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.

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...
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.

Endoscopy_UCTN_Code_CCL_1AB_2AC_3AH

Competing interests

None

The Authors

Kazuya Takahashi, Yui Ishii, Kazunao Hayashi, Satoshi Ikarashi, Hirokazu Kawai, Yuichi Sato, Shuji Terai
Division of Gastroenterology and Hepatology, Graduate School of Medical and Dental Science, Niigata University, Niigata, Japan

Corresponding author

Kazuya Takahashi, MD, PhD
Division of Gastroenterology and Hepatology, Graduate School of Medical and Dental Science, Niigata University, 1-757 Asahimachi-dori, Chuo-ku, Niigata City, Niigata, 951-8510, Japan
Fax: +81-25-2270776
kazuya911@med.niigata-u.ac.jp

References


Bibliography

DOI http://dx.doi.org/10.1055/s-0043-100691
Endoscopy 2017; 49: E95–E96
© Georg Thieme Verlag KG
Stuttgart · New York
ISSN 0013-726X