Thyroglossal duct cyst: Variable presentations

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Abstract Background: Thyroglossal duct cysts are the most common cause of midline congenital cyst formation in the neck that may present at any age. Classically, it presents as an anterior midline neck swelling that moves with deglutition and protrusion of the tongue. Occasionally, thyroglossal duct cyst presents in atypical manner either clinically or radiologically, which may pose a diagnostic challenge.

Objective: The aim of this study is to review cases diagnosed with thyroglossal duct cysts as regards clinical and radiological presentation, focusing on cases with atypical presentation.

Patients and methods: The medical records of patients who were diagnosed with thyroglossal duct cysts from January 2004 till October 2007 were retrospectively reviewed. A total of 22 patients were included in the study. They were classified as typical and atypical according to the clinical and radiological presentation.

Results: There were 10 males (45.5%) and 12 females (54.5%) with mean age of 17.3 years. The site of the cyst was infrahyoid in 20 cases (91%), suprahyoid in one case (4.5%) and intralingual in one case (4.5%). Clinically, 16 cases (72.7%) showed typical presentation and six cases (27.3%) were atypical in the form of: thyroglossal duct cyst with intralaryngeal extension, intralingual cyst,
1. Introduction

Thyroglossal duct cyst (TGDC) is the most common congenital anomaly of the neck in childhood, representing more than 75% of congenital midline neck masses. Although TGDCs often occur in pediatric patients, at least half are diagnosed in the second decade of life and they can also present later in adulthood. TGDCs originate from persistent epithelial remnants of the thyroglossal duct that are present during the descent of the thyroid gland from the foramen cecum to its final position in the anterior neck.¹

TGDC usually presents as a painless midline neck swelling that moves with deglutition and protrusion of the tongue with the typical radiological finding of a simple cystic swelling in the neck. Occasionally, TGDC can show atypical presentation either clinically or radiologically, which may pose a diagnostic challenge. Failure to anticipate the possibility of a TGDC may be associated with the performance of an inadequate surgical procedure such as simple incisional biopsy or enucleation, both of which are associated with significant recurrence rates.²

We report our experience with TGDCs regarding their clinical and radiological presentations with special emphasis on cases with atypical presentation.

2. Patients and methods

The medical records of all patients who were admitted to the Department of Otorhinolaryngology, Alexandria Faculty of Medicine, Egypt, with the diagnosis of TGDC during the period from January 2004 till October 2007 were retrospectively reviewed.

Charts were reviewed for demographic data, clinical presentation, available radiological assessment, and management plans. Cases were classified as typical and atypical according to the clinical and radiological presentation. TGDC was considered typical when it presented clinically as a painless midline neck swelling that moved with deglutition and protrusion of the tongue with the typical radiological appearance of a simple cystic swelling in the neck. Any variation in the clinical and/or radiological presentation was considered as atypical TGDC. Operative data and postoperative histopathological results were also assessed.

3. Results

Twenty two patients diagnosed as having TGDCs were included in the study. Twelve were females (54.5%) and 10 were males (45.5%). Their age ranged from 2 to 50 years with mean of 17.3 years and median age of 12.5 years. The site of the cyst was infrahyoid in 20 cases (91%), suprhyoid in one case (4.5%) and intralingual in one case (4.5%).

Clinically, 16 cases (72.7%) showed the typical presentation of a cyst in the anterior part of the neck moving with deglutition and protrusion of the tongue. Six cases (27.3%) showed atypical presentation.

Radiological evaluation included neck ultrasonography (USG) in 19 cases, computed tomography (CT) scan in four cases and magnetic resonance imaging (MRI) in four cases. USG showed the typical presentation of well defined, smooth, uniformly anechoic lesion with posterior acoustic enhancement in 17 cases. In one case, USG showed multiloculation with intracystic septae. In another case, it showed an intracystic solid component (Fig. 4a).

CT scan showed well circumscribed, low-density lesions with smooth, thin wall. Peripheral rim contrast enhancement was observed in two cases denoting previous infection. MRI was performed in four patients with atypical presentation. It showed homogenous hypointense signal on T1 weighted images and hyperintense signal in T2 weighted images suggestive of uncomplicated TGDCs. The diagnosis was confirmed in three cases by showing an upward tapering hyperintense tract extending towards the tongue base in T2 weighted images. In one case, it showed another downward tapering tract extending from the lower part of the cyst towards the thyroid gland.

