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CASE REPORT Open Access

A rare cause of dysphagia due to retropharyngeal foregut duplication cyst: case report and review of the literature

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Abstract

Background The retropharyngeal space is a deep compartment of the head and neck region which extends from the base of the skull to the mediastinum, between the posterior pharyngeal wall and vertebral muscles, delimited laterally by carotid sheaths. Pathological processes of the retropharyngeal space are rarely encountered, generally are isolated and painless masses often cystic, and they usually originate from branchial arch anomalies, but only in rare cases, they turn out to be foregut duplication cysts. Foregut duplication cyst is rare congenital malformations arising along primitively derived alimentary tract during the first trimester in the developing embryo, mostly seen in the thorax and abdomen, with just few cases reported in the head and neck region. We report an extremely rare case of a foregut duplication cyst lined with respiratory epithelium located in the retropharyngeal space, at the level of the oropharynx, of an adult patient with dysphagia surgically treated, and we also made an analysis of the published literature about this very uncommon condition.

Case presentation A 63-year-old male patient with chronic dysphagia was diagnosed with a retropharyngeal cystic lesion, which was surgically treated. Final pathologic evaluation confirmed the diagnosis of a rare foregut duplication cyst lined with respiratory epithelium. A review of the pertaining published literature about the head and neck foregut duplication cysts was made, with a particular emphasis on retropharyngeal ones.

Conclusions Retropharyngeal foregut duplication cysts are a very rare congenital cyst of the head and neck. Clinical symptoms such as dysphagia and dyspnea should be evaluated with fibrolaryngoscopy, and CT and MRI scans are of great significance for definitive diagnosis, which should include the possibility of a foregut duplication cyst in the differential diagnosis. Surgical excision is the elective treatment for this lesion, in order to prevent complications including infection and compression symptoms or eventually malignant transformation.

Keywords Dysphagia, Foregut duplication cyst, Retropharyngeal, Transoral surgery, Case report

Background

The retropharyngeal space is a deep compartment of the head and neck region which extends from the clivus to the upper mediastinum, posterior to the pharynx and esophagus, and anterior to the prevertebral musculature, laterally delimited by carotid sheaths [1, 2]. It is bounded by the buccopharyngeal fascia anteriorly, the prevertebral fascia posteriorly, and the thin alar fascia, a part of the deep layer of the deep cervical fascia, which extends from the medial border of the carotid space on either side and divides the retropharyngeal space into 2 components: the

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anteriorly proper retropharyngeal space and the posteriorly situated "danger" space, which extends inferiorly into the posterior mediastinum to the level of the diaphragm and is named as such because it provides a conduit for spread of infection from the pharynx to the mediastinum. The main components of the retropharyngeal space are areolar fat and lymphatic tissue, with lymph nodes draining the pharynx, nasal cavity, paranasal sinuses, and middle ears [3].

This posterior compartment could be involved by pathological processes which are rarely encountered, especially inflammatory and neoplastic disease, but generally are isolated and painless cystic lesions, and among them, branchial cysts are the most frequent [4], but only in very rare cases they turn out to be foregut duplication cysts.

Foregut duplication cysts are benign congenital malformations which can arise along the primitively derived alimentary tract during the first trimester of embryonal development [5–7]. Although the exact pathogenesis still remain unclear, the hypothesis generally accepted suggests that it is most likely a congenital abnormality that arises from the embryonic development of the foregut, which in the 4th week divides into a ventral part, containing endoderm cells, which leads to the development of the laryngo-tracheo-bronchopulmonary tree, and a dorsal part that becomes the proximal gastrointestinal tract. In this period of differentiation, embryonal heterotrophic germs of foregut, with their native epithelium, could migrate and be entrapped in the wall of pharyngeal arches, forming duplication cysts [8–10]. According to Veeneklaas, abnormal notochord adherence to the primordial gut endoderm may have a role in the pathogenesis of foregut duplication cyst [11].

Most foregut duplication cysts are found in the thorax or abdomen, and they account for one-third of all duplication cysts of the alimentary tract cysts, with an overall incidence rate of 1 in 4500. Head and neck localization is very rare and comprises only 0.3% of cases, usually occurring in children; most cases of foregut duplication cysts previously described occurred in oral cavity, especially the tongue, with very few cases described in the pharynx, so due to the low incidence rate, the existing literature on head and neck foregut duplication cysts consists mostly of single case reports [12].

