	Retracted dermal scar above the hyoid bone.	Midline neck fistula (thyroglossal duct cyst)	Excision

(Age in years; Sex: M=male, F= female; Rt=Right, Lt=Left, Lat= Lateral, Ant = Anterior; PTC =papillary thyroid carcinoma; VATS=Video assisted thoracoscopic surgery)

Presentation

Most of the patients with ectopic thyroid are asymptomatic, while some cases are detected incidentally. Lesions which usually affect the eutopic thyoid gland can also involve these ectopic thyroid tissues, though it carries a low risk of malignancy 10 Symptoms are usually related to size and location of the ectopic gland and associated endocrine dysfunction. The

most frequent location of ectopic thyroid tissue is at the base of tongue, particularly at the region of the foramen cecum (lingual thyroid), accounting for about 90% of the published cases. Apart from the lingual thyroid, ETT has been described in many other sites between the base of the tongue and its final pre-tracheal position, as well as in the mediastinum and distant sub-diaphragmatic organs. Hypothyroidism is commonly present because of the absence of a normal thyroid gland in most instances. Hyperthyroidism is very rare. A thorough search of

English medical databases, using key word 'ectopic thyroid tissue' revealed some interesting case reports, which were compiled in Table 1.^{6,11-2}

Differential diagnosis

Differential diagnoses of cervical ETT include salivary gland tumors, thyroglossal duct cysts, midline branchial cysts, subhyoid bursitis, lymphadenopathy, lymphangioma, epidermal cysts, hemangioma, adenoma, fibroma, lipoma and dermoid cyst.

Investigations

High resolution ultrasonography (USG) is generally favoured as the initial imaging test of choice for evaluation of any neck mass which can differentiate between solid and cystic lesions. If goitre is suspected in USG then thyroid profile including fT4 and TSH is necessary to determine presence of thyroid endocrine abnormality. Next line investigation is FNAC to get tissue diagnosis. The most important diagnostic modality for ectopic thyroid is radio-nuclear scanning with technetium-99 m, it also can detects presence of ETT in other parts of the body (dual/triple ectopic) as well. However, computed tomography (CT), and magnetic resonance (MRI) imaging may help to define the extension and location of the ectopic thyroid gland.

Management

Asymptomatic and euthyroid patients do not require any treatment but should be kept under follow up. For those with obstructive symptoms and hypothyroidism, initial suppressive therapy with levothyroxine is recommended which helps to achieve euthyroid state and reduce the size of the thyroid gland. Surgical intervention is reserved in cases which fail to respond to thyroxine trial, if there is any pathology involving ETT or for cosmetic reasons where large lesion causing disfigurement. Radio-iodine ablation therapy is an alternative in patients who are not fit or refusing surgery. Ablative radioiodine therapy usually avoided in children and young adults, due to its potential deleterious effects on the gonads. However, after surgery and ablative therapy life-long levothyroxine supplementation is needed. 39,222

CONCLUSION

ETT although rare, should be kept in clinician's mind in differential diagnosis of midline neck swelling specially in childrens and young adults, as it is often misdiagnosed as thyroglossal cyst. Thyroid scintigraphy plays an important role in establishing the diagnosis. Other investigation tools, mainly USG and FNAC may aid in diagnosis. In symptomatic and hypothyroid cases a trial of levothyroxine can solve the problem otherwise surgical intervention is necessary followed by life-long thyroid hormone replacement therapy.

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