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KEY WORDS:
Pseudoachalasia, Aganglionosis, Leiomyomatosis, Endoscopic ultrasonography, Gastric pull through surgery.

DIFFUSE LEIOMYOMATOSIS OF THE ESOPHAGUS – A RARE CAUSE OF PEDIATRIC PSEUDOACHALASIA.

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INTRODUCTION:

Achalasia is an esophageal motility disorder with progressive dysphagia for liquids and solids. It occurs due to absence or degeneration of myenteric plexus of nerves in the distal esophagus. The term pseudoachalasia refers to achalasia like symptoms and imaging manifestations caused by diseases other than aganglionosis [1]. It can be due to stricture, extrinsic compression, malignant neoplasm or proliferation of smooth muscle cells as in leiomyomatosis. Diffuse leiomyomatosis of the esophagus is a very rare cause of pseudoachalasia and requires total esophagectomy with gastric pull through surgery. We present a rare case of a seven year old boy with diffuse leiomyomatosis of esophagus presenting with pseudoachalasia, aspiration pneumonitis and bronchiectasis secondary to recurrent aspirations.

Case Report:

A seven year old boy presented to the pediatric OPD with cough and fever. Detailed history reveled that he had repeated attacks of respiratory infections since 4 years and had developed difficulty in swallowing from past 3 years. His general physical examination was normal except for mild fever (99 ^{0}C). His laboratory investigations were normal except for mild leucocytosis. Ultrasonography of the abdomen was normal.

Chest x ray of the patient showed large tubular right paracardiac opacity (Fig.1). Nasogastric tube was visualized on the right side of the thorax. CT scan of the thorax was performed to evaluate the opacity. It showed grossly dilated esophagus with transverse diameter of 35mm. The distal esophagus showed tapering & smooth wall thickening with wall thickness of 30mm (Fig. 2A). The wall thickening extended to the gastric fundus & cardia distally and mid esophagus proximally (Fig. 2B). The stomach was seen below the diaphragm and there was no hiatus hernia (Fig. 2C). The lower lobe of both the lungs showed multifocal patchy consolidations and bronchiectasis (Fig.3). Barium swallow with NG tube in situ demonstrated features of achalasia cardia (Fig.4).

Based on the imaging findings, a diagnosis of pseudoachalaia was made and endoscopy was suggested. Endoscopy also revealed features suggestive of achalasia with intact mucosa. The patient was posted for surgical myotomy. As there was wall thickening in the esophagus, he underwent surgical biopsy. It showed diffusely proliferated smooth muscle cells of the esophagus in multiple pockets of whorled pattern. The mucosa of the esophagus was intact. The proliferated cells appeared benign without mitotic activity. Final diagnosis of diffuse leiomyomatosis of the esophagus with pseudoachalasia was made. Because of its rarity, this case is presented here.

DISCUSSION:

Achalasia cardia or cardiospasm is a misnomer. It is caused by the absence of myenteric plexus in the distal esophagus leading to increased tone of GE junction and resultant esophageal dilatation. It initially starts in young adolescence with dysphagia for only liquids and progresses to severe dysphagia for both liquids & solids by middle age [2].

The term pseudoachalasia refers to achalasia like imaging appearance and clinical features secondary to some other cause. It can be due to systemic diseases like amyloidosis, sarcoidosis, scleroderma & sjogren's syndrome, extrinsic compression of the esophagus caused by pseudocyst of the pancreas or aortic aneurysm, post surgical complication of bariatric surgery or banding of varices, secondary to benign tumors of the esophagus like neurofibromatosis, esophageal leiomyoma or diffuse hypertrophy of muscle layer of the esophagus [2]. The commonest malignant cause of pseuoachalasia is carcinoma of distal esophagus. Other malignant causes include carcinoma of lung, pancreas, hepatobiliary malignancies, mesothelioma and lymphoma [3,4].

Mucosal integrity is the key differentiating feature between true achalasia and pseudoachalasia due to carcinoma. However, long standing achalasia is a risk factor for carcinoma of esophagus. The mechanism of dysphagia in pseudoachalasia of carcinoma is encasement of the lower esophageal sphincter by tumor / infiltration of the myenteric plexus / para neoplastic neuropathy [4].

Diffuse leiomyomatosis of the esophagus is a rare hamartomatous disorder described for the first time by Hall [5]. It is characterized by diffuse unencapsulated proliferation of the smooth muscle layer of the esophagus with intact mucosa. In contrast, leiomyoma is a focal encapsulated proliferation of smooth muscle cells. Esophageal leiomyomatosis is commonly associated with Alport syndrome [6]. It may also be seen as a part of diffuse visceral leiomyomatosis of multiple organs of the body [6]. In our case, no such association was found.

