Esophageal smooth muscle tumor in a 25-year-old woman with congenital malformations

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We recently treated a 25-year-old woman with an esophageal smooth muscle tumor with congenital malformations. Although the large size of the tumor and the presence of hemorrhagic lesion suggested the tumor to be leiomyosarcoma, histological studies revealed it to be leiomyoma. According to previous reports in the English-language literature, the coincidence of esophageal smooth muscle tumor with congenital malformations is relatively rare, and the coincidence of such a tumor with malformations of the type seen in this patient has never been reported. The congenital malformations in our patient were ocular hypertelorism, platyrrhiny, bilateral divergent strabismus, clubbed fingers and toes, fingerprint abnormality, and mild mental retardation. These congenital malformations cannot be explained by any reported syndromes.

Key words: esophagus, leiomyoma, congenital malformation

Introduction

Although leiomyoma is a commonly seen benign tumor in the esophagus, its incidence is much lower than that of carcinoma of the esophagus.1 According to previous reports,1–3 the occurrence of leiomyoma is male-dominant, the age distribution is about equal in each decade from 20 to 60 years, and almost the half of the patients are asymptomatic. However, the occurrence of this tumor in an earlier decade of life may indicate the presence of some hereditary or circumstantial factor. There have been few reports about the coincidence of congenital malformations and leiomyoma,4–6 and only one report of gastrointestinal tract leiomyosarcoma coincidental with congenital malformations, suggesting the association of the smooth muscle tumor with some kinds of genetic anomalies.7

Here we report a case of esophageal smooth muscle tumor in a 25-year-old woman with congenital malformations that could not be explained by any known syndromes.

Case report

A 25-year-old woman was seen because of right lower chest pain of 2 months, duration. The pain had not changed in strength and had disappeared after she took analgesics. There were no other symptoms referable to the gastrointestinal system. On admission, her general status was good. On physical examination, several congenital malformations were found; ocular hypertelorism, strabismus, clubbed fingers and toes, fingerprint abnormality, and mild mental retardation. These congenital malformations cannot be explained by any reported syndromes.

Results of routine laboratory tests were essentially normal. Chromosomal analysis showed no abnormalities. Abdominal ultrasonography revealed a heterogeneous round mass on the back of the left lobe of the liver (Fig. 1). Posteroanterior and lateral roentgenograms of the chest showed a large, well circumscribed mass in the lower lung field. An esophagogram demonstrated extrinsic compression in the distal esophagus (not shown). Upper gastrointestinal endoscopy showed intact esophageal mucosa compressed externally from the right anterior (not shown). A computed tomogram of the chest demonstrated a large mediastinal mass (Fig. 2). The esophagus at the margin of the huge mass was...
round, indicating that this mass was of esophageal origin. There was neither calcification nor central necrosis in the tumor. In a frontal section on magnetic resonance imaging, the tumor appeared to be present on the diaphragm cone, and protruded into the abdominal cavity through the hiatus (Fig. 3). On sagittal section, the tumor compressed the liver forward, and separated the aorta and the inferior vena cava. The tumor was spindle-shaped along the esophagus. Angiography showed a hypervascular tumor around the epigastrium (Fig. 4).

After the above examinations, tumor resection was performed. The tumor was a 12.6 × 9.0 × 5.5-cm irregular mass adhering to a 2.5 × 2.0-cm portion of the esophagus, and located in the proper muscle, with ulceration in the lumen. No lymph node metastasis was found. On cross section, the tumor had a firm, fish-flesh appearance and had areas of necrosis and hemorrhage.