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LUMPY BUMPY ESOPHAGUS: A RARE CAUSE OF PSEUDOACHALASIA

Pathology

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ABSTRACT

Benign esophageal tumors are slow growing but they rarely grow to the extent that may lead to pseudoachalasia like symptoms making complete surgical resection mandatory. We report a case of young girl presenting with dysphagia since 6 months and showing diffuse lesion on radiology involving esophagus and part of stomach.

KEYWORDS

Benign lesion, pseudoachalasia, Leiomyomatosis, Esophagus

INTRODUCTION

The most common benign esophageal tumor is leiomyoma which is usually single, solitary tumor[1]. Diffuse leiomyomatosis is a rare entity, which may involve the entire esophagus and proximal part of the stomach[2]. We report a case of a young girl presenting with progressive dysphagia and weight loss. Radiological investigations revealed a large diffuse mass involving distal esophagus and proximal stomach, which was diagnosed as diffuse esophageal leiomyomatosis.

CASE PRESENTATION

A 13-year-old girl came with complaints of progressive dysphagia, initially for solids, then liquids for 6 months. Patient also had failure to thrive, loss of weight and appetite. She had no history of vomiting, chest pain or cough.

Upper gastrointestinal scopy showed a 6 x 5 cm heterogeneous mass with well-defined borders. Overlying mucosa was normal without any vascularity near gastro-esophageal junction (GEJ). Computed tomography (CT) showed asymmetric circumferential mildly enhancing wall thickening of thoracic and abdominal esophagus for length of 16 cm and maximum wall thickness of 3.5 cm, extending from D3 to D12 vertebral level. There was significant luminal narrowing and upstream gross dilatation of upper thoracic and cervical esophagus along its entire length. There was no regional lymphadenopathy. (Figure 1).

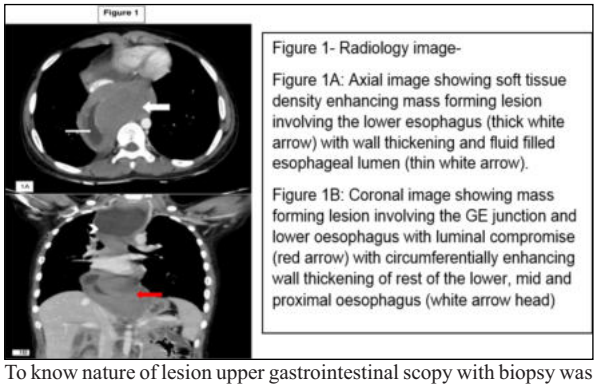

Figure 1- Radiology image-

Figure 1A: Axial image showing soft tissue density enhancing mass forming lesion involving the lower esophagus (thick white arrow) with wall thickening and fluid filled esophageal lumen (thin white arrow).

Figure 1B: Coronal image showing mass forming lesion involving the GE junction and lower oesophagus with luminal compromise (red arrow) with circumferentially enhancing wall thickening of rest of the lower, mid and proximal oesophagus (white arrow head)

To know nature of lesion upper gastrointestinal scopy with biopsy was done which showed features suggestive of leiomyoma. As patient had dysphagia leading to failure to thrive esophagectomy with partial gastrectomy was performed for symptomatic relief. We received segment of esophagus measuring 14.5 cm in length and part of the stomach measuring 2 cm. Esophageal adventitia, gastric serosa was normal with no evidence of perforation. On cut opening, multiple, diffuse, well circumscribed firm nodules with overlying normal mucosa were seen throughout the wall of distal esophagus and stomach. The largest nodule was measuring 3x2.5x1.5 cm and smallest measuring 0.5 cm in diameter. On cut surface whorling was seen. There was no evidence of necrosis or infiltrating borders.

Microscopic examination of multiple sections revealed normal esophageal and gastric mucosa with underlying tumour arising from the inner layer of muscularis propria arranged in interlacing fascicles, whorls and bundles. Tumour cells were spindle-shaped having elongated bland nuclei with blunt ends. There was no evidence of atypia, mitosis or necrosis. Differential diagnosis of GIST (Gastrointestinal Stromal Tumour) and leiomyoma were considered. Immunohistochemistry (IHC) panel was done for antibodies SMA, Desmin, CD117, CD34 and MIB1. Amongst these markers SMA and desmin were strong diffuse positive and CD117 and CD34 were negative. MIB1 labelling index (Ki 67 index) was <1%. (Figure 2).