Leiomyomatosis can occur at any age but most commonly seen in children and young adults. Usual age of presentation is about 10-14 years. Federici et al have reported a case in a 6 month old baby [7]. It is slightly more common in females (F:M = 1.6:1). Our patient is a boy of 7 years.

The incidence of childhood achalasia is 0.1 per 1 lakh children [8]. Diffuse leiomyomatosis is a very rare cause of childhood achalasia. It commonly presents with clinical features of vomiting, dysphagia, regurgitation, and weight loss [8]. Atypical presentations include recurrent pneumonia, nocturnal cough, hoarseness of voice, aspiration and feeding difficulties [9]. Our patient presented with recurrent chest

infections and fever due to regurgitation and aspiration pneumonitis.

In a comparative study of achalasia & pseudoachalasia due to neoplasm, Tracey et al have observed that duration of symptoms was shorter & weight loss for the duration was longer in patients with malignancy than idiopathic achalasia [10]. They concluded that negative findings on imaging and endoscopy should not give false re assurance. Rather, repeated biopsies/surgical exploration should be performed to ascertain the cause of achalasia. Similarly, in our case also the findings were confirmed on surgical biopsy and the diagnosis was established.

As the disease entity is rare, diagnosis of diffuse leiomyomatosis is difficult. Chest x ray may show mediastinal widening/para cardiac opacity. Barium swallow mimics achalasia and can not differentiate between true and pseudoachalasia. CT and MRI show thickening of the muscle layer of the esophagus. Extension of the thickening to fundus and cardia of the stomach is a characteristic feature. Such thickening is not seen in true achalasia [11]. In our case also, extensive thickening of the esophageal muscle layer was seen with extension into gastric cardia and fundus.

Endoscopic ultrasonography (EUS) is the best diagnostic study to confirm leiomyomatosis and useful to avoid potentially harmful surgery of myotomy [12,13]. In our case, however, endoscopic ultrasonography was not performed. Average wall thickness in leiomyomatosis can be upto 4cm. The involvement is common in the middle and distal esophagus [13]. In our case, the wall thickness was 2.5cm with extension upto mid esophagus proximally and gastric cardia & fundus distally.

Total esophagectomy is preformed to prevent recurrences as small pockets of leiomyomas may be left over if subtotal esophagectomy is performed. The food way is reconstructed by either gastric or colonic transposition. Gastric pull through is preferred as the long term outcome is good [14].

CONCLUSION:

Diffuse leiomyomatosis of the esophagus is a rare cause of pseudoachalasia. Thickening of the muscular layer of the distal esophagus and extension of the thickening into the gastric fundus and cardia are the key findings in the diagnosis. All the cases should be confirmed on biopsy. Total esophagectomy & gastric pull through surgery is the definitive treatment.

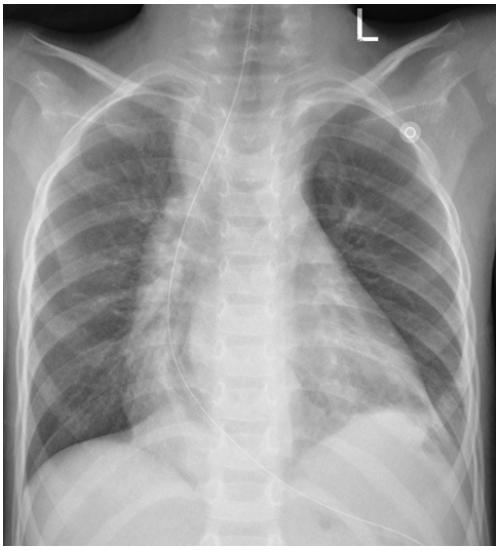

Fig.1: Chest Radiograph Showing Right Para Cardiac Opacity With NG Tube In The Right Side Of The Thorax.

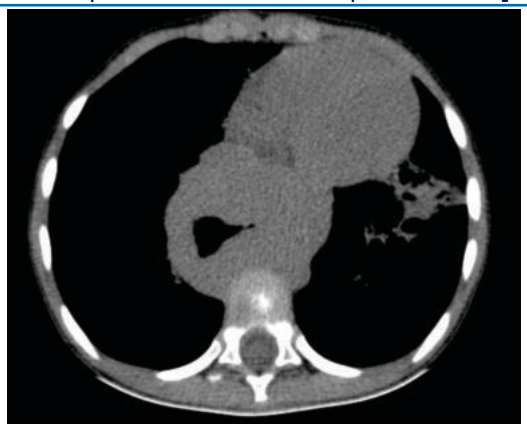

Fig. 2A: Axial CT Section In Mediastinal Window, Showing Circumferential Wall Thickening Of The Esophagus.

