Esophageal Gastrointestinal Stromal Tumor Presenting as a Mediastinal Mass: a case report

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Although gastrointestinal stromal tumor (GIST) is the most common mesenchymal neoplasm of the gastrointestinal tract, it rarely arises from the esophagus and is thus very rarely the cause of a mediastinal mass. We report a case of esophageal GIST presenting as a mediastinal mass in a 59-year-old male. If such a tumor has extremely exophytic growth, it may result in a huge mediastinal mass without causing dysphagia. The CT features of a huge well-demarcated mass with heterogeneous enhancement and extensive central necrosis may mimic that of a primary sarcoma of the mediastinum. Esophageal GIST should be in the differential diagnosis of a mediastinal mass, even if there is little evidence of luminal involvement of the esophagus demonstrated on imaging.

Key words: Esophageal Neoplasm; Gastrointestinal stromal tumor

Gastrointestinal stromal tumors (GIST) arise most commonly from the stomach but very rarely from the esophagus [1, 2]. Most esophageal mesenchymal tumors are leiomyomas, and GISTs are rare in this organ [3]. We have found only a few reports of esophageal GIST, and the imaging features have not been documented in detail [3, 4, 5]. Understandably, therefore, esophageal GIST would not easily come to mind when a patient presents with a mediastinal mass. We describe the clinical and imaging features of such a case.

CASE REPORT

A 59-year-old male was admitted with the symptoms of left back pain for 3 months, weight loss of 10 kg over 6 months, and cough and dyspnea for 2 weeks. There was no dysphagia, nausea, vomiting, fever, or hematochezia. Decreased breath sounds were heard in the left chest. There was a palpable mass in the left upper abdomen. Laboratory tests were within normal range.

A chest radiograph showed a huge mass occupying the left middle and lower hemithorax associated with left pleural effusion (Fig. 1). Chest CT showed a left-sided mediastinal mass located between the esophagus and descending thoracic aorta that measured approximately $15 \times 16 \times 15$ cm. The tumor was well delineated and contained small area of high attenuation indicating focal hemorrhage (Fig. 2a). Contrast-enhanced images revealed heterogeneous enhancement of the tumor with extensive central low attenuation (Fig. 2b). There was no mediastinal lymphadenopathy. The mass effect of the tumor resulted in anterior and upward displacement of the heart and the left lower lobe of the lung and downward displacement of the left hemidiaphragm (Fig. 2). The tumor also caused compression of the lower esophagus, and a small portion of the tumor seemed to protrude into the esophageal lumen (Fig. 2b, 2c). Based on the CT findings, a primary sarcoma of mediastinum was the preoperative diagnostic impression.
After the imaging study, the patient underwent percutaneous needle biopsy of the mass. Microscopically, it was a stromal tumor composed primarily of spindle cells with elongated cigar-shaped nuclei arranged in fascicles and whorls (Fig. 3a). Immunohistochemical staining was strongly and diffusely positive for CD117 (c-kit) (Fig. 3b) and CD34 protein. The tumor cells were negative for SM-actin, desmin, α-1-antitrypsin and S-100 protein. The histologic features and staining pattern were consistent with GIST, and the esophagus was the most likely origin. At surgery, the tumor was seen to arise from muscular layer of the distal esophagus, confirming the diagnosis of esophageal GIST.

DISCUSSION

GIST is the most common subset of mesenchymal tumors of the gastrointestinal tract [1-2]. It arises most commonly from stomach (60% to 70%), followed by small bowel (20% to 25%), anorectum (7%), and, rarely, colon and esophagus [2]. In stomach, small intestine, colon, and anorectum, GISTs account for almost all mesenchymal tumors, since leiomyomas and leiomyosarcomas in these sites are very rare. The esophagus is the only site where leiomyomas predominate [1].

In a series of 68 esophageal stromal tumors reported by Miettinen, et al., GISTs accounted for 17, compared with 48 leiomyomas and 3 leiomyosarcomas. The median age for patients with GIST was 63, versus 35 for patients with leiomyomas [3].

The presenting signs and symptoms of esophageal GIST depend on the size and location of the tumor. Typically, they cause dysphagia, suggesting the possibility of carcinoma [3]. Less commonly they are detected as large mediastinal masses involving the esophagus. Other manifestations include cough, gastrointestinal bleeding, and weight loss. Occasionally, it may be an incidental finding.

GISTs arising from the stomach or small bowel most commonly have an exophytic growth pattern and manifest as masses prominently outside the organ of origin. Dominant intramural and intraluminal masses are less common manifestations. Endoscopy and barium study classically show a smooth, discrete submucosal mass, usually with intact overlying mucosa, or occasionally with central mucosal ulceration. A shortcoming of endoscopy and barium study is that they tend to underestimate the lesion size for those predominantly submucosal or exenteric GISTs. CT is commonly used in delineating the full extent of tumors as well as to detect local invasion and distant metastasis. GISTs characteristically have hemorrhage, necrosis, or cyst formation that appears as focal areas of low attenuation on CT images. Small lesions are usually homogeneous with attenuation values similar to those of muscle [7].

