Case Report

Desmoid Fibromatosis of the Oesophagus Creating an Oesophageal Diverticulum in a 2-year-old Girl

Divya Murli[1], Vasundhara Smriti[2,3], Subhash Yadav[2,4], Bhakti Trivedi[2,5], Sajid S. Qureshi[1,2]

[1]Department of Surgical Oncology, Division of Paediatric Surgical Oncology, Tata Memorial Hospital and Advanced Centre for Training Research and Education in Cancer, Tata Memorial Centre, Departments of [3]Radiology, [4]Pathology and [5]Anaesthesia, Tata Memorial Hospital and Advanced Centre for Training Research and Education in Cancer, Tata Memorial Centre, [2]Homi Bhabha National Institute, Mumbai, Maharashtra, India

Abstract

Extra-abdominal desmoid fibromatosis arising from the oesophagus and the contemporaneous traction diverticula due to an oesophageal tumour is extremely rare. We present this complex situation in a 2-year-old girl which posed a surgical challenge requiring simultaneous management of multiple pathologies. Surgery addressed both the entities and the presence of the diverticulum facilitated achieving negative surgical margins.

Keywords: Desmoid fibromatosis, oesophageal diverticulum, oesophagus

Introduction

The World Health Organization defines desmoid fibromatosis (DF) as a clonal fibroblastic proliferation that arises in deep soft tissues and is characterised by infiltrative growth and a tendency towards local recurrence with no ability to metastasise.[1] It accounts for 3.5% of all fibrous tumours and 0.03% of all neoplasms.[2,3] DF may occur at abdominal, intra-abdominal or extra-abdominal sites.[2,3] The oesophagus as an extra-abdominal site has been scarcely reported in the literature, especially in children.[4] Oesophageal diverticula (EDs), particularly mid-oesophageal, are exceedingly rare. They are typically traction diverticula caused due to adhesions with the inflammatory lymph nodes commonly seen in tuberculosis or histoplasmosis.[5] EDs secondary to oesophageal tumours are extremely rare and never reported in children.[6,7] We report a unique case of EDs secondary to DF of the oesophagus which was successfully managed by surgical resection.

Case Report

A 2-year-old girl presented with vomiting following intake of food for the past 5 months. The vomitus contained intact ingested food. The child was malnourished and weighed 8.9 kg. A barium swallow revealed a diverticulum arising from the mid-oesophagus along with a soft-tissue mass in the anterior mediastinum. A computed tomography scan showed an anterior mediastinal mass measuring 9.6 cm × 7.7 cm with a fistulous communication with the oesophagus [Figure 1]. Upper gastrointestinal endoscopy revealed a large-mouthed diverticulum about 8 cm from the cricopharynx with compression of the oesophageal lumen. The biopsy of the mediastinal mass was suggestive of a benign low-grade spindle cell lesion. On immunohistochemistry, the spindle cells showed nuclear positivity for beta-catenin [Figure 2]. The clinicoradiological picture supported by the pathological findings compelled for upfront surgery due to the presence of obstructive symptoms. A median sternotomy approached was utilised, and the mass was dissected from the adjacent structures easily. The mass was entrapping the tip of the oesophageal diverticulum, whereas the body and mouth were free of the tumour [Figure 3]. The mass was excised along with the diverticulum utilising a surgical stapler. The post-operative course was uneventful. The histopathology confirmed DF with infiltration of the oesophageal diverticulum and negative

Received: 27-08-2022 **Revised:** 27-08-2022 **Accepted:** 28-08-2023 **Available Online:** 09-08-2024

Access this article online

Quick Response Code:

Website: https://journals.lww.com/AJPS

DOI: 10.4103/ajps.ajps_120_22

Address for correspondence: Prof. Sajid S. Qureshi, Department of Surgical Oncology, Division of Paediatric Surgical Oncology, Tata Memorial Hospital and Advanced Centre for Training Research and Education in Cancer, Tata Memorial Centre, Ernest Borges Road, Parel, Mumbai - 400 012, Maharashtra, India.
E-mail: sajidshafiques@hotmail.com

This is an open access journal, and articles are distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 4.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as appropriate credit is given and the new creations are licensed under the identical terms.

For reprints contact: WKHLRPMedknow_reprints@wolterskluwer.com

How to cite this article: Murli D, Smriti V, Yadav S, Trivedi B, Qureshi SS. Desmoid fibromatosis of the oesophagus creating an oesophageal diverticulum in a 2-year-old girl. Afr J Paediatr Surg 2024;21:210-2.