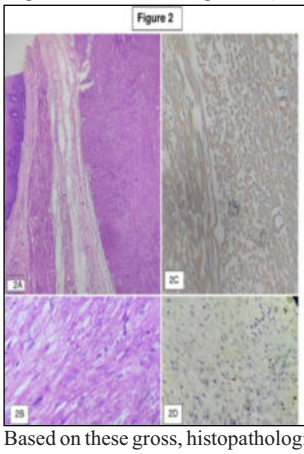

Figure 2- Histology and IHC

Figure 2A (10x H&E stain)- Normal esophageal mucosa, submucosa with tumour arising from muscularis propria,

Figure 2B (40x H&E stain) - Tumour cells are spindly with bland nuclei arranged in fascicles,

Figure 2C - IHC Smooth muscle actin - strong diffuse positive,

Figure 2D- IHC Ki-67 index - <1%.

Based on these gross, histopathological and IHC findings, diagnosis of diffuse esophageal leiomyomatosis was confirmed.

DISCUSSION

Benign esophageal tumors are rare and constitute less than 5% of overall esophageal tumor[1]. These tumors are usually asymptomatic but they may present as slowly progressing dysphagia. Based on their cell of origin mesenchymal benign esophageal tumours can be divided into GIST, Smooth muscle origin (leiomyoma, leiomyomatosis) or neural origin (schwannoma)[1].

Non-invasive radiological diagnostic modalities that can be used to know extent and character of lesions as initial steps are CT Scan, EUS, MRI, endoscopy, chest x-ray, barium swallow. Characteristic of benign lesions on radiology are that they do not erode or ulcerate overlying mucosa as seen in malignant lesions[3]. Our patient had diffuse involvement of mid, distal esophagus with gastroesophageal junction and proximal stomach with intact overlying mucosa.

Biopsy is the gold standard for diagnosis of these tumors. Biopsy was interpreted as leiomyoma in our patient. As patient was symptomatic surgical management was mandatory. Disease was diffusely involving most of part of esophagus and proximal stomach, esophagectomy with partial gastrectomy was performed instead of enucleation which is usually done in cases of leiomyoma[4].

Diffuse esophageal leiomyomatosis (DEL) is a rare smooth muscle benign hamartomatous lesion[3]. A review of the literature was performed by Federici et al.[5] in 1997 and included 25 cases of patients under the age of 14, and again by Ziogas et al[6]. in 2018 reported 35 cases under age of 18. DEL are common in children and young adults and are 1.6 times more common in women with usual age of presentation being 10 to 14 years. It is commonly associated with Alport syndrome[7]. In our patient there were no symptoms of hearing loss or haematuria which ruled out Alport syndrome.

Partial or complete esophagectomy with partial gastrectomy depending on involved segment and symptoms is the treatment of choice in patients of DEL. Complete surgical resection is usually curative and show good prognosis[8]. Our patient had good post-operative symptomatic relief and currently on tube feeds. These lesions rarely undergo sarcomatous change and their relapse is not described in children after complete resection[5].

CONCLUSION

Diffuse Esophageal Leiomyomatosis is a rare condition. Histopathology including immunohistochemistry is the gold standard for diagnosis of Diffuse Esophageal Leiomyomatosis. Surgical resection is the treatment of choice in patients with severe and progressive symptomatic presentation.

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REFERENCES

1. Ha C, Regan J, Cetindag IB, Ali A et al. Benign esophageal tumors. Surg Clin North Am. 2015; 95:491-514.
2. Ray S, Saluja SS, Gupta R et al. Esophageal leiomyomatosis -- an unusual cause of pseudoachalasia. Can J Gastroenterol. 2008; 22:187-189.
3. Lekawale H, Khandeparkar SS, Deshmukh S et al. A rare case of giant diffuse esophageal leiomyomatosis. Med J DY Patil Univ 2015; 8:384-6
4. Cheng YL, Hsu JY, Hsu HH et al. Diffuse leiomyomatosis of the esophagus. Dig Surg. 2000; 17:528-31
5. Federici S, Ceccarelli PL, Bernardi F et al. Esophageal leiomyomatosis in children: report of a case and review of the literature. Eur J Pediatr Surg.1998; 8:358-63.
6. Ziogas IA, Mylonas KS, Tsoulfas G et al. Diffuse Esophageal Leiomyomatosis in Pediatric Patients: A Systematic Review and Quality of Evidence Assessment. Eur J Pediatr Surg. 2019; 29:487-494.
7. Crouch G, Devitt PG, Thompson SK. Diffuse esophageal leiomyomatosis. Dis Esophagus. 2011; 24:145-6
8. Rapp JB, Ciullo S, Mallon MG. Diffuse esophageal leiomyomatosis: A case report with surgical correlation. Clin Imaging. 2019; 58:161-165.