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Clinical Record

Foregut duplication cyst of the pharynx

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Abstract

Introduction: Foregut duplication cysts are heterotrophic rests of foregut-derived epithelium which are usually found in the abdomen and thorax; rarely are they found in the head and neck.

Case report: We describe the case of a pharyngeal foregut duplication cyst presenting with airway obstruction in a neonate. We also review the pathology, investigation and management of this rare condition.

Discussion: The occurrence of a foregut duplication cyst in the head and neck region mandates vigilance with respect to the airway. Magnetic resonance imaging is a useful part of pre-operative evaluation but cannot be relied upon for definitive diagnosis. Although foregut duplication cysts are benign lesions, definitive cure ultimately requires surgical excision, and this is often the means by which a definitive diagnosis is made. The prognosis for these lesions is excellent, with no reports in the literature of recurrence following excision.

Key words: Congenital Defects; Cyst; Pharynx; Airway Obstruction

Introduction

Foregut duplication cysts are heterotrophic rests of foregut-derived epithelium which are usually found in the abdomen and thorax; rarely are they found in the head and neck.

We describe the case of a pharyngeal foregut duplication cyst presenting with airway obstruction in a neonate. We also review the pathology and management of this rare condition.

Case report

A 15-day-old female neonate was referred to the ear, nose and throat clinic with a three-day history of right submandibular swelling. The child had been born at term by spontaneous vaginal delivery and had no feeding or breathing difficulties.

Ultrasonography of the lesion confirmed a multicystic mass measuring 25 × 25 × 23 mm and containing echogenic debris. The differential diagnosis was either a cystic hygroma or branchial cyst with associated haemorrhage or infection.

The child was initially treated with a course of antibiotics, which appeared to settle the swelling over the next week.

At 28 days of age, the child was readmitted to hospital with respiratory distress related to a sudden increase in size of the mass. Examination revealed significant right-sided oropharyngeal swelling, causing supero-lateral deviation of the tongue. The child was intubated pre-emptively to prevent impending airway obstruction and then transferred to the operating theatre.

Direct laryngoscopy revealed a normal larynx and a supraglottic swelling involving the lateral wall of the pharynx and tongue. The cyst was aspirated; microscopy of the purulent fluid showed numerous white cells and coagulase-negative staphylococci.

Despite antibiotic therapy, the cyst reaccumulated over the next two days. Magnetic resonance imaging (MRI) of the neck confirmed a complex, multicystic structure lying anteromedial to the right sternomastoid muscle and extending from the retromandibular space down to the level of C5. The lesion was seen to be causing a considerable mass effect, compressing the larynx and displacing the tongue and oropharynx. The child was taken back to theatre for full excision of the lesion.

At operation, the appearance of the lesion was unusual, with both solid and cystic elements and a significant amount of surrounding soft tissue inflammation and oedema. Post-operatively, after a short stay in the intensive care unit, the child was extubated and later discharged home successfully.

Histopathological analysis of the resected specimen revealed a foregut duplication cyst. Sections of the multicystic lesion, which measured 35 × 25 × 17 mm, were seen to have both solid and cystic components exhibiting specific forms of differentiation (Figure 1). The solid elements included mucin-predominant salivary gland and pancreatic exocrine and endocrine tissue, with positive immunohistochemical staining for insulin (Figure 2) and glucagon. The cystic components comprised a range of different epithelial-lined structures of varying morphological complexity, including mucin-secreting gastric epithelium and small intestinal villous mucosa with underlying submucosa, muscularis mucosae and muscularis propria. The lesion had been fully excised.

The child was reviewed six months post-operatively and had no evidence of recurrence.