Foregut duplication cysts are mainly symptomatic according to the site, size, and occurrence of infection, and generally presented by swelling, deglutition disorders and dyspnea, accounting for the execution of endoscopic study for airway evaluation [13]. Radiological study, with CT and MRI, is mandatory for better preoperative planning to reveal the extent and the relationship between the cyst and the adjacent structures [14]. Distinguishing them from other cervical cystic lesions such as thyroglossal duct, branchial cleft cysts, dermoid cyst, cystic hygroma, and metastatic cervical lymph nodes is difficult preoperatively due to their similar radiological features, with rates of misdiagnosis high. Definitive diagnosis mainly depends on intraoperative findings and postoperative pathology [15].

Case presentation

We present the clinical case of a 63-year-old man, who came at our attention in October 2022 complaining a globus pharyngeus and dysphagia for several years, with worsening in the last 2 months. His pathological anamnesis revealed that he was affected by granulomatosis with polyangiitis. On clinical examination of the neck, no mass could be palpated. Oropharyngoscopy revealed, confirmed by fibrolaryngoscopy, a mass from the posterior pharyngeal wall expanding on the right side, with nonpathological mucosa on the surface, in front of the base of the tongue and the tip of epiglottis (Fig. 1). Routine blood tests were normal. The patient underwent preoperative radiological assessment, including contrast-enhanced computed tomography (CT) and magnetic resonance imaging (MRI) of the neck. Contrast-enhanced CT scan showed well-defined mass on the right side of the posterior pharyngeal wall, caudal to the ipsilateral palatine tonsil, not enhanced with contrast. MRI scan with gadolinium demonstrated enhancement on T2-weighted images, supposing cystic nature of mass, due to mucinous and proteinaceous debris. The mass was well-delineated into the retropharyngeal space expanding on the right side, in close contact with the carotid sheath (Fig. 2).

Due to the localization of the cyst, at the level of the posterior wall of the oropharynx, transoral surgical removal was planned. The pharyngeal mucosal wall was cut using electric monopolar cutting knife, exposing the cystic mass. During surgical maneuvers, the cystic wall was accidentally ruptured with leaking of mucinous material, and then the cyst was totally removed. The surgical breach was closed with a barbed suture technique (Fig. 3).

Histopathological examination confirmed the cystic nature of the mass and showed pseudo-stratified ciliated columnar epithelium lining the cystic capsule and surrounding muscle of the pharynx, suggestive for a foregut duplication cyst (Fig. 4). Clinical examination after 1 week revealed no swelling of the pharyngeal wall, and the patient's initial symptoms had completely disappeared.

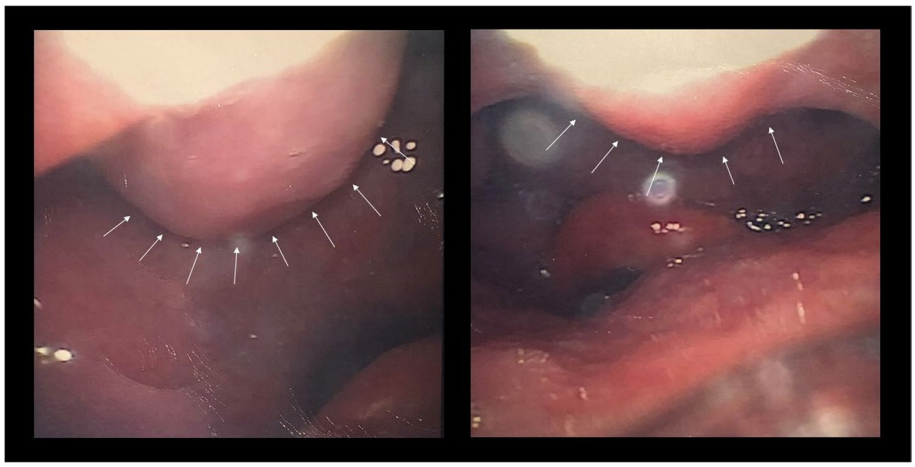

Fig. 1 Fibrolaryngoscopy showed oropharyngeal mass from the posterior wall (white arrows) at the level of the base of the tongue and the tip of the epiglottis

