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Esophageal Leiomyomatosis-A Rare but Debilitating Clinical Entity!

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Introduction

Esophageal leiomyomatosis is defined as diffuse hypertrophy of esophageal wall muscles except muscularis mucosae. It is mostly seen in children and young adults.[1] Moreover , it has been reported in old age as well. [2] They come from smooth muscles principally and are a diffuse variant of leiomyomas of esophagus which constitute 0.4 % of neoplasms of this organ.[3] They are slow growing tumors and are generally asymptomatic. Size greater than 5cm have been correlated with symptomatology in these cases.[4] Familial variety has been seen in association with Alport syndrome.[5] The common imaging findings include smooth tapering of the distal esophagus on barium swallow and thickening of the distal esophageal wall on computed tomography scan.[6]

Case Report

A 21year old male, smoker, presented to us with complaints of slowly progressive difficulty in swallowing solids and then liquids over 3-4 years. His dysphagia was not intermittent, with food sticking initially in lower chest and afterwards in upper chest. He had associated productive cough, regurgitation of food but not through nose or at night, and dyspepsia for which he used to take proton pump inhibitors. He had history of repeated chest infections from first decade of life and more so over the last 4 years before presenting to us. He did not have any positive history of GI malignancy in the family, infectious disease, lympho or myeloproliferative disorder, respiratory tract malignancy or radiation exposure. He did not report drooling of saliva or coughing and choking during food consumption, hematemesis, skin lesion or intake of antibiotics, anti-inflammatories, or bisphosphonates. His labs were unremarkable and no evidence of hematuria, proteinuria was present. He underwent equivocal upper GI endoscopy. Repeat endoscopy and EUS with FNB revealed esophageal leiomyomatosis. HRCT chest revealed large well defined soft tissue density mass approximately 10x7.5x7.2cm at distal end of esophagus extending into stomach cardia. CT also revealed marked circumferential thickening of middle esophagus with retrograde dilatation. Anteriorly, the narrowed middle esophagus was abutting the trachea and carina and displacing them anteriorly. Aorta was encased by lesion more than 90^0 but less than 180^0.

