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The *WJCC* is now abstracted and indexed in Science Citation Index Expanded (SCIE, also known as SciSearch®), Journal Citation Reports/Science Edition, Current Contents®/Clinical Medicine, PubMed, PubMed Central, Scopus, Reference Citation Analysis, China National Knowledge Infrastructure, China Science and Technology Journal Database, and Superstar Journals Database. The 2022 Edition of Journal Citation Reports® cites the 2021 impact factor (IF) for *WJCC* as 1.534; IF without journal self cites: 1.491; 5-year IF: 1.599; Journal Citation Indicator: 0.28; Ranking: 135 among 172 journals in medicine, general and internal; and Quartile category: Q4. The *WJCC*'s CiteScore for 2021 is 1.2 and Scopus CiteScore rank 2021: General Medicine is 443/826.

RESPONSIBLE EDITORS FOR THIS ISSUE

Production Editor: *Ying-Yi Yuan*; Production Department Director: *Xiang Li*; Editorial Office Director: *Jin-Lei Wang*.

NAME OF JOURNAL

World Journal of Clinical Cases

ISSN

ISSN 2307-8960 (online)

LAUNCH DATE

April 16, 2013

FREQUENCY

Thrice Monthly

EDITORS-IN-CHIEF

Bao-Gan Peng, Jerzy Tadeusz Chudek, George Kontogeorgos, Maurizio Serati, Ja Hyeon Ku

EDITORIAL BOARD MEMBERS

<https://www.wjgnet.com/2307-8960/editorialboard.htm>

PUBLICATION DATE

September 26, 2022

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INSTRUCTIONS TO AUTHORS

<https://www.wjgnet.com/bpg/gerinfo/204>

GUIDELINES FOR ETHICS DOCUMENTS

<https://www.wjgnet.com/bpg/GerInfo/287>

GUIDELINES FOR NON-NATIVE SPEAKERS OF ENGLISH

<https://www.wjgnet.com/bpg/gerinfo/240>

PUBLICATION ETHICS

<https://www.wjgnet.com/bpg/GerInfo/288>

PUBLICATION MISCONDUCT

<https://www.wjgnet.com/bpg/gerinfo/208>

ARTICLE PROCESSING CHARGE

<https://www.wjgnet.com/bpg/gerinfo/242>

STEPS FOR SUBMITTING MANUSCRIPTS

<https://www.wjgnet.com/bpg/GerInfo/239>

ONLINE SUBMISSION

<https://www.f6publishing.com>

Misdiagnosis of an elevated lesion in the esophagus: A case report

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Specialty type: Medicine, research and experimental

Provenance and peer review:

Unsolicited article; Externally peer reviewed.

Peer-review model: Single blind

Peer-review report's scientific quality classification

Grade A (Excellent): 0

Grade B (Very good): B

Grade C (Good): C, C, C

Grade D (Fair): D

Grade E (Poor): 0

P-Reviewer: Mohamed SY, Egypt; Nakamura K, Japan; Suresh Kumar VC, United States; Villa E, United States

Received: April 20, 2022

Peer-review started: April 20, 2022

First decision: June 19, 2022

Revised: June 30, 2022

Accepted: August 15, 2022

Article in press: August 15, 2022

Published online: September 26, 2022



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Abstract

BACKGROUND

Esophageal carcinosarcoma (ECS) is a rare biphasic tumor and a type of esophageal malignancy, which presents as protruding or elevated lesions. ECS patients are often not hospitalized until they have severe dysphagia. ECS is easily misdiagnosed as a benign tumor due to its atypical characteristics under endoscopy. With the popularization of endoscopic treatment, these patients are often referred to endoscopic treatment, such as endoscopic submucosal dissection (ESD). However, there is a lack of consensus on the endoscopic features and therapies for ECS. Here, we report a case of ECS and discuss the value of endoscopic diagnosis and therapeutic strategies.

