GASTROESOPHAGEAL LEIOMYOMATOSIS IN A YOUNG WOMAN WITH VULVAR LEIOMYOMATOSIS AND GALLSTONE DISEASE: REPORT OF A RARE CASE

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ABSTRACT

Gastroesophageal-vulvar leiomyomatosis is a very rare condition; it is characterized by diffuse, ill-defined proliferation of smooth muscle in the esophagus and vulva. We present an interesting case of esophageal leiomyomatosis in a woman with a history of vulvar leiomyomatosis and a gall bladder full of stones. She was 18-years old and had a 4 year history of vulvar mass and clitoromegaly, with subclinical dysphagia. This paper represents the first reported simultaneous occurrence of these three pathologic entities in the English literature.

Esophago-vulvar leiomyomatosis should be considered in a young patient with vulvar mass and long-standing dysphagia in whom a smooth, tapered esophageal narrowing on barium study and circumferential esophageal wall thickening on CT scan are seen. An esophagectomy combined with a reconstruction procedure is indicated.

INTRODUCTION

Leiomyomatosis is a rare condition of unknown etiology; there is diffuse thickening of the esophageal musculature. Few cases of leiomyomatosis have been described so far in the literature. The largest series, from Mexico, only described six cases, four of which belonged to one family, indicating that heredity may have a role in pathogenesis. The patients are often children and young adults, and females may be affected more often than males. It may be isolated or found in association with other intrathoracic and genital localizations or Alport syndrome (nephropathy, sensorineural deafness, and ocular lesions). Genital localizations, exclusively in women (clitoral hypertrophy, vulvar leiomyomatosis), in association with esophageal leiomyomatosis comprise the esophago-vulvar syndrome. In this article, we present an interesting case of esophago-vulvar leiomyomatosis in a woman with gallstone disease. This paper represents the first reported simultaneous occurrence of these three pathologic entities in the English literature.

CASE REPORT

An 18 year old woman presented with clitoromegaly and retrovulvar mass. She hadn’t told her problem to anybody until she got married and her husband referred her to a physician.

On physical examination, the lungs were clear, there was no palpable abdominal mass but abnormal genitalia because of clitoromegaly and a retrovulvar mass was detected (Fig. 1). Examination of hymen and vagina was normal. Chest radiographs (Fig. 2) demonstrated a large middle mediastinal mass in the region of the esophagus. Further questioning revealed mild dysphagia, which limited her to small bites of food. UGI Endoscopy didn’t show intraluminal invasion and a minimal degree of mucosal inflammation was confirmed by biopsy. The patient had no signs of Alport’s syndrome such as renal disease, sensorineural hearing loss, and ocular impairment. Family history for similar abnormalities was unremarkable.

Barium esophagogram (Fig. 3) showed a consider-
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Fig. 1. The patient with esophagovulvar leiomyomatosis.

Fig. 2. CXR (PA) demonstrates a large middle mediastinal mass in the region of the esophagus.

Fig. 3. Barium esophagogram demonstrates considerable variability in the caliber of the esophagus with regions that were quite distended. No passage disturbance was present.

Fig. 4. Contrast-enhanced computed tomography of the chest reveals a long segment of circumferential soft tissue mass encircling the esophagus.

able variability in the caliber of the esophagus with regions that were quite distended. The maximum diameter in some areas extended up to 15 centimeters and was similar to a stomach! The mass extended to the gastroesophageal junction but did not seem to involve the fundus per se. Sonography and CT scan revealed the soft tissue bicomponent masses intimately associated with the esophagus though the lumen of the esophagus was quite smooth (Fig. 4). No adenopathy, either in the gastrohepatic region or in the mediastinum was defined on
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Fig. 5. Gross specimen of the esophagus (A-P view) reveals a circumferential multilobulated solid mass encircling the esophagus which has an intact mucosa.

Fig. 6. Cut surface of the esophageal specimen shows bulk of the tumor at the lower end and abnormal muscular hypertrophy throughout the whole esophagus.

The gross specimen of the esophagus. The complex bicomponent mass was within the esophageal wall. It extended several centimeters proximal to the azygos vein to the hiatus. Intraoperatively, it was decided that esophageal preservation was not possible. After resection of the thoracic esophagus (Figs. 5, 6) the patient was turned supine and a laparotomy and left neck incision were performed. The stomach was mobilized and transposed through the posterior mediastinum to the neck and a cervical esophagogastronomy was performed. The genital mass was excised and the remaining skin was reapproximated. The gallbladder containing more than 30 stones also was removed (Fig. 7).

Postoperatively the patient needed further ventilation support for 2-3 days but made a smooth postoperative recovery.

The gross specimen of the vulvar lesion was a well-demarcated tumor measured 11 x 5 x 4 cm with a bulging gray cut surface.

Histologic examination showed poorly defined interlacing bundles of smooth muscle cells with blunt ended, cigar shaped nuclei and eosinophilic cytoplasm. There was no necrosis (Fig. 8). Cytoplasmic immunoreactivity for desmin, actin, and vimentin was present by...
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Fig. 8. Histopathologic examination of mass (H & E; original magnification x 100), show diffuse proliferation of smooth muscle cells as interlacing bundles with blunt ended, cigar shaped nuclei and eosinophilic cytoplasm.

Abdominal exploration was negative for other pathologies but the gall bladder was full of stones, and a cholecystectomy was performed.

DISCUSSION

Leiomyomas are benign tumors of smooth muscle origin and represent only 0.4% of all esophageal tumors.\textsuperscript{4} The lesions are mostly solitary, and multiple lesions are rare.\textsuperscript{4} Only a few cases of diffuse esophageal leiomyomatosis have been reported. On the other hand, smooth muscle neoplasms of the vulva are also exceedingly rare, a comprehensive study included only 32.\textsuperscript{18} Genital localizations, exclusively in women (clitoral hypertrophy, vulvar leiomyomatosis), in association with esophageal leiomyomatosis comprise the esophago-vulvar syndrome. Only a few cases of this rare syndrome have been described so far in the literature.\textsuperscript{5,7,19,20,21,23}

Leiomyomatosis represents a diffuse proliferation of smooth muscle that forms ill-defined nodules. Some authors have questioned whether they represent true neo-

Fig. 9. (a, b, c). Immunohistochemical examination of vulvar mass biopsy with desmin-vimentin-actin markers show positive tumoral cell immunoreactivity (Immunostaining desmin-vimentin-actin x 250).
The clinical history of slowly insidious progressive dysphagia in a young patient and radiologic findings of smooth mucosa lining seen on the esophagogram are helpful in differentiating this benign entity from a malignant tumor.\textsuperscript{4,13} We didn't find any abnormality in hematologic and biochemical profiles related to the syndrome.

Upon reviewing the literature, esophageal-vulvar leiomyomatosis with cholelithiasis has not been reported yet and therefore this case may be the first case report. In conclusion, recognition of esophageal-vulvar leiomyomatosis is important because of its benign prognosis. Furthermore, awareness of this uncommon clinical condition may facilitate the proper diagnosis and treatment. Our case is typical in that the patient was young and presented with a 4 years history of vulvar mass and clitoromegaly, with subclinical dysphagia. We emphasize that leiomyoma of the vulva should prompt X-ray studies of the esophagus, and leiomyoma of the esophagus should prompt a search for vulvar leiomyoma.

REFERENCES

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