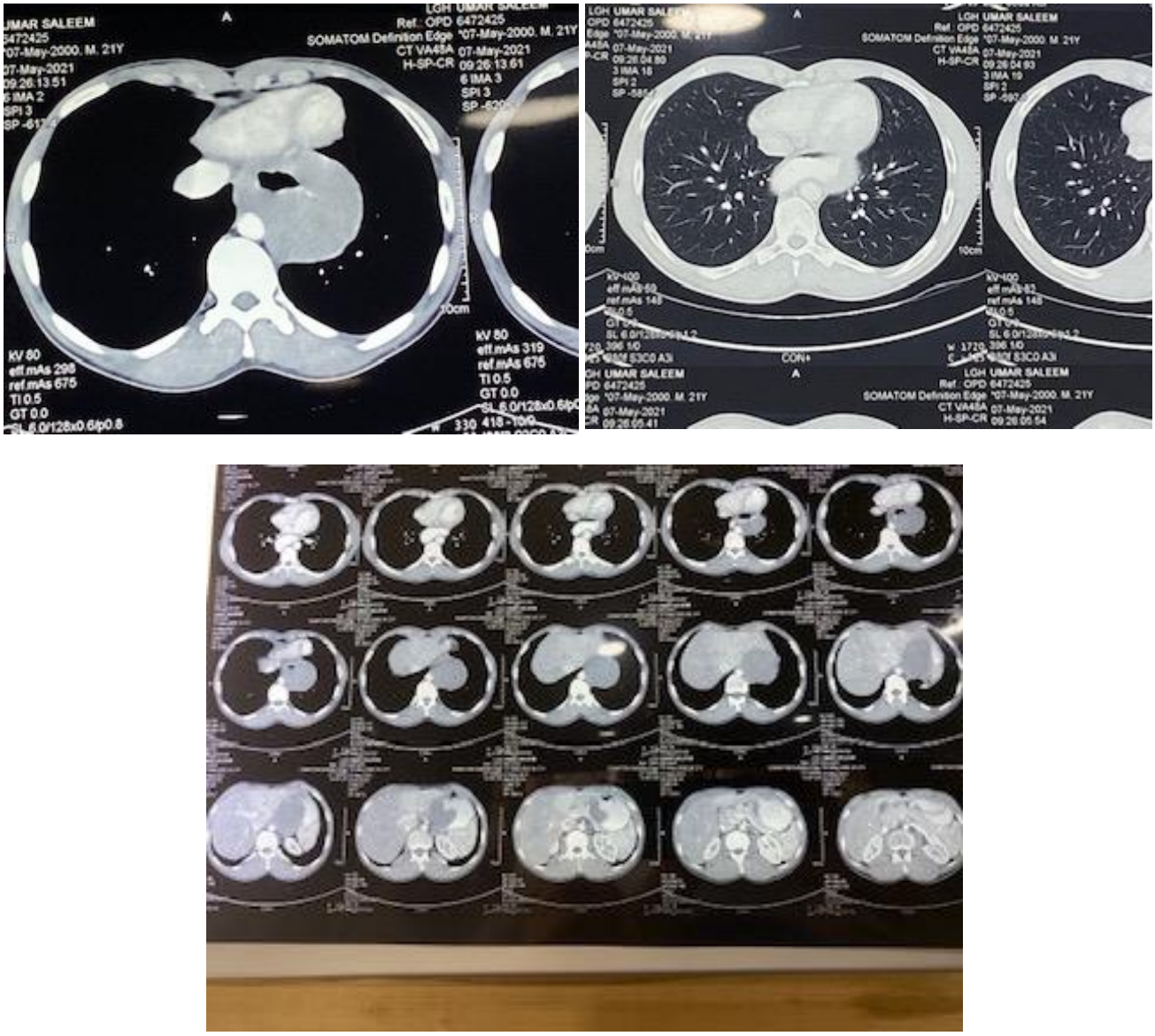


Fig 1: CT Chest showing circumferential luminal thickening with encasement of aorta and involvement of cardia of stomach.

After anesthesia fitness protocols, Transhiatal esophagectomy with stomach pull up and esophagogastric anastomosis in the neck was performed. Histopathology report revealed esophageal leiomyomatosis without cellular pleomorphism, necrosis, atypia, and benign squamous mucosa without ulceration. Immunoprofiling turned out to be positive for desmin and h-Caldemon. Proximal and distal resection margins were unremarkable. Post-operative course of the patient remained smooth, and he was discharged on 12th post-operative day for follow up with us.