4. Cases with atypical presentation

4.1. Case 1

A 50 year old male patient presented with neck swelling since 3 months. Examination revealed right paramedian cystic neck swelling that was mobile with deglutition and protrusion of the tongue. On routine laryngoscopic examination, there was fullness and medial displacement of the right aryepiglottic fold. CT scan showed right paramedian neck swelling extending to the preepiglottic space. A diagnosis of saccular cyst was proposed. T1 weighted MRI revealed hypointense cystic neck swelling extending to the preepiglottic space (Fig. 1a). T2 weighted MRI showed a short tract extending towards the tongue base which raised the suspicion of TGDC with intralaryngeal extension (Fig. 1b). Intraoperatively, the preepiglottic extension of the cyst and the upward tract were identified (Fig. 1c and d). Excision of the cyst with body of the hyoid bone and the tract up till the tongue base was performed. Postoperative histopathological
examination confirmed the diagnosis of TGDC with intralaryngeal extension.

4.2. Case 2

Male child aged 4 years presented with sleep apnea with attacks of stridor. Examination revealed a well circumscribed mass at the base of the tongue (Fig. 2c). MRI showed cystic lesion occupying the base of the tongue which was hypointense in T1 and hyperintense in T2 weighted image. It was protruding into the valeculla pushing the epiglottis posteriorly (Fig. 2a and b). Laser excision of the cyst with the surrounding muscles was performed (Fig. 2d). Histopathological examination confirmed the diagnosis of lingual TGDC.

4.3. Case 3

A 45 year old female patient presented with a tender oblong shaped anterior neck swelling extending from the level of the thyroid gland till the hyoid bone (Fig. 3c). She gave a history of anterior neck swelling that was present since childhood with a previous attack of acute severe neck pain associated with spasm of the neck muscles 2 years ago. She reported receiving medical treatment with relief of pain and neck spasm. MRI showed isointense T1, hyperintense T2 irregular shaped swelling surrounding the strap muscles with the characteristic upward tapering towards the tongue base (Fig. 3a and b). Neck exploration revealed amalgamated diffuse anterior neck swelling extending from the thyroid gland up till the hyoid bone (Fig. 3d). Dissection of the swelling was done till the level of the hyoid bone where the body was removed and the tract was followed up till the tongue base (Fig. 3e and f). Postoperative histopathological examination revealed chronic inflammatory cells, fibrous tissue, muscle and remnants of the thyroglossal cyst. The diagnosis of chronic neck inflammation following the rupture of a TGDC was proposed.

4.4. Case 4

Female patient aged 32 years presented with anterior neck swelling that moved with deglutition and protrusion of the tongue. USG revealed cystic swelling with large intracystic soft tissue shadow (Fig. 4a). USG of the thyroid gland revealed normal in-place thyroid gland. Ultrasound guided fine needle aspiration biopsy from the solid component revealed normal thyroid tissue. Excision of the cyst with its tract up till the tongue base was done including the body of the hyoid bone. The cut surface of the surgical specimen revealed a solid component within the cyst (Fig. 4b). Postoperative histopathological examination confirmed the diagnosis of TGDC with normal intracystic thyroid tissue.

4.5. Case 5

A 6 year old boy presented with a paramedian neck swelling at the level of the thyroid gland. It was mobile with swallowing but not with protrusion of the tongue (Fig. 5a). USG revealed cystic swelling at the level of the thyroid isthmus with normal appearance of both thyroid lobes (Fig. 5b). The diagnosis of thyroid isthmic cyst was proposed. The patient was euthyroid.
Fine needle aspiration revealed benign squamous cells and mucoid proteinaceous material suggestive of TGDC. T2 weighted MRI revealed a cystic swelling that showed the characteristic upward tapering of hyperintense tract extending to the tongue base (Fig. 5c). On intraoperative exploration, the cystic swelling was found to be separated from the thyroid gland and had a long tract extending to the tongue base (Fig. 5d). Histopathological examination confirmed the diagnosis of TGDC.