Discussion

The embryological foregut gives rise to the pharynx and its associated structures, as well as to the lower respiratory tract, oesophagus, stomach, duodenum and hepatobiliary

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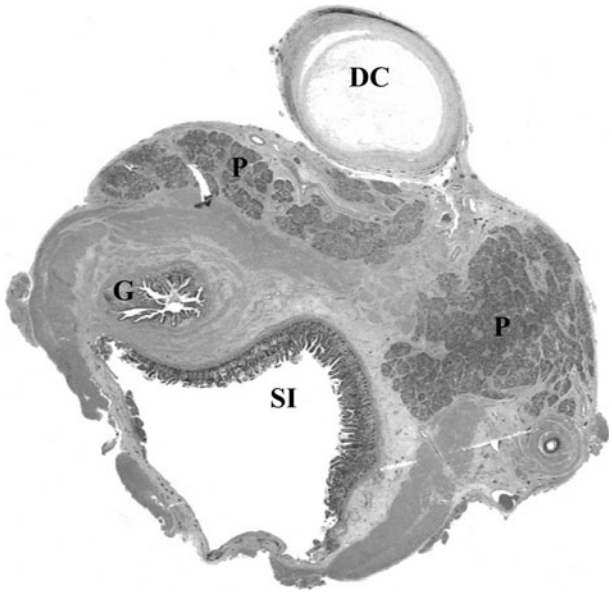

FIG. 1

Photomicrograph of operative specimen demonstrating a variety of foregut-derived epithelial structures within the cyst. DC = degenerate cyst; P = pancreatic tissue; G = gastric mucosa (seen with surrounding muscularis mucosae and muscularis propria); SI = small intestinal villous mucosa (H&E; ×25).

tract. Rarely, heterotrophic rests of foregut-derived epithelium cause duplications of these structures and form what is known as a foregut duplication cyst. Alimentary tract duplications can occur anywhere from mouth to anus; foregut duplication cysts are the least common of these, accounting for only 33 per cent of all cases.[1] Foregut duplication cysts are most commonly found within the thorax, followed by the abdomen;[2] rarely are they found in the head and neck. Most cases of foregut duplication cysts described previously within the head and neck have involved the tongue; very few have been found in the pharynx.[3,4]

Foregut duplication cysts have been previously known as cystic choristomas, heterotrophic gastrointestinal cysts and eterocystomas. Rickham *et al.*[5] established three diagnostic criteria for foregut duplication cysts; the cyst must be: covered by a smooth muscle coat; contain epithelium derived from the foregut; and be attached to a portion of the foregut. The epithelium may be gastric, intestinal, respiratory or pancreatic, and varying numbers of parietal, goblet, argentaffin and Paneth cells are seen.[6] The mucosa observed in the cyst may not necessarily correspond to the normal gastrointestinal mucosa found at the anatomical level of the cyst, and mixed mucosal types within a single cyst are common.[7]

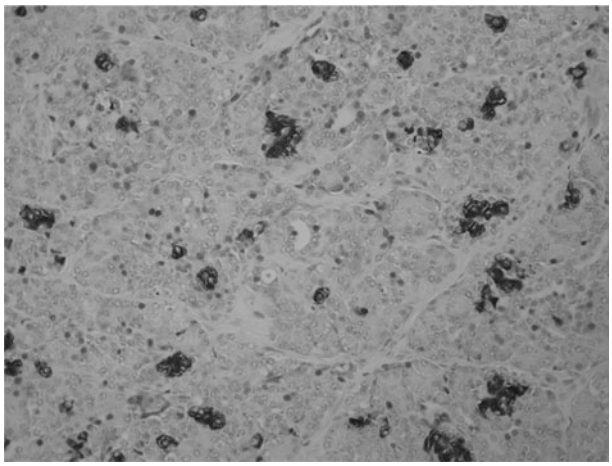

FIG. 2

Photomicrograph showing pancreatic tissue staining positive for insulin on immunohistochemical staining (×250).

Foregut duplication cysts have not yet been attributed to a single definitive embryological process, although several mechanisms have been proposed. In 1970, Gorlin and Jirasek[6] suggested that foregut duplication cysts in the tongue may develop following the misplacement of embryonal rests of gastric mucosa during embryological development. However, this theory fails to explain the presence of intestinal or pancreatic tissue in these cysts.