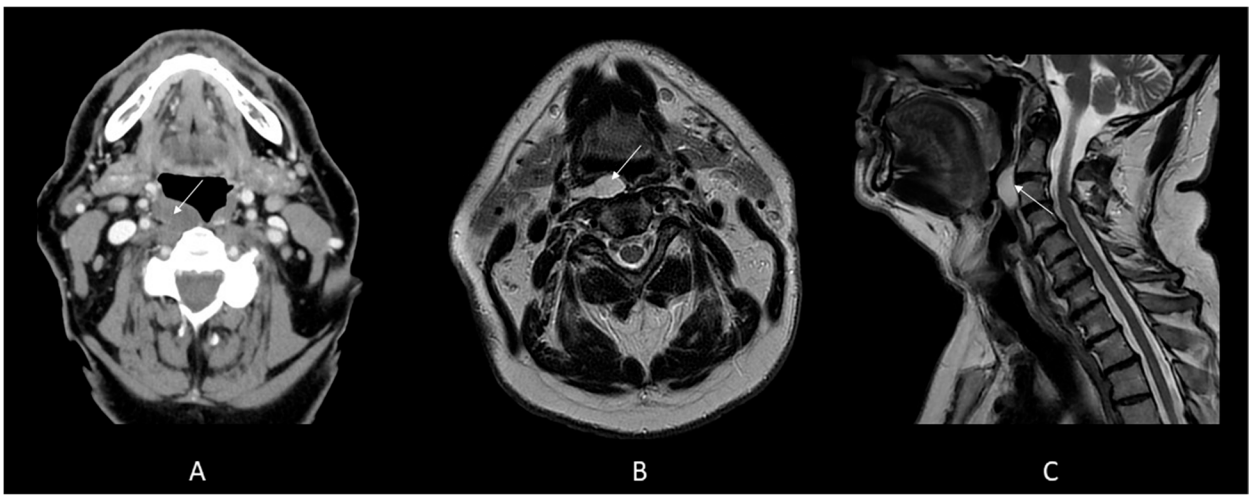

Fig. 2 Contrast-enhanced CT scan imaging of the neck: encapsulated mass (white arrow) occupying the retropharyngeal space, expanding on the right side, in close contact with carotid sheath (**A**). MRI axial and sagittal scans with gadolinium showed a retropharyngeal lesion with uniform high signal intensity in T2-weighted images (**B** and **C**). Note that the mass, arising from the retropharyngeal space, causes anterior displacement of the pharyngeal mucosa, leading to the pharyngeal globus sensation. CT, computed tomography. MRI, magnetic resonance imaging

Discussion

Foregut duplication cysts are uncommon developmental anomalies of the embryonal primitive alimentary tract, and they rarely develop in head and neck region [16]. These lesions are usually diagnosed in the pediatric population and are very rare in adults. The first case of esophageal duplication cyst was reported in 1711 by Balasius [17], and in published literature, approximately 70 cases of foregut duplication cysts in the head and neck region have been reported so far, almost totally in oral cavity; in particular, only 2 cases of retropharyngeal foregut duplication cysts have been described [18–20]. To date, the age of presentation was 3 years old and 30 years old, with no gender predilection. The patients were treated by complete excision of the cyst, and no recurrence was reported (Table 1).

Cysts localized in head and neck region may cause globus pharyngeus, dysphagia, or respiratory distress in some individuals, and if suprainfection occurs, worsening of the symptoms may result with also external drainage of purulent material. Clinical symptoms such as dysphagia and dyspnea should be evaluated with fibrolaryngoscopy, and CT and MRI scans are of great significance for definitive diagnosis, which should include the