CASE SUMMARY

A 63-year-old man was admitted to the hospital with dysphagia. During the endoscopic examination, an elevated lesion was found with an erosive and hyperemic surface covered with white pseudomembranous inflammation. Endoscopic ultrasonography (EUS), biopsies, and enhanced thoracic computed tomography were performed, suggesting that it was a benign lesion and located within the submucosal layer. This lesion was diagnosed as a fibrovascular polyp with a Paris classification of 0-Ip. The patient was then referred to ESD treatment. However, the post-ESD pathological and immunohistochemical study showed that this lesion was ECS with a vertical positive margin (T1b stage), indicating that we made a misdiagnosis and achieved a noncurative resection. Due to the potential tumor residue, additional open surgery was performed at the patient's request. In the postoperative pathological study, no tumor remnants or metastases were discovered. The patient was followed for 1 year and had no recurrence.

CONCLUSION

ECS can be misdiagnosed at the initial endoscopy. EUS can help to identify the tumor stage. Patients with T1b stage ECS cannot be routinely referred to ESD

treatment due to the high risk of metastasis and recurrence rate.

Key Words: Esophageal carcinosarcoma; Misdiagnosis; Endoscopic ultrasonography; Endoscopic submucosal dissection; T1 stage; Case report

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Core Tip: Esophageal carcinosarcoma (ECS) is a rare type of esophageal malignancy. ECS commonly presents as a pedunculated characteristic (0-Ip), which is often misdiagnosed due to the lack of specific features. Endoscopic ultrasonography can help to evaluate whether ECS invasion is within the submucosal layer (T1 or T2 stage) but cannot further distinguish whether it is T1a or T1b stage. Due to the high risk of metastasis and recurrence based on the literature review, endoscopic submucosal dissection treatment cannot be routinely recommended for ECS patients with T1b stage disease.

Citation: Ma XB, Ma HY, Jia XF, Wen FF, Liu CX. Misdiagnosis of an elevated lesion in the esophagus: A case report. *World J Clin Cases* 2022; 10(27): 9828-9833

URL: <https://www.wjgnet.com/2307-8960/full/v10/i27/9828.htm>

DOI: <https://dx.doi.org/10.12998/wjcc.v10.i27.9828>

INTRODUCTION

Patients routinely undergo endoscopic evaluation for dysphagia, during which protruding or elevated lesions are frequently found. Some of the lesions are presented as pedunculated lesions, including esophageal adenoma, inflammatory polyps, fibrovascular polyps, carcinosarcoma[1-3], *etc.* However, they lack specific features and have similar endoscopic ultrasonography (EUS) characteristics. Therefore, it is difficult to conclusively diagnose the lesion without the support of postsurgical pathology.

Herein, we report a rare case of esophageal carcinosarcoma (ECS), which was assessed as a benign tumor and treated by endoscopic submucosal dissection (ESD). Nevertheless, post-ESD pathology indicated that it was preoperatively misdiagnosed. Therefore, we systematically evaluated the endoscopic and clinicopathological characteristics of ECS and analyzed the feasibility of endoscopic treatment.

CASE PRESENTATION

Chief complaints

Dysphagia for 3 mo.

History of present illness

A 63-year-old man was admitted to the hospital with dysphagia for 3 mo. The patient can only swallow semi-solid food for 2 wk, with intermittent swallowing pain.

History of past illness

The patient was in good health in the past.

Personal and family history

The patient had no personal and family history.