4.6. Case 6

A 34 year old male patient presented with a paramedian upper neck swelling that was mobile with deglutition and protrusion of the tongue (Fig. 6a). Its lateral location along the anterior border of the sternocleidomastoid raised the suspicion of a second branchial cleft cyst. However, the diagnosis of TGDC was confirmed intraoperatively by detecting a tract that extended upwards from the cyst till the tongue base. Another tract extending inferiorly towards the isthmus of the thyroid gland was identified (Fig. 6b). Sistrunk operation was performed and histopathological examination was consistent with the diagnosis of TGDC.

5. Discussion

TGDCs are the most common cause of midline congenital cyst formation in the neck.\textsuperscript{3} Though they may present at any age, the prevailing thought has been that the peak incidence is in the 1–10-year age group; however, recently it has been shown that TGDCs are more common in the adult population than previously believed. It has been demonstrated that TGDCs may have a bimodal distribution with peaks at 6 and 45 years of age.\textsuperscript{4} In our study, age ranged from 2 to 50 years with mean age of 17.3 years. Thirteen patients (59.1%) were below the age of 18 years.

While the classic presentation is of a midline anterior cervical cyst or mass that moves with deglutition and protrusion of the tongue, TGDCs may occasionally present in non-classic form. As regards the site, Allard collected 381 cases of well-documented TGDCs and reported the following locations: 2.1% lingual, 24.1% suprathyroid, 60.9% infrahyoid and 12.9% suprasternal.\textsuperscript{5} In this study, most of the cases (91%) were infrahyoid. Suprathyroid and lingual locations accounted for 4.5% each. No juxtahyoid or suprasternal cysts were reported. In one of the infrahyoid lesions, the cyst presented as a lateral neck swelling, its mobility with deglutition and protrusion of the tongue raised the suspicion of TGDC. Few reports in the literature described laterally placed TGDCs along the anterior border of the sternocleidomastoid muscle and included TGDC in the differential diagnosis of cysts originating from branchial clefts. The presence of a medial tail like component of TGDC that ‘dives’ into the hyoid bone, that can be detected by preoperative MRI or intraoperatively, will differentiate it from the second branchial cleft cyst.\textsuperscript{6–8}
Lingual TGDCs are rare. In a series of 300 TGDCs treated during a 29-year period, only two cases (0.67%) were in the region of the foramen cecum. The low incidence of lingual TGDC may be related to the fact that the duct initially atrophies from the oral side, where thyroid descent first begins. This low incidence makes overall experience with these lesions uncommon. Lingual TGDCs are primarily posterior; however, anterior location was previously reported. It accounts for 2.1% of intralingual TGDCs. Large lingual TGDCs on the base of the tongue might cause severe airway obstruction by a mass effect on the hypopharynx and by backward displacement of the epiglottis. This mechanism has been described as a ball valve effect between the cyst and laryngeal inlet. The location of these cysts often contributes not only to the development of respiratory symptoms such as stridor, dyspnea, raspy respiration and periodic cyanosis, but it can also cause sudden infant death. Clinical examination supplemented with flexible fiberoptic nasopharyngoscopy is of vital importance in the diagnosis of lingual TGDC. Imaging is required to confirm the diagnosis and to evaluate the airway before surgical intervention. Our case of lingual TGDC cyst was posterior. MRI showed the criteria of cystic swelling at the base of the tongue which was hypointense in T1 and hyperintense in T2 weighted images. It was protruding into the vallecula pushing the epiglottis posteriorly. Differential diagnosis includes dermoid cyst and vallecular cyst. As regards vallecular

