Loss of peristalsis of the esophagus due to diffuse esophageal leiomyomatosis

A 15-year-old girl was referred to our hospital with a tumor in the mediastinum that had been found on a chest radiograph during a medical check-up. An esophagram showed a large mediastinal mass (60 mm in diameter) (▶ Video 1). No peristalsis of the thoracic esophagus (smooth muscle area) was apparent, and dilatation of the esophagus and retention of the barium were observed on the proximal side of the tumor. High resolution manometry (HRM) findings showed complete loss of peristalsis of the smooth muscle area, whereas peristalsis of the striated muscle area was preserved (▶ Fig. 1). Although the esophageal lumen was narrowed by the tumor, the endoscope could be passed through it (▶ Fig. 2). Computed tomography (CT) scanning demonstrated a mildly enhancing tumor with a diameter of 60 mm, which had circumferentially surrounded the lower esophagus. The tumor was not invading other organs (▶ Fig. 3). Biopsy specimens

The patient underwent endoscopic ultrasound-guided fine needle aspiration (EUS-FNA), with the endoscopic ultrasound showing that the tumor had a mosaic echo, and that there was a clear boundary between the tumor and other organs (▶ Fig. 5). Biopsy specimens

▶ Video 1: Results of the esophagram showing a large mediastinal tumor with a diameter of 60 mm. The esophageal lumen is narrowed by the tumor, and there is no peristalsis of the smooth muscle area visible. Dilatation of the esophagus and retention of the barium are observed on the proximal side of the tumor.
showed spindle cells without atypia on hematoxylin and eosin (H&E) staining; they were positive for smooth muscle actin and desmin, and negative for c-kit by immunohistochemistry (▶ Fig. 6). A diagnosis of diffuse esophageal leiomyomatosis (DEL) was made but, as she was asymptomatic, surgery was not indicated.

DEL is a rare, benign disease characterized by diffuse hypertrophy of the esophageal muscular layer; some cases of DEL are associated with Alport syndrome [1–4]. Although patients with DEL usually present with dysphagia [1,2], our patient was asymptomatic with no signs of Alport syndrome. Esophageal motility in DEL investigated by HRM has rarely been reported [5]. This is the first report showing loss of peristalsis of the esophagus due to DEL, although the mechanism of this remains unknown.

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Competing interests

None

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