Physical examination

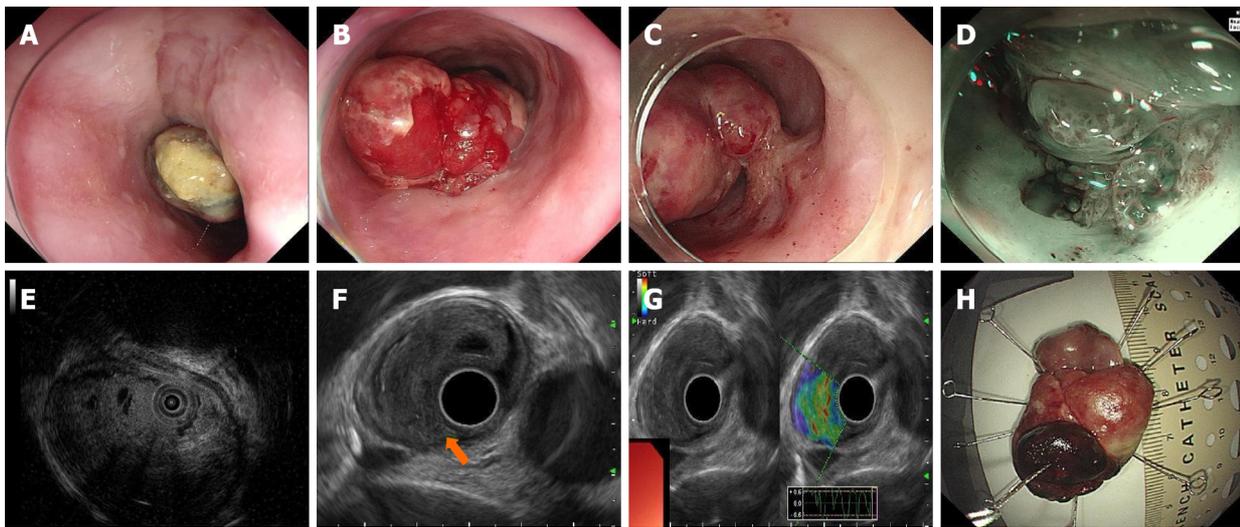
The patient was in good condition. The physical examination was completely normal.

Laboratory examinations

Routine laboratory tests were all within the normal range.

Imaging examinations

During the endoscopic examination, an elevated lesion with an erosive and hyperemic surface covered with white pseudomembranous inflammation was found. It had a short peduncle connected to the mid-



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Figure 1 Endoscopic features of the lesion. A: The oral side of the esophageal carcinosarcoma (ECS). The surface was covered with white pseudomembranous inflammation; B: The body of the ECS. The surface was hyperemic and eroded; C: The short peduncle connected to the wall of the esophagus; D: The peduncle component revealed by narrow-band imaging; E: Ultrasonic mini-probe; F: Endoscopic ultrasonography revealed that the origin of the ECS was from the submucosal layer and the inherent muscle layer was clear; G: Ultrasonic elastography revealed that the lesion was blue-green, with a tough texture; H: Macroscopic findings of the resected specimen.

esophagus wall and was 30 mm × 40 mm in size. EUS revealed a lesion derived from the submucosal layer with an intact inherent muscle layer, and this lesion was hypoechoic and consisted of internal multicystic components. Ultrasonic elastography revealed that the lesion was blue-green, indicating a tough texture (Figure 1). Multiple biopsies showed necrosis and active fibroblast proliferation. An enhanced thoracic computed tomography scan showed a protuberant lesion in the middle of the esophagus, suggesting a benign tumor. A multidisciplinary consultation was performed, and we preliminarily diagnosed this lesion as a fibrovascular polyp.