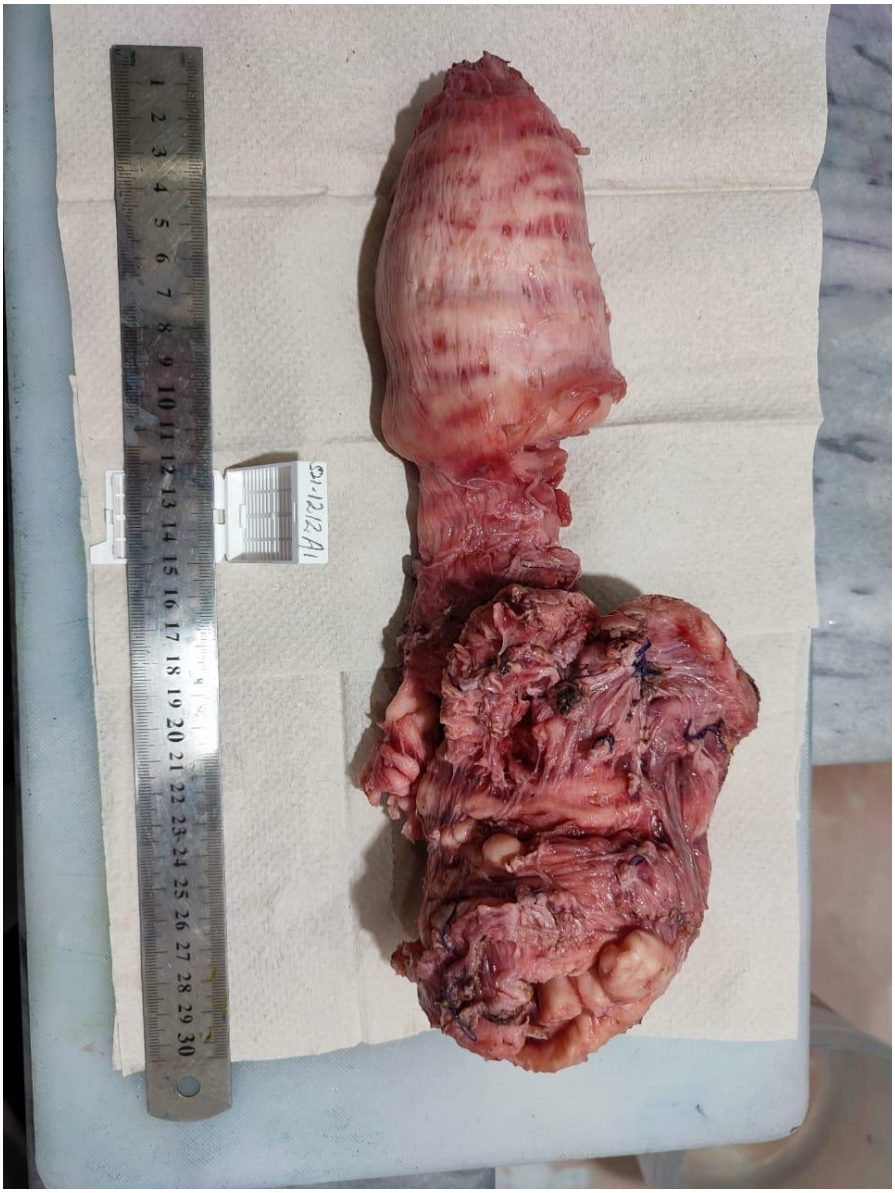
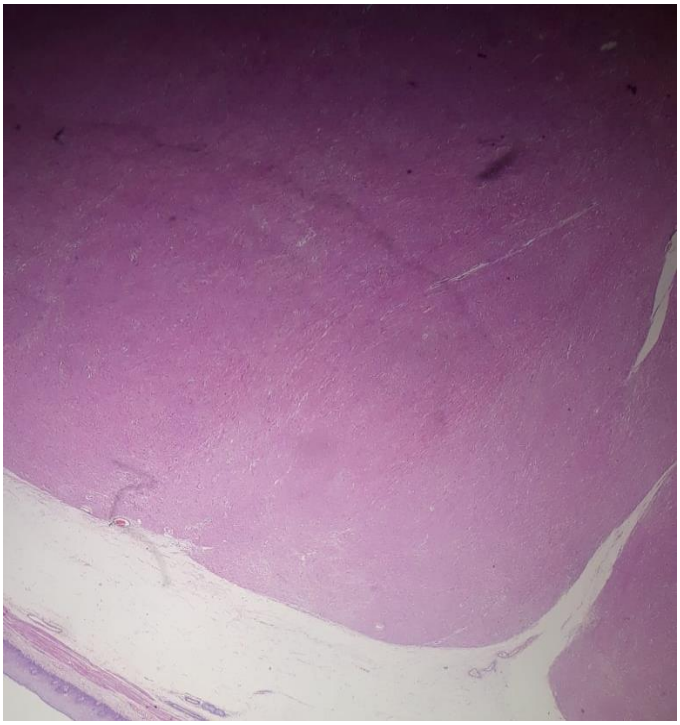
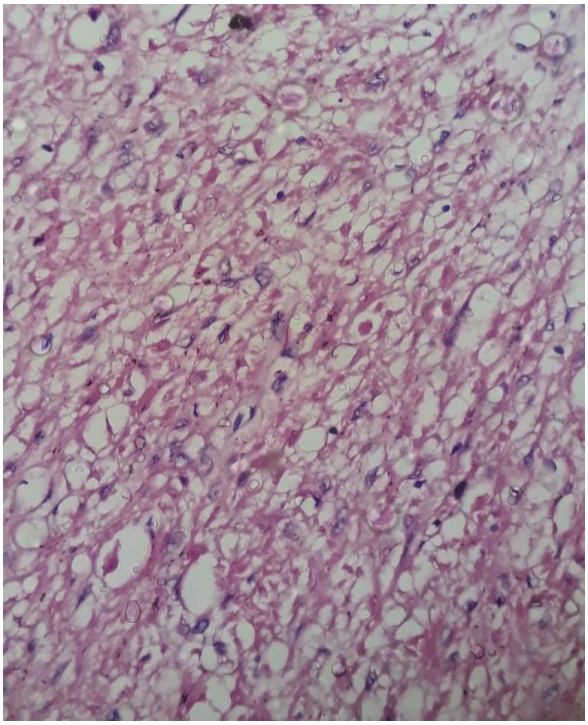


Fig: Histopathology view of Transhiatal esophagectomy specimen.