Figure 3  Ruptured TGDC with subsequent chronic inflammation. (a) T1-weighted sagittal image showing isointense diffuse anterior neck mass (asterisk) surrounding the strap muscles. (b) T2-weighted sagittal image showing hyperintense irregular mass in the anterior part of the neck with hyperintense tract (arrow) extending towards the tongue base. (c) Preoperative view showing oblong shaped swelling (arrows) along the midline of the neck. (d) Intraoperative view showing diffuse irregular amalgamated mass at the anterior part of the neck. (e) Intraoperative view after dissection of the mass up till the tongue base. Remnant of the cyst was seen within the fibrous tissue (asterisk). (f) The resected specimen with the body of the hyoid bone.
cyst, it is a retention submucosal cyst that bulges between the tongue base and the free anterior margin of the epiglottis and not extending to the tongue muscles.14 Dermoid cyst is usually located above the geniohyoid muscles presenting at the floor of mouth and rarely occurs in the intralingual area.15,16 MRI of a dermoid cyst shows a heterogeneous, multiloculated cyst with components similar in intensity to fat as a result of sebum or fat in the cyst. These components are hyperintense on T1-weighted MR images.17 Although Sistrunk procedure is still the traditional management for TGDC, endoscopic CO2 laser surgery is an alternative for endogenous TGDCs without any projecting neck masses.18 However, meticulous examination of the preoperative MRI is crucial to rule out cervical extension of the cyst or the presence of any caudal ductal remnant that will require combined intraoral and cervical approaches. Some authors believe that simple marsupialization of lingual TGDCs provides excellent and definitive treatment reserving formal Sistrunk operation for recurrent cases.19 Newborns with lingual TGDC require special attention. They have low tolerance ability for surgery and their oral cavities are small and narrow. Therefore, total surgical removal may be difficult. Puncture method was advocated as a simple and effective method for initial debulking to be followed by close follow up till they are old and fit enough to withstand definitive surgical excision.19 Despite close relation of TGDC to laryngeal structures, a TGDC with intralaryngeal invasion mimicking an intralaryngeal mass is an extremely rare condition and only 10 cases have been reported in the literature.20 Symptoms of hoarseness, dyspnea, and dysphagia should make one consider intralaryngeal extension of TDC. Slotnik et al. reported three cases of laryngeal invasion in a series of 21 patients with TGDCs.21 Generally the intralaryngeal extension of thyroglossal cysts is seen as a secondary phenomenon resulting from massive enlargement over a long period of time.22 However, Lübben et al. reported a 62 yr old male patient having TGDC with intralaryngeal extension.23 The patient presented with change of voice for 6 months followed by appearance of painless neck swelling. They suggested that the cyst was primarily intralaryngeal in the preepiglottic space and extended to the neck. The primary origin from the preepiglottic space was explained by the fact that during embryological development the hyoid bone rotates to assume its normal position, pulling the thyroglossal tract with it posteriorly and cranially. Therefore, remnants of the tract might stay deep posterior and inferior to the hyoid bone within the preepiglottic space.24 In our case, it is likely that the cyst was primarily in the neck and then extended to the preepiglottic space based on the following findings: (1) the patient had no laryngeal symptoms or signs, and (2) the extralaryngeal portion of the cyst is larger than the intralaryngeal portion. Differentiation from saccular cyst was based on the identification of a small tract extending to the tongue base in T2 weighted MRI. Metabolic dissection of the cyst from the laryngeal mucosa is of utmost importance to decrease the likelihood of incomplete surgical excision or iatrogenic entry into the airway that may result in postoperative surgical emphysema. If entry in the airway occurred during surgery, reconstruction of the laryngeal framework by the local strap muscles can solve the problem.25

As the thyroglossal remnants remain connected to the tongue base by a tract, there is risk of being infected, with or without abscess formation which could be the initial presentation of TGDC. Liu et al. reported signs of inflammation, either acute or chronic, in 48 percent of resected specimens of TGDCs.26 Thyroglossal cyst abscess might discharge to the skin of the neck either spontaneously or following an inappropriate surgical incision resulting in a fistulous tract. Alternatively, it might discharge via a sinus tract to the tongue base resulting in resolution with a residual fibrous remnant. A true cyst will remain or develop only if an intact capsule forms. Without a capsule, or full resolution, a chronically infected duct remains.26,27 Our case of infected TGDC is possibly a ruptured cyst into the anterior neck compartment with incomplete resolution leaving a chronically inflamed duct.

