Endoscopic submucosal dissection for a rare cause of esophageal mass: Von Recklinghausen's neurofibromatosis





▶ Fig. 1 a Cutaneous manifestations of neurofibromatosis observed during physical examination. b Upper endoscopy showing a mass originating from the lower esophagus to the gastric cardia. c Hypoechoic, submucosally-originating mass with multiple anechoic projections into the esophageal cavity, as revealed by endoscopic ultrasound. d Complete removal of the mass via endoscopic submucosal dissection. e The resected specimen. f Histological analysis confirming the presence of neurofibromatosis.

A 72-year-old woman was admitted to our hospital with a 20-year history of intermittent subxiphoid pain, which intensified in the past 2 months. Physical examination revealed dermal manifestations suggestive of neurofibromatosis (> Fig. 1 a). Enhanced computed tomography identified thickening of the distal esophageal wall. During upper endoscopy, a mass was observed stretching from the lower esophagus to the gastric cardia, leading to partial esophageal obstruction (> Fig. 1 b). Endoscopic ultrasound depicted a hypoechoic submucosal mass, distinguished by several anechoic lesions extending into the esophageal lumen (**Fig. 1 c**). With informed consent from the patient, the mass was successfully excised using endoscopic submucosal dissection (▶ Fig. 1 d, ▶ Video 1). The extracted specimen measured 3.0×5.0× 5.0 cm (**Fig. 1e**). Histopathological evaluation confirmed neurofibromatosis, identified by neurofibromatous cells encircling the specimen (▶ Fig.1 f). The patient experienced no symptoms and showed no signs of recurrence or residual mass on follow-up endoscopic evaluations over 1 year.

Von Recklinghausen's neurofibromatosis, an autosomal dominant condition, is generally characterized by dermal neurofibromas and distinctive café-au-lait spots. Though predominantly affecting the jejunum, stomach, ileum, and colorectum, esophagus involvement is scarce [1, 2]. To the best of our knowledge, this is the inaugural report of an esophageal neurofibromatosis case managed with the ESD technique, demonstrating its safety and efficacy. This case underscores the need for endoscopists to be vigilant for signs of gastrointestinal neurofibroma, particularly in patients with dermal nodules, and suggests that endoscopic approaches like endoscopic submucosal dissection can provide an accurate diagnosis and effective treatment.



Video 1 Endoscopic submucosal dissection for a rare cause of esophageal mass: Von Recklinghausen's neurofibromatosis.

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Conflict of Interest

The authors declare that they have no conflict of interest.

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