Discussion

Esophageal leiomyomatosis has been described as a hamartomatous entity with sporadic and hereditary profiles. It can be associated with gastrointestinal leiomyomata, tracheobronchial and genital lesions. Slowly progressive and long-standing dysphagia are classic features. Achalasia is its most common mimicker.The average time from symptom onset to diagnosis may be 3 years. **[**7**]** It commonly involves middle and distal third of esophagus but can reach up to cardia in 80% of cases. In 35% of cases, it can involve whole of esophagus. Increased length of stricture on barium swallow differentiates it from achalasia as does the eccentric bulge indicating submucosal lesion. CT scan can depict extension of soft tissue lesion into the cardia of the stomach, not typically seen with achalasia. **[**8**]** Once diagnosis is established, family can be screened for possibility of Alport syndrome, esophageal vulvar syndrome.**[**9**]** The treatment of choice is esophagectomy with transposition of stomach to restore the GI continuity.**[**10**]** Partial gastrectomy and colonic interposition may be required in some cases. **[**11**]**

Conclusion

Esophageal leiomyomatosis can cause significant debilitation to the patient and warrants clinical suspicion incorporated in the diagnostic plan and treatment to avoid morbidity and mortality.

Reference

1. Bloch P, Quijada J. Léiomyomatose diffuse de l'oesophage. Analyse d'un cas et revue de la littérature **[**Diffuse leiomyomatosis of the esophagus. Analysis of a case and review of the literature**]**. Gastroenterol Clin Biol. 1992;16(11):890-3. French. PMID: 1483560.

2. Crouch G, Devitt PG, Thompson SK. Diffuse esophageal leiomyomatosis. Dis Esophagus. 2011 Apr;24(3):145-6. doi: 10.1111/j.1442-2050.2010.01114.x. Epub 2010 Oct 11. PMID: 21489040.

3. Reinbold WD, Oglodek C, Hillemanns A, Grönniger J. Leiomyomatose des Osophagus **[**Leiomyomatosis of the esophagus**]**. Radiologe. 1994 Sep;34(9):534-6. German. PMID: 7800802.

4. Misra M, Maziak DE, Shamji FM, Michaud C, Perkins DG, Matzinger F. Esophageal leiomyomatosis. Med Sci Monit. 2003 Nov;9(11):CS98-101. PMID: 14586279.

5. Delteil C, Macagno N, Daniel L, D'Journo XB, Guisiano S, Garcia S, Secq V. Léiomyomatose œsophagienne diffuse et syndrome d'Alport : rapport d'un cas **[**Diffuse esophageal leiomyomatosis and Alport's syndrome: A case report and review of the literature**]**. Ann Pathol. 2019 Feb;39(1):36-39. French. doi: 10.1016/j.annpat.2018.08.007. Epub 2018 Oct 12. PMID: 30322718.

6. Levine MS, Buck JL, Pantongrag-Brown L, Buetow PC, Lowry MA, Sobin LH. Esophageal leiomyomatosis. Radiology. 1996 May;199(2):533-6. doi: 10.1148/radiology.199.2.8668807. PMID: 8668807.

7. Gupta V, Lal A, Sinha SK, Nada R, Gupta NM. Leiomyomatosis of the esophagus: experience over a decade. J Gastrointest Surg. 2009 Feb;13(2):206-11. doi: 10.1007/s11605-008-0701-x. Epub 2008 Sep 26. PMID: 18818979.

8. Ray S, Saluja SS, Gupta R, Chattopadhyay TK. Esophageal leiomyomatosis -- an unusual cause of pseudoachalasia. Can J Gastroenterol. 2008 Feb;22(2):187-9. doi: 10.1155/2008/603105. PMID: 18299739; PMCID: PMC2659141.

9. Rapp JB, Ciullo S, Mallon MG. Diffuse esophageal leiomyomatosis: A case report with surgical correlation. Clin Imaging. 2019 Nov-Dec;58:161-165. doi: 10.1016/j.clinimag.2019.07.008. Epub 2019 Jul 23. PMID: 31377438.

10. Ziogas IA, Mylonas KS, Tsoulfas G, Spartalis E, Zavras N, Nikiteas N, Schizas D. Diffuse Esophageal Leiomyomatosis in Pediatric Patients: A Systematic Review and Quality of Evidence Assessment. Eur J Pediatr Surg. 2019 Dec;29(6):487-494. doi: 10.1055/s-0038-1676507. Epub 2018 Dec 21. PMID: 30577043.

11. Thomas LA, Balaratnam N, Richards DG, Duane PD. Diffuse esophageal leiomyomatosis: another cause of pseudoachalasia. Dis Esophagus. 2000;13(2):165-8. doi: 10.1046/j.1442-2050.2000.00106.x. PMID: 14601910.