FINAL DIAGNOSIS

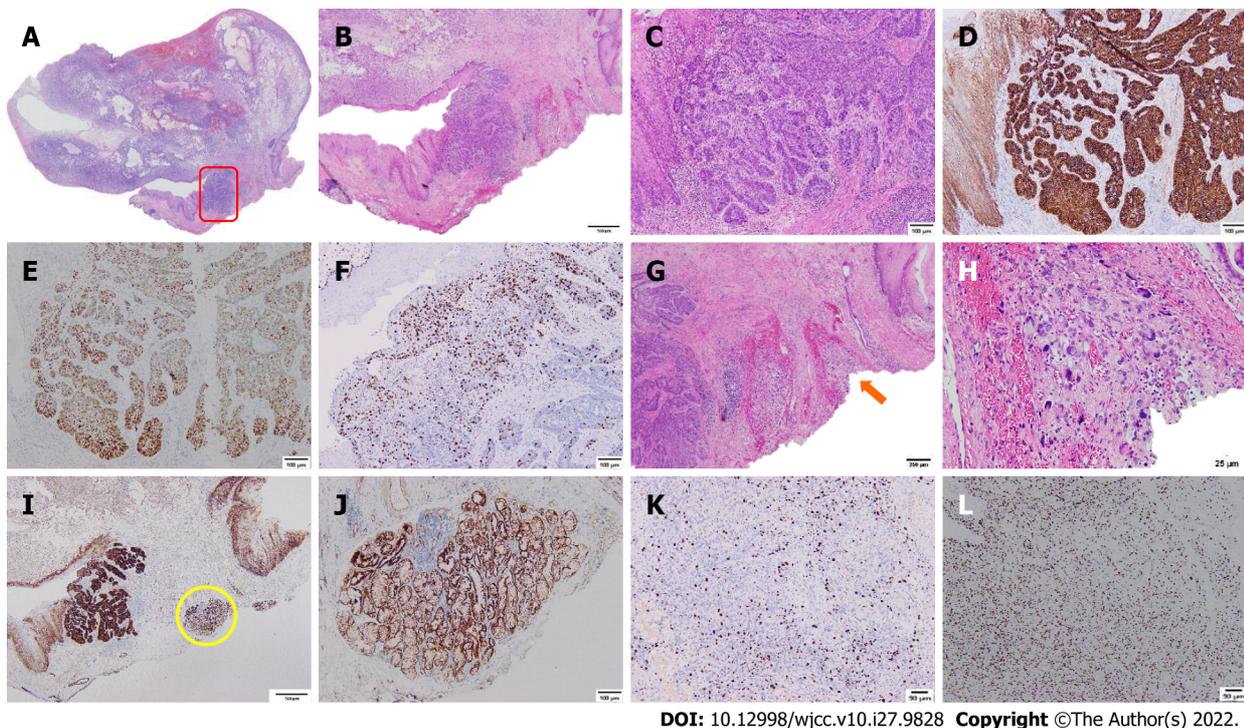
The post-ESD pathological study showed that this lesion was composed of a malignant fibroblast apoptosis component and a basal-like squamous cell carcinoma (BSC) component, indicating ECS. The polypoid mass was predominantly composed of malignant fibrous histiocytoma with a vertical positive margin, horizontal negative margin, and no evidence of vascular or lymphatic invasion. Immunohistochemical (IHC) staining showed CK (-), vimentin (+), CD68 (+), β-catenin (+), p53 (+), S-100 (-), CD34 (-), SMA (-), desmin (-), Twist1 (+), ZEB1 (+), Snai2 (-), PDGFR alpha (-), and a Ki-67 index of 20%. BSC was observed in the neck of the tumor, and its vertical and horizontal margins were negative. IHC staining showed CK (+), E-cadherin (+), p53 (+), vimentin (-), S-100 (-), CD34 (-), CD68 (-), SMA (-), desmin (-), and a Ki-67 index of 30% (Figure 2).

TREATMENT

The following ESD treatment was successful with no obvious adhesion in the submucosal layer after the patient's informed consent was obtained.

OUTCOME AND FOLLOW-UP

Due to the potential tumor residue, additional open surgery was performed at the patient's request. No tumor remnants or metastases were discovered in the postoperative pathological study. The patient was followed for 1 year and had no recurrence.



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Figure 2 Histological and immunohistochemical findings of the esophageal carcinosarcoma. A: Histological mapping of the esophageal carcinosarcoma; B and C: Hematoxylin-eosin staining of the basal-like squamous cell carcinoma (BSC) component (red rectangle, B \times 20, C \times 100); D: β -catenin staining of the BSC component (positive, \times 100); E: P53 staining of the BSC component (positive, \times 100); F: Ki-67 staining of the BSC component (30%, \times 100); G and H: Hematoxylin-eosin staining of the sarcoma component (orange arrow, G \times 40, H \times 400); I and J: Immunopositive carcinomatous cells for β -catenin are closely adjacent to the invasion depth (yellow circle, I \times 20, J \times 100); K: Ki-67 staining of the sarcoma component (30%, \times 100); L: P53 staining of the sarcoma component (positive, \times 100).

