Rare disease

Isolated cystic cervical oesophageal duplication: a rare congenital anomaly

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Summary

Oesophageal duplication is a rare congenital anomaly. The cystic form is the usual presentation of the duplication, found predominantly in the lower third. Less than 20 cases of isolated cystic oesophageal duplication in the cervical region have been reported till date in the English literature. We are reporting this case because of its rarity.

BACKGROUND

Oesophageal duplication is a rare congenital anomaly with an estimated incidence of 1 in 8200,¹ representing about 10% of all foregut duplications.² The cystic form is the usual presentation, whereas the tubular type accounts for only 5–10% of cases.³ Cystic duplications are commonly found in the lower third (60%)⁴ and on the right side^{4–5} of the native oesophagus. Less than 20 cases of isolated cervical cystic oesophageal duplication have been reported till date in the English literature.^{6–7} We are reporting this case because of its rarity.

CASE PRESENTATION

A 2-year-old female child presented to us with a history of stridor for 6 months duration along with a swelling on the right side of the neck. Stridor was progressively worsening with increase in the size of the swelling. The swelling was aspirated multiple times by a local practitioner, revealing mucoid material with transient improvement in stridor. There was no history of coughing, choking, feeding difficulty or fever.

On examination, the swelling was soft, cystic, non-tender, non-compressible, mobile only side to side and free from the overlying skin.

INVESTIGATIONS

Ultrasonography (USG) of the neck showed a 2.8×1.6 cm well-circumscribed, thick-walled (3 mm) cystic lesion with internal echoes and posterior acoustic enhancement, medial to right common carotid artery and posterior to the right lobe of thyroid, reaching up to the thoracic inlet.

On contrast-enhanced (CE) CT of the neck and chest, a well-defined hypodense lesion (52–65 HU) measuring 5.2×2.8×2.2 cm was seen in the right tracheo-oesophageal groove, extending from the level of the thyroid gland up to the thoracic inlet. It was displacing the trachea and oesophagus to the left side and had a well-defined hyper-enhancing wall in its superior aspect. There were no septae, mural calcification, nodularity or intraspinal extension (figure 1). CECT abdomen showed no evidence of associated duplications.

DIFFERENTIAL DIAGNOSIS

The differential diagnosis of a cystic neck mass in a child includes branchial apparatus anomaly, cystic hygroma, thyroglossal duct cyst, cervical thymic cyst, cystic teratoma or a laryngocele.

TREATMENT

Excision was undertaken via a cervical incision. Intraoperatively, a cystic structure sharing a common wall with the cervical oesophagus was found (figure 2). This was excised completely and the oesophagus was closed over a nasogastric tube.

OUTCOME AND FOLLOW-UP

Feeding was started with the nasogastric tube on the second postoperative day. A contrast oesophagogram, done a week later, showed no evidence of leak or stricture. The child was gradually shifted to normal oral diet and discharged. She has been asymptomatic till now after 2 years of surgery.

The histopathological examination was suggestive of an oesophageal duplication cyst. The cyst wall showed a stratified squamous epithelium, with a subepithelium consisting of loose connective tissue and bundles of smooth muscles beneath and the muscularis propria was composed of skeletal muscles.

DISCUSSION

Several theories have been proposed to explain the occurrence of oesophageal duplications viz. (1) abortive twinning theory, (2) persistent embryological diverticula theory, (3) bronchopulmonary foregut malformation theory, (4) split notochord theory, (5) aberrant luminal recanalisation theory and (6) intrauterine vascular accident theory. Out of these, split notochord and aberrant luminal recanalisation are considered to be the most likely theories explaining oesophageal duplication.

The location and size of duplication determine the clinical presentation. Most oesophageal duplications present in the first year of life with respiratory problems, feeding

Figure 1 (A, B) Contrast-enhanced CT neck and chest showing a well-defined hypodense lesion (C) on the right side of the neck displacing the trachea (T) to the left side.

difficulties, neck swelling and failure to thrive. Our patient presented with stridor and neck swelling.

Most of the congenital cystic neck swellings can be differentiated after thorough history and examination only. Imaging begins with USG to know the cystic or solid nature, extent and relation with the surrounding structures.⁸ A well-defined cystic mass with an inner echogenic mucosal layer is seen in relation to the oesophagus in a patient with duplication. CT or MRI is the next investigation, usually indicated for deep and extensive lesions and when USG is inconclusive.⁸

Contrast oesophagogram can be done to delineate communication of the cyst with oesophagus. Endoscopic ultrasonography may be helpful in the diagnosis of intramural cysts.⁹ Endoscopic management, though more commonly used for tubular duplications, has been tried successfully in cystic type also.¹⁰ We did not perform an endoscopy in our patient.

Thoracic and cervical vertebral anomalies are the commonly associated malformations. Other duplications, non-rotation, annular pancreas and dural communication with the duplication have also been reported.

Complete surgical extirpation is the standard treatment, especially when symptoms occur. It has been argued that surgery should always be performed so as to avoid the risk of complications and malignant degeneration. Mucosa should be removed in patients where complete excision is not possible.

To be a true duplication, these lesions should have three characteristics: (1) they should be firmly attached to at least one point of the alimentary tract; (2) they should have a well-developed smooth muscle coat; and (3) the epithelial lining of the duplication should resemble some part of the alimentary tract.¹¹

Learning point

- Oesophageal duplications, although rare, should be included in the differential diagnosis of a child with neck swelling, airway difficulties and swallowing problems.

Figure 2 (A) Cystic structure in the neck in relation to the oesophagus. (B) Oesophageal lumen after complete excision of the cyst.

Competing interests None.

Patient consent Obtained.

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