There is little information in the literature describing the radiologic appearance of esophageal GISTs. In Miettinen’s series, esophageal GISTs ranged from 2.6 to 25 cm in size and were most commonly located in the distal third of the esophagus [3]. Levy et al. described the radiologic findings in 64 GISTs, in which there was only one located in the esophagus. This was a distal esophageal mass that distorted and widened the esophageal lumen on barium esophagram. Based on the general appearance of GIST arising from the stomach or small bowel, they suggested that an esophageal GIST might appear as a smooth intramural mass or a large ulcerated mass on barium study, while on CT it might have homogeneous or heterogeneous attenuation, including central areas of hemorrhage, necrosis, or cystic degeneration [6].

Levy et al. also stated that “The radiologic differential diagnosis for esophageal GISTs is based on size and growth pattern. Small lesions confined to the esophageal wall are more likely to represent leiomyomas. Other lesions of submucosal origin, such as duplication cyst, lipoma, and hemangioma should also be considered. GISTs that extend into the esophageal lumen may simulate a polypoid mass of mucosal
Esophageal GIST presenting as a mediastinal mass

Figure 2. Chest CT revealed a huge left-sided mediastinal mass with massive pleural effusion. 

a. Unenhanced axial image showed area of high attenuation in the tumor (arrow), indicating focal hemorrhage.

b. Contrast-enhanced axial image demonstrated well defined lobulated mass with heterogeneous enhancement and extensive central low attenuation. The tumor located between the esophagus and descending thoracic aorta, and caused anterior displacement of the heart and compression of the esophagus.

c. Contrast-enhanced coronal image revealed upward displacement of the heart and collapsed left lower lobe of lung. The tumor also resulted in downward displacement of left hemidiaphragm. A small portion of the tumor seemed to protrude into the esophageal lumen (arrowhead in b and c).

Figure 3. Histologic examination of percutaneous needle biopsy.

a. The tumor composed primarily of spindle cells with elongated cigar-shaped nuclei arranged in fascicles and whorls (H&E stain, x100; right lower quarter, x400).

b. Immunohistochemical staining (x400) demonstrated strong and diffuse positive for CD-117.
origin, like papilloma, adenoma, fibrovascular polyp, and carcinoma. Large, highly aggressive GISTs that extend into the mediastinum have radiologic appearances similar to those of advanced stage carcinoma and leiomyosarcoma” [6]. In our case, the tumor had an extremely exophytic growth pattern and therefore didn’t cause dysphagia. For this reason, we initially suspected a mediastinal rather than an esophageal tumor. Mediastinal tumors that may have a similar CT appearance of a huge well-demarcated mass with heterogeneous enhancement and extensive central necrosis include primary sarcoma of the mediastinum, tumors of neural origin, and lymphoma. The absence of mediastinal lymphadenopathy made the diagnosis of lymphoma less likely. The location of the tumor anterior to the descending aorta decreased the odds of it being a neurogenic tumor. However, its appearance was consistent with a primary sarcoma of the mediastinum, including leiomyosarcoma, spindle cell sarcoma, or malignant fibrous histiocytoma [8]. The slight luminal involvement of the esophagus by the tumor demonstrated on CT (Fig. 2b, 2c) is probably the only clue that might lead to the consideration of a submucosal esophageal tumor.

With extremely exophytic growth, esophageal GIST may result in a huge mediastinal mass without causing dysphagia. Esophageal GIST should be considered in the differential diagnosis of a mediastinal mass, particularly if there is evidence of luminal involvement of the esophagus.

REFERENCES
食道的胃腸道間質細胞腫瘤以縱膈腔腫瘤表現：病例報告

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雖然胃腸道間質細胞腫瘤是胃腸道最常見的間質腫瘤，但是它很少發生在食道，也因此很少是造成縱膈腔腫瘤的原因。在此，我們提出一個59歲男性以縱膈腔腫瘤表現的食道胃腸道間質細胞腫瘤。當整個腫瘤幾乎都向外生長時，食道胃腸道間質細胞腫瘤可以長成相當大且擴大的縱膈腔腫瘤而不造成病人吞嚥上的症狀；此時電腦斷層影像上的特徵可能無法與縱膈腔的惡性肉瘤區分。因此，雖然影像上食道管腔內腫瘤的侵犯很少，我們也必須將食道胃腸道間質細胞腫瘤列入縱膈腔腫瘤的鑑別診斷中。

關鍵詞：食道的胃腸道間質細胞腫瘤；胃腸道間質細胞腫瘤；縱膈腔腫瘤