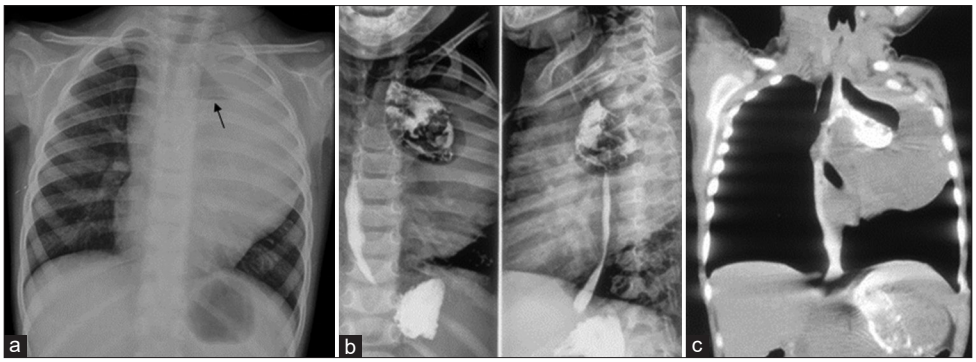


Figure 1: Radiological image demonstrating a homogeneous opacity occupying the left haemothorax with an outpouching (arrow) filled with air on chest X-ray (a), barium on barium swallow study (b) within the mass suggestive of an oesophageal diverticulum. Coronal (c) sections of computed tomography scan showed the anterior mediastinal mass around the oesophageal diverticulum

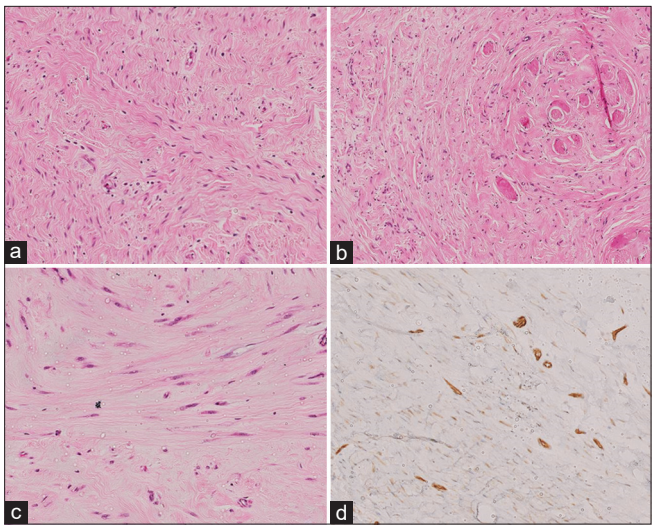


Figure 2: (a-c) Histology showed a sparsely cellular infiltrative spindle cell tumour with no specific growth pattern in a markedly sclerotic stroma. The tumour is low grade without any cellular atypia and infiltrates the skeletal muscle fibres of the oesophageal diverticulum (c). On immunohistochemistry, the spindle cells showed nuclear positivity for beta-catenin (d)

resection margins [Figure 2d]. At a follow-up of 40 months, the child has remained disease-free with satisfactory weight gain.

Discussion

The present case highlights the uncommon association of two different disease entities peculiarly in a 2-year-old child. Although DF may be observed in nearly every part of the body, it frequently involves the extremities, the trunk and the abdominal cavity; however, the oesophagus as the site of origin is exceptional.[2] Only an isolated case of fibromatosis of the oesophagus has been reported in the past.[4]

Similarly, EDs are rare and can occur at any oesophageal level including pharyngo-oesophageal, mid-oesophageal and epiphrenic.[5] Based on the aetiopathogenesis, the EDs are classified as pulsion diverticula resulting from an altered pressure gradient inside the oesophageal lumen or traction diverticula due to a chronic inflammatory process starting from the mediastinal lymph nodes which involve the oesophageal wall.[5] In the present case, the diverticulum was found at the level of the mid-oesophagus along with the DF shrouding it. The sheer size of the mass seems to be the likely reason for the traction that led to the formation of the diverticulum. Conventionally, mid-EDs tend to be small with a large neck, and they rarely become symptomatic or require medical attention; however, the synchronous presence of the DF produced the mass effect which led to vomiting, reduced nutritional intake and weight loss. Traction diverticula secondary to benign oesophageal pathologies such as giant oesophageal lipoma and leiomyoma have been reported previously.[6,7] Contrastingly, long-standing diverticula provide a risk of developing malignancy *de novo*.[7]

In the present case, although two diseases, DF and diverticula, were evident with the investigations, the oesophageal origin was confirmed only at the surgery. Since DF of the mediastinum could be non-visceral from the connective tissue, and due to their inherent property to infiltrate locally, a complete excision of the mediastinal disease may not be feasible in view of the presence of critical structures.[8] Hence, the surgical challenge was to deal with multiple pathologies and ensuring a safe and complete resection of both the entities. The oesophageal origin incidentally facilitated surgical resection with negative margins along with the diverticulectomy.