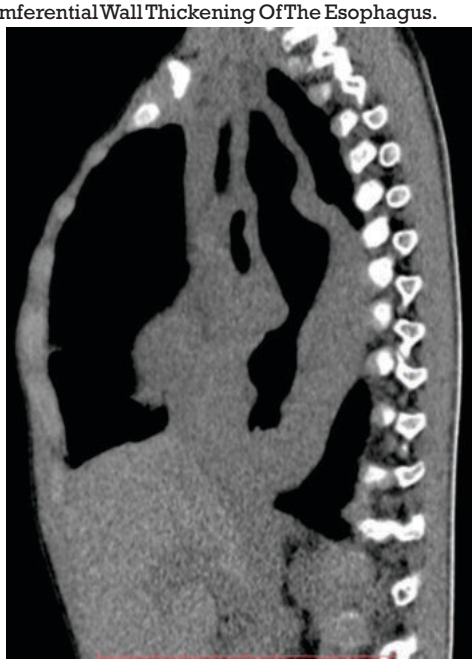

Fig. 2B: Sagittal CT Section In Mediastinal Window Showing The Extension Of Wall Thickening Till The Mid Esophagus.

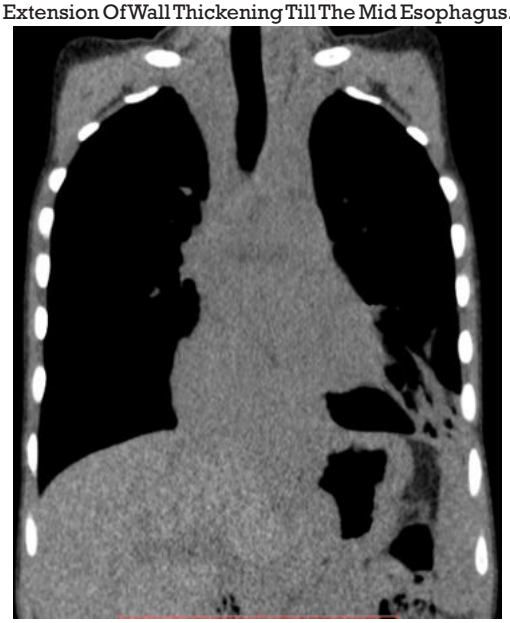

Fig. 2C: Coronal CT Section In Mediastinal Window Showing The Wall Thickening Of Gastric Fundus And Presence Of The Fundus Below The Diaphragm.

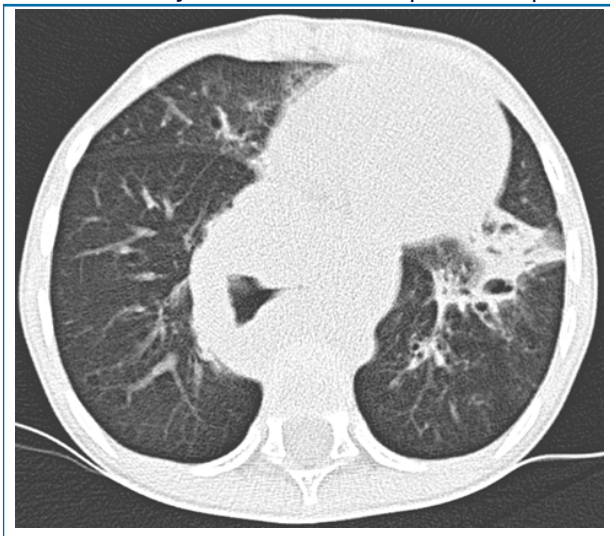

Fig.3: Lung Window Image Showing Bilateral Lower Lobe Consolidations And Bronchiectasis.

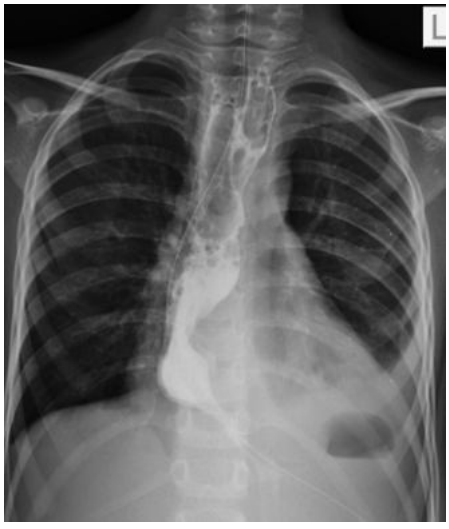

Fig.4: Barium Swallow Image Showing Dilated Esophagus With Smooth Tapering Of The Distal End.

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