In 1952, Veeneklaas[8] proposed the currently accepted theory of abnormal notochord adherence. During normal embryogenesis, the notochord fuses with the primordial gut endoderm and then separates to form a rod-like structure that regresses to form the nucleus pulposus. Veeneklaas proposed that if the notochord remained adherent to the oesophagus, the distracted part of the oesophagus might separate and therefore form a cyst or duplication. This same theory of abnormal notochord adherence applies to other sites in the foregut as well.

Alternatively, the split notochord theory suggests that abnormal adherence of endoderm to neural tube derived ectoderm, and subsequent herniation through a sagitally split notochord, leads to a posterior neuroenteric fistula. This fistula, if not obliterated, leads to the formation of duplication cysts or fistulae.[1]

Given such a variety of embryological explanations, it remains unclear whether foregut duplication cysts represent a spectrum of lesions with similar but not identical origin, rather than one homogenous group.

Most foregut duplication cysts of the head and neck are diagnosed in asymptomatic neonates, although they have the potential to go undetected for years if the cyst is small.[9] As seen with our patient, foregut duplication cysts within the head and neck can give rise to significant respiratory distress and feeding difficulties if the cyst enlarges rapidly or becomes infected.

Magnetic resonance imaging, with its lack of ionising radiation and superior soft tissue resolution, is the imaging study of choice.[7] Although MRI allows an accurate assessment of the extent of infiltration for pre-operative planning, foregut duplication cysts are often indistinguishable from dermoid cysts on MRI because of the presence of proteinaceous fluid.[7] Both appear as cystic lesions that do not enhance with intravenous contrast.

- **Foregut duplication cysts are rare in the head and neck**
- **They are usually diagnosed in asymptomatic neonates, but occasionally cysts remain undetected into adulthood**
- **Histologically, these lesions are benign, heterotrophic rests of foregut-derived epithelium with a smooth muscle coat, and are attached to a portion of the foregut**
- **Cysts can enlarge rapidly and present with airway obstruction**
- **Surgical excision is curative**

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The role of ultrasound is debatable. Although two cases of intraoral foregut duplication cyst have been detected in utero by prenatal ultrasound,[10] the ultrasonic appearance of foregut duplication cysts is extremely variable and is unreliable for diagnosis.

Although foregut duplication cysts are benign lesions, definitive cure ultimately requires surgical excision, and this is often the means by which a definitive diagnosis is made. The prognosis for these lesions is excellent, with no reports in the literature of recurrence following excision.

References

1 Qi BQ, Beasley SW, Williams AK. Evidence of a common pathogenesis of foregut duplications and esophageal atresia with tracheo-esophageal fistula. *Anat Rec* 2001; **264**:93–100
2 Azzie G, Beasley S. Diagnosis and treatment of foregut duplications. *Semin Pediatr Surg* 2003;**12**:46–54
3 Edwards J, Pearson S, Zalzal G. Foregut duplication cyst of the hypopharynx. *Arch Otolaryngol Head Neck Surg* 2005; **131**:1112–15
4 Frenkiel S, Remsen KA. Foregut duplication arising from the pharynx. *J Otolaryngol* 1990;**19**:279–81
5 Rickham PP, Lister J, Irving IM, eds. *Neonatal Surgery*. London: Butterworths, 1978
6 Gorlin RJ, Jirasek JE. Oral cysts containing gastric or intestinal mucosa: an unusual embryological accident or heterotopia. *Arch Otolaryngol* 1970;**91**: 594–7
7 Eaton D, Billings K, Timmons C, Booth T, Biavati JM. Congenital foregut duplication cysts of the anterior tongue. *Arch Otolaryngol Head Neck Surg* 2001;**127**: 1484–7
8 Veeneklaas GMH. Pathogenesis of intrathoracic gastrogenic cysts. *Am J Dis Child* 1952;**83**:500–7
9 Velchek FT, Klotz DH, Hill CH. Tongue lesions in children. *J Paediatr Surg* 1979;**14**:238–45
10 Chen MK, Gross E, Lobe TE. Perinatal management of enteric duplication cyst of the tongue. *Am J Perinatol* 1997;**14**:161–3

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