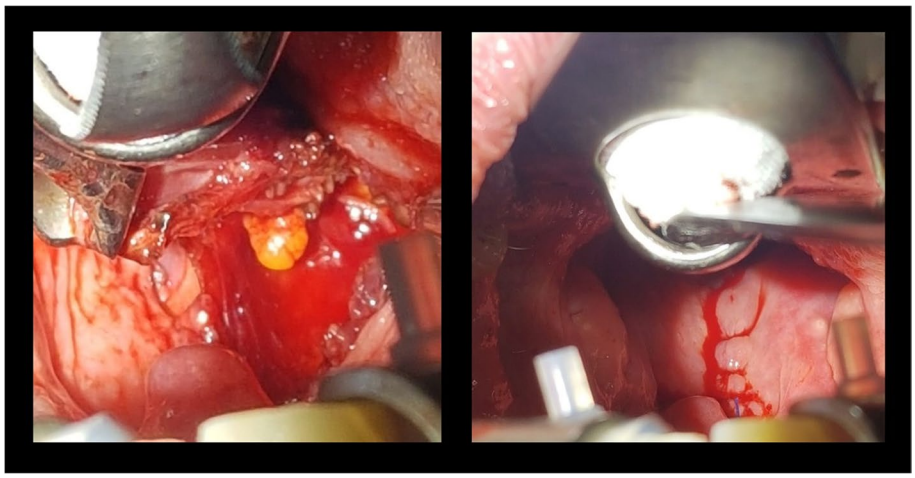

Fig. 3 Intraoperative picture after the removal of the cyst, fatty areolar tissue of the retropharyngeal space is exposed. Mucosal wound was closed with barbed suture technique

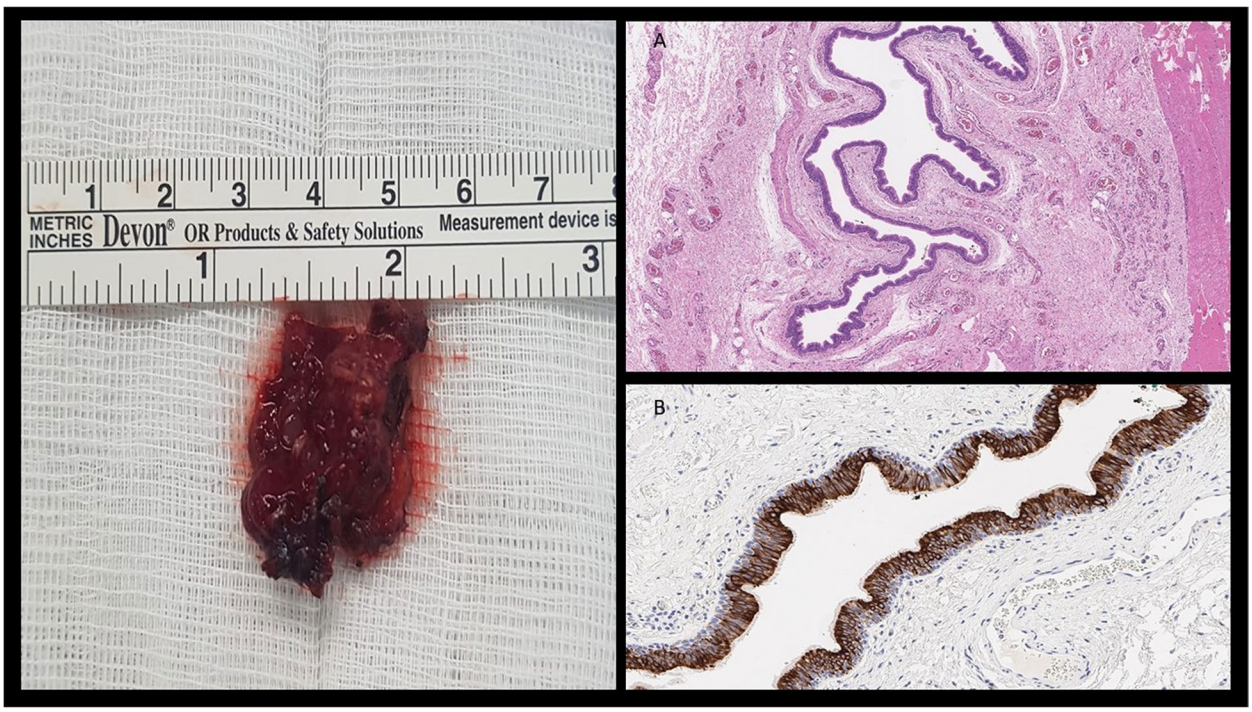

Fig. 4 The mass was analyzed by an expert head and neck pathologist, who confirmed the cystic nature of the lesion, lined by pseudostratified columnar-ciliated respiratory epithelium without any cartilage or mucinous glands, confirming the diagnosis of foregut duplication cyst lined with respiratory epithelium (H&E stain 4× (**A**) and immunohistochemistry using CD7 as respiratory tract marker, 20× (**B**))

possibility of foregut duplication cyst in the differential diagnosis [21, 22].