The treatment of DF witnessed a paradigm shift due to a better understanding of the disease biology.[9,10] Active surveillance and medical treatment frequently are the initial lines of treatment.[10] However, a lesion producing symptoms or resulting in complications requires individualisation of the treatment approach as was performed in the present case which led to complete relief of symptoms. DF has a high incidence of local recurrence which may be independent of wide negative margins.[2,3] Radiotherapy has been used for unresectable or recurrent disease, whereas adjuvant radiotherapy has been randomly used in the presence of positive surgical margins.[2,3] An array of systemic therapy has been utilised for DF including anti-oestrogenic drugs (tamoxifen and toremifene), non-steroidal anti-inflammatory drugs (meloxicam, indomethacin, sulindac and celecoxib), cytotoxic chemotherapy (methotrexate and

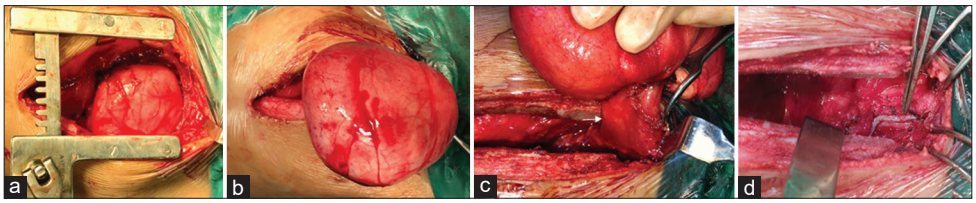


Figure 3: Serial intraoperative pictures showing the mass in the anterior mediastinum (a) not involving other mediastinal structures delivered through the sternotomy (b). The mass involved the distal portion of the oesophageal diverticulum (c) while the uninvolved proximal part (arrow) ensured the excision of the diverticulum and the desmoid fibromatosis (d)

vinblastine) and tyrosine kinase inhibitors (imatinib, sunitinib, pazopanib, sorafenib and sirolimus) for achieving disease stability.[2,3]

In conclusion, DF of the oesophagus and the contemporaneous traction ED are rare and may present a unique surgical challenge requiring simultaneous management of multiple pathologies. Although a favourable outcome was achieved with surgery for the ED and the DF, the inherently unpredictable nature of the latter obligates long-term follow-up. However, due to the unconventional oesophageal origin and the achievement of negative surgical margins due to the presence of diverticula, a favourable eventual outcome is anticipated in the present case.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the legal guardian has given his consent for the patient's images and other clinical information to be reported in the journal. The guardian understands that the patient's name and initials will not be published and due efforts will be made to conceal the patient's identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Fletcher CD, Bridge JA, Hogendoorn P, Mertens F. WHO Classification of Tumours of Soft Tissue and Bone (IARC WHO Classification of Tumours). 4th ed. Lyon, France: IARC; 2013.
2. Ganeshan D, Amini B, Nikolaidis P, Assing M, Vikram R. Current update on desmoid fibromatosis. J Comput Assist Tomogr 2019;43:29-38.
3. Garcia-Ortega DY, Martín-Tellez KS, Cuellar-Hubbe M, Martínez-Said H, Álvarez-Cano A, Brener-Chaoul M, *et al.* Desmoid-type fibromatosis. Cancers (Basel) 2020;12:1851.
4. Wolf Y, Katz S, Lax E, Okon E, Schiller M. Dysphagia in a child with aggressive fibromatosis of the esophagus. J Pediatr Surg 1989;24:1137-9.
5. Costantini M, Zaninotto G, Rizzetto C, Narne S, Ancona E. Oesophageal diverticula. Best Pract Res Clin Gastroenterol 2004;18:3-17.
6. Yekeler E, Yazicioglu A, Subasi M, Boztepe H. Giant esophageal lipoma as an uncommon cause of diverticula. Case Rep Gastroenterol 2016;10:446-51.
7. Herbella FA, Dubecz A, Patti MG. Esophageal diverticula and cancer. Dis Esophagus 2012;25:153-8.
8. Bhat V, Raju P, Rao S, Ramaiah S. Infantile fibromatosis: A rare cause of anterior mediastinal mass in a child. J Clin Imaging Sci 2015;5:34.
9. Kasper B, Raut CP, Gronchi A. Desmoid tumors: To treat or not to treat, that is the question. Cancer 2020;126:5213-21.
10. Timbergen MJ, Schut AW, Grünhagen DJ, Sleijfer S, Verhoef C. Active surveillance in desmoid-type fibromatosis: A systematic literature review. Eur J Cancer 2020;137:18-29.