Classically, TGDC is mobile with deglutition and protrusion of the tongue. In one of our cases, the cyst was mobile with deglutition but not with tongue protrusion. The diagnosis of an isthmic cyst was proposed. On USG, an intrathyroid TGDC appears as a well-defined anechoic cystic lesion and is indistinguishable from other benign cysts within the thyroid gland, including intrathyroidal lymphoepithelial cyst or branchial cleft cyst.28 However, fine needle aspiration results raised the suspicion of TGDC. The finding on MRI of a large cyst with the characteristic upward tapering and a hyperintense tract extending to the tongue base on T2 weighted images supported the preoperative diagnosis of TGDC. Less than 10 cases of intrathyroidal TGDCs were reported in the literature. Loss of mobility with tongue protrusion may be explained by the presence of a long tract interfering with its mobility or due to adhesion to the thyroid gland. Diagnosis of TGDC is mainly clinically. On reviewing the literature, it becomes clear that there is no consensus regarding preoperative imaging in patients with TGDCs. In most instances, such decision making relies on common sense and on the surgeon's previous experience. However, there is a general agreement that high resolution USG remains the ideal initial investigation of choice, particularly in children, as it does not involve ionizing radiation or sedation, is readily available, inexpensive and provides the surgeon with the necessary pre-operative information.29,30 TGDCs have previously been described by ultrason as well defined, smooth, and uniformly anechoic with posterior enhancement.30 The thyroid gland can also be examined during the same sonographic study. According to several studies, the demonstration of a thyroid gland with normal echogenicity, contour, site and location in a patient without clinical or laboratory evidence of hypothyroidism should

Figure 4 TGDC with solid intracystic component. (a) USG showing a cyst (asterisk) at the anterior part of the neck with large intracystic solid component, (b) postoperative view after excision and exploration of the cyst showing the solid component.
be sufficient to exclude an only functioning ectopic thyroid tissue within the TGDC.\textsuperscript{31} Thereby, obviates the need for radiouclide scanning. However, USG was not able to visualize a tract extending towards the tongue base in any of our cases.

Figure 5  TGDC mimicking thyroid isthmus cyst. (a) Preoperative view showing an infrahyoid midline neck swelling (arrow) which was mobile with swallowing but not with tongue protrusion. (b) US showing a cystic swelling within the thyroid isthmus. (c) T2-weighted sagittal image showing hyperintense anterior neck swelling with hyperintense tract (arrow) extending towards the tongue base. (d) Intraoperative view showing the cyst (C) with a long tract (arrow) extending towards the tongue base.

Figure 6  Laterally located TGDC. (a) Preoperative view showing left lateral neck swelling (arrow) which was mobile with deglutition and protrusion of the tongue. (b) Intraoperative view showing the cyst (C) which is related to the hyoid bone (HB) with another tract (arrow) extending inferiorly towards the thyroid gland.
Similarly, in a study done by Ahuja et al., on 23 patients, USG did not demonstrate the tract in any case. CT scan is rarely justified during the preoperative assessment of a presumed TGDC. However, if it is performed, a TGD cyst is seen as a well circumscribed, low-density lesion with a smooth, thin wall. Rim contrast enhancement may also be observed. Again CT scan was not able to demonstrate a tract in any of our cases. MRI was performed in four cases. It showed the typical appearance of a simple cystic swelling which was hypointense in T1 weighted image and hyperintense in T2 weighted image. However, this appearance is not constant as TGDCs with high protein content and those with previous episodes of infection or hemorrhage will display a high signal intensity on T1 weighted MRI. USG did not demonstrate the tract in any case. MRI was performed in four cases. Several authors and should be considered during planning of surgical resection to avoid recurrence.

In conclusion, TGDCs are the most common cause of midline congenital cyst formation in the neck that may present at any age. Clinically, it presents as an anterior midline neck swelling that is mobile with deglutition and protrusion of the tongue. However, non-classic presentation is not uncommon. Variability in the site, clinical presentation and radiological appearance must be anticipated. Although USG is the ideal method for investigation, MRI T2-weighted image is the only sure imaging diagnostic modality as it can visualize the tract ascending to the tongue base and sometimes, multiple or arborized tracts can be also identified. Therefore, MRI should be considered in the diagnostic workup of TGDCs especially in cases with atypical presentation.

References