The differential diagnosis for cervical foregut duplication cyst includes branchial cleft cyst, thyroglossal duct cyst, thymic and thyroid cysts, cystic hygroma, dermoid cyst, lymphangiomas, teratoma, and also metastatic lymph node. It could be very difficult to differentiate the cyst from metastatic lymph node or other cystic lesions preoperatively and intraoperatively as well, so histopathological confirmation is essential for the diagnosis as there are no specific clinical or radiological criteria for differentiation from other lesions, especially cystic lesions. Traditionally, Rickham et al. established three diagnostic criteria that must be met to make a diagnosis

Table 1 Summary of published case series with retropharyngeal foregut duplication cyst: demographic information, clinical characteristics at presentation, and treatment

Series (author, year, reference)	No. of cases, sex	Age at onset (years)	Symptoms	Site of lesion	Treatment/recurrence
Roddie (1960) [19]	1 F	30	Globus pharyngeus	Right posterolateral pharyngeal wall (retropharyngeal space)	Aspiration (recurrence 12 months later and definitive transoral surgical excision)
Kieran et al. (2010) [20]	1 M	3	Loud snoring	Left posterolateral pharyngeal wall (retropharyngeal space)	Transoral surgical excision

of foregut duplication cyst; it must be covered by a smooth muscle coat, contain epithelium derived from the foregut, and be attached to a portion of the foregut [23]. Duplication cysts may be lined by 1 or more types of epithelium: ciliated respiratory-type epithelium, stratified squamous epithelium, and gastric or intestinal mucosa, so based on their epithelial type and other features, foregut duplication cysts may appear to closely resemble airway or digestive tracts [24]. Therefore, the term foregut duplication cyst includes bronchogenic cyst, esophageal duplication cyst, and enteric duplication cyst [20].

In our particular case, we described a unique case of retropharyngeal foregut duplication cyst, the third case reported in the scientific literature, in head and neck region, bulging in the oropharyngeal wall, where the presence of the respiratory epithelium lining the wall of the cyst promoted the diagnosis of foregut duplication cyst and differentiated it from the uncommon parapharyngeal/retropharyngeal bronchogenic cyst, lined with respiratory epithelium, with the presence of cartilage, seromucous glands, and smooth muscle, suggesting bronchial origin [20].

Patients with an intramural pharyngeal foregut duplication cyst could be asymptomatic, but symptomatic cyst should be surgically removed. Cyst enlargement, infection in the cyst, hemorrhaging in surrounding tissues, may compress adjacent organs and eventually determine dysphagia and airway obstruction caused by cyst hyperplasia. The size and location of the lesion and its relationship with the surrounding organs will determine the specific operation procedure, but it will usually involve the complete excision of the cyst so as to avoid recurrence and other complications, such as malignant transformation [21]. Carcinomas arising from foregut duplication cysts have been reported in the literature, emphasizing the importance of total surgical excision since recurrence has been reported after incomplete resection [25, 26]. After surgery, clinical and radiological follow-ups should be carried out regularly to prevent recurrence [27].

Conclusions

Foregut duplication cyst is a benign malformation rarely located in the head and neck region. In extraordinary cases, it could be located in the parapharyngeal or retropharyngeal space. The impact on clinical symptoms depends on the location and size of the cyst, so endoscopic exploration of upper airways and CT and MRI scanning is of great significance for differential diagnosis and for surgical planning, especially in children because cyst enlargement can rapidly lead to obstruction and severe respiratory failure. Surgery is the most effective way to treat a foregut duplication cyst, and follow-ups should be carried out regularly to prevent cyst recurrence.

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Code availability
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Authors' contributions
All authors contributed to the study conception and design. Material preparation, data collection, and analysis were performed by GC, PDA, and PA. GM supervised the study and approved the final manuscript.

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Availability of data and materials
Availability of data and material is possible upon reasonable request, deidentified for maintenance of anonymity.

Declarations

Ethics approval and consent to participate
All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards. The ethics committee for clinical trials of the Provinces of Verona and Rovigo exempted this study from its approval. Informed consent was obtained, written, from all individual participants included in the study.

Consent for publication
Consent for publication was obtained, written, for every individual person's data included in the study.

Competing interests
The authors declare that they have no competing interests.

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