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Familial massive leiomyoma with esophageal leiomyomatosis: an unusual presentation in a father and his 2 daughters

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Abstract Esophageal leiomyomatosis and leiomyoma are benign neoplastic lesions composed of proliferating smooth muscle cells. Although rare, these 2 conditions may occur simultaneously in an individual patient. Symptomatic patients often require surgical management. We describe the first reported cases of family members presenting with esophageal leiomyomatosis and concomitant massive esophageal leiomyoma.

Esophageal leiomyomatosis is a rare, benign, neoplastic condition characterized by thickening of the esophageal wall as a result of aberrant smooth muscle proliferation. This condition predominantly affects children and young adults but can present at any age. Esophageal leiomyomatosis may occur in isolation or as a familial disorder and has been associated with other conditions such as Alport syndrome and visceral leiomyomatosis [1-4]. We present 3 cases of esophageal leiomyomatosis with concurrent esophageal leiomyoma occurring in a single family. Affected members included the father, a 15-year-old daughter, and her 12-year-old sister.

1. Case report

A 15-year-old girl presented to a migrant farm workers' clinic for a public school admission medical evaluation. Because of a positive screening purified protein derivative of tuberculin, a chest radiograph was obtained (Fig. 1). The chest x-ray revealed a large mass in the right chest. Subsequent computed tomography (CT) scan demonstrated a 15-cm distal esophageal mass consistent with a large leiomyoma (Fig. 2). On further questioning, it became apparent that she had progressive dysphagia over several years. Unfortunately, while completing her medical evaluation and plans for resection, her father, a 36-year-old immigrant from Mexico, collapsed and died. A postmortem examination revealed multiple and massive esophagogastric leiomyomata, one of which was necrotic and had recently bled. His cause of death was massive exsanguination because of bleeding into the esophagus and upper gastrointestinal track.

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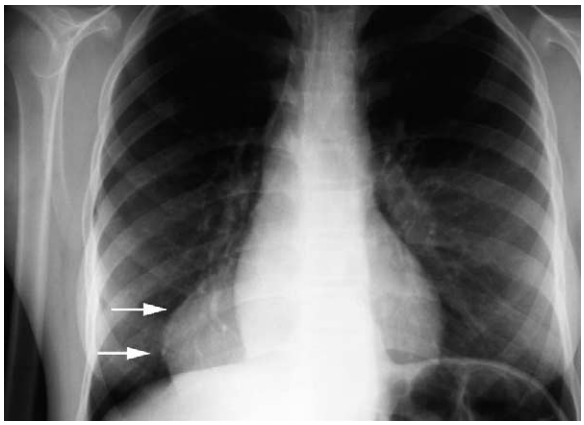

Fig. 1 Chest radiograph of the 15-year-old girl demonstrating large mass located to the right of the cardiac border (arrows).

The 15-year-old girl underwent a total esophagectomy via a right thoracotomy, upper midline laparotomy, and left neck incision. At thoracotomy, a large esophageal tumor extending down into the costophrenic sulcus was identified and resected (Fig. 3). Gastrointestinal continuity was reestablished with creation of a gastric tube and cervical esophagogastric anastomosis carried out through a left cervical incision. She required a repeat laparotomy in the perioperative period for herniation of colon through the hiatus. The patient is now tolerating a regular diet and doing well 1 year after resection.

The resected esophagus was 25 cm in length and appeared as a thickened tube averaging 3.7 cm in diameter. Enveloping the distal 14 cm of the specimen was a 14 × 8 cm circumferential tumor. Inspection of the cut surface of the esophageal wall revealed the muscular layer diffusely thickened to 0.8 cm (Fig. 4). The mucosa was intact and unremarkable. Microscopic examination showed marked hypertrophy of both layers of the muscularis propria and, to a lesser extent, the muscularis mucosa. This

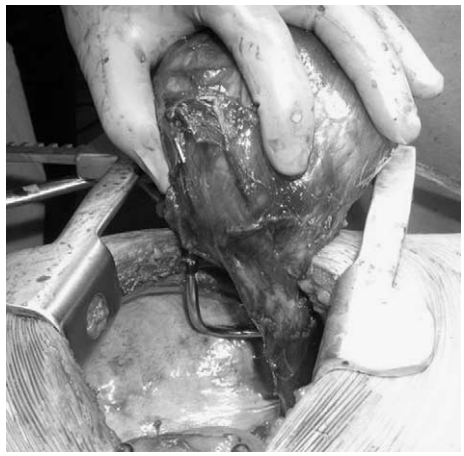

Fig. 3 Operative photograph showing massive distal leiomyoma resected from the 15-year-old girl.

diffuse lesion was composed of hypertrophic smooth muscle cells and displayed patchy infiltration by eosinophils as well as focal areas of calcification and fibrosis. Immunohistochemical analysis revealed positive staining for desmin, muscle-specific actin, and smooth muscle actin, and negative staining for S-100. The distal tumor displayed similar histopathology and was determined to be an esophageal leiomyoma.

The 12-year old sister also had a positive purified protein derivative of tuberculin and had a chest radiograph performed which revealed a mediastinal mass protruding into the right chest (Fig. 5). CT and magnetic resonance imaging (MRI) confirmed a large distal esophageal soft tissue mass consistent with a leiomyoma (Fig. 6). Operative findings and management were similar to her sister. Histopathologic findings were also similar to those of her sister, with diffuse lobulated thickening of the esophageal wall because of hypertrophy of smooth muscle cells. Features of the distal tumor were consistent with an

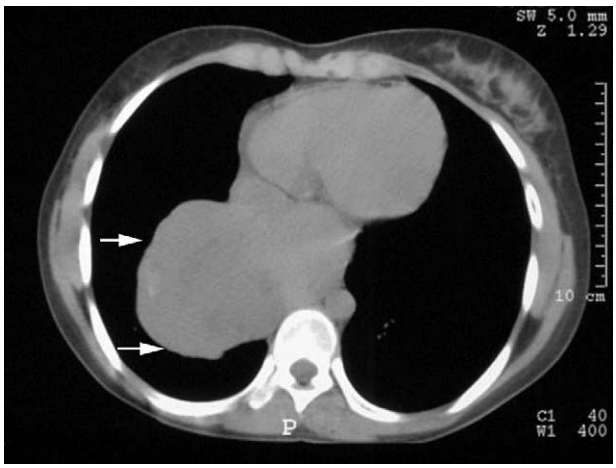

Fig. 2 CT scan of the 15-year-old girl confirms the presence of a massive distal esophageal leiomyoma (arrows).

Fig. 4 Photograph of the cut edge of the mid esophagus showing diffuse esophageal leiomyomatosis. Note the markedly thickened esophageal wall resulting from hypertrophy of both layers of the muscularis propria.