DISCUSSION

ECS is a rare biphasic tumor that accounts for 0.2%-2.8% of all esophageal malignancies. It is characterized by the presence of both malignant epithelial and mesenchymal components[4]. ECS usually presents as a large intraluminal polypoid mass on the upper and middle esophagus, with a median diameter of 55-75 mm. The endoscopic features of this lesion may include a hyperemia surface, erosion, ulceration, brittleness, and easy bleeding, which lack specificity for endoscopic diagnosis[5].

The diagnosis of ECS mainly relies on pathological studies[4]. However, untargeted endoscopic biopsies of this lesion usually reveal components of sarcoma, which makes it easily misdiagnosed. Efforts can be made to potentially improve the biopsy accuracy by targeting the root or peduncle as the epithelial cancer component always exceeds the mass in the range[6].

EUS evaluation of the lesion plays a role in the assessment before treatment. Although lacking specificity in diagnosis, EUS can provide information on invasion depth. According to a report from Taiwan[7], five of six ECS patients were correctly assessed on the invasion depth by EUS. However, all lesions were in the deep invasion (T2 stage). Our preoperative EUS showed that the origin of the ECS was derived from the T1 stage, which was proven by postoperative pathology. However, we also noticed that EUS could not further distinguish whether it was T1a or T1b stage. The reason could be that the echo of sarcoma that invaded the submucosa was similar to the original interstitial composition and therefore could not be distinguished by EUS.

Data on lymph node metastases of ECS at T1 stage are limited[7,8]. In 2006, Sanada *et al*[9] reviewed 57 cases of ECS reported in Japan between 1995 and 2004, among which one was a T1a stage case and 17 were T1b stage cases[9]. Seven (41%) of the T1b stage cases were found to have lymph node metastasis compared with none of the T1a stage cases. In 2021, Chen *et al*[10] reported that none of the ten ECS patients at T1 stage were found to have lymph node metastasis, with no report of T1 subtypes[10]. Since lymph node metastasis is related to prognosis, a detailed assessment is required before treatment.

Data on the prognosis of ESD treatment for ECS in the T1 stage are also limited. One Korean case reported by Cha *et al*[11] in 2014 is very similar to ours[11]. The lesion located within the submucosal layer without evidence of metastasis was treated by ESD. The post-ESD pathological study reported ECS with a vertical positive margin (T1b stage). In contrast, the patient from this Korean study refused to receive additional surgery, and a recurrence was found during an endoscope examination 21 mo later. Two Chinese cases were also treated by ESD, and one case was followed by additional surgery. Unfortunately, neither of them had long-term follow-ups[12,13]. Therefore, robust data on the prognosis

of ESD for ECS are needed.

CONCLUSION

We report a rare case of ECS with BSC, which can be misdiagnosed due to the lack of specific characteristics. Targeted biopsies on the root or peduncle after observation by narrow-band imaging or iodine staining may potentially improve the diagnostic accuracy. EUS can help to evaluate the layer of the origin (T1 or T2 stage) but cannot further distinguish whether it is at the T1a or T1b stage. ESD treatment should not be routinely recommended to ECS patients with T1b stage disease due to the risk of metastasis and high recurrence rate.

FOOTNOTES

Author contributions: Ma XB and Ma HY reviewed the literature and contributed to manuscript drafting; Jia XF was the patient's EUS and ESD surgeon; Wen FF was involved in pathology evaluation; Liu CX was responsible for revising the manuscript for important intellectual content; all the authors provided final approval for the version of the manuscript to be submitted.

Informed consent statement: Written informed consent was obtained from the patient for the publication of this report and any accompanying images.

Conflict-of-interest statement: All the authors report no relevant conflicts of interest for this article.

CARE Checklist (2016) statement: The authors have read the CARE Checklist (2016), and the manuscript was prepared and revised according to the CARE Checklist (2016).

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Country/Territory of origin: China

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S-Editor: Fan JR

L-Editor: Wang TQ

P-Editor: Fan JR

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