Discover Oncology

Case Report

Rare rectal elastofibroma: diagnostic challenges and case report

Chen-Xin Xiang[1,2] · Yu Long[1,2] · Yi-Ning Xiang[3] · Fei Huang[1,2] · Yun Ke[1,2] · Yi-Ran Yao[4] · Yun-Huan Zhen[1]

Received: 7 February 2025 / Accepted: 2 May 2025
Published online: 19 May 2025

Abstract
Elastofibroma (EF) is a rare soft tissue tumor that typically occurs in the scapular region, with its occurrence in the rectum being extremely uncommon. In this report, we present a case of rectal EF in a 72-year-old female, which was definitively diagnosed through histopathology, immunohistochemistry, and special staining analyses. This case emphasizes the diagnostic challenges posed by rectal EF and underscores the necessity of histopathological and immunohistochemical assessment in distinguishing it from other spindle cell neoplasms, particularly when imaging findings are inconclusive.

Keywords Elastofibroma · Rectum · Neuroendocrine tumors · Special staining · Case report

1 Introduction

Elastofibroma (EF) is a rare and benign soft tissue tumor first described by Jarvi and Saxen in 1961 [1]. EF is most commonly found in the subscapular region; however, recent studies have revealed a more diverse range of locations [2–4]. This case reports the occurrence of EF in the rectum, a site that has been rarely documented. Despite being a benign tumor, EF's unique pathological features and varied locations make it a significant topic in the study of soft tissue pathology.

In preparing this case report, a literature review yielded only one previous report of rectal EF from 1992 [5]. This report aims to enhance awareness of this rare entity by detailing the diagnostic process and surgical outcomes, emphasizing the need for further investigations when imaging studies fail to provide a definitive diagnosis, to avoid missed or incorrect diagnoses.

2 Case presentation

2.1 Chief complaint

A 72-year-old female patient presented to the outpatient clinic with recurrent upper gastrointestinal discomfort accompanied by abdominal pain.

✉ Yun-Huan Zhen, yunhuanzhen72@163.com | [1]Department of Colorectal Surgery, The Affiliated Hospital of Guizhou Medical University, No. 16 Beijing Road, Yunyan District, Guiyang 550001, Guizhou, China. [2]Department of Colorectal Surgery, Guizhou Medical University, Guiyang 550001, Guizhou, China. [3]Department of Pathology, The Affiliated Hospital of Guizhou Medical University, Guiyang 550001, Guizhou, China. [4]Department of Gastroenterology, The Affiliated Hospital of Guizhou Medical University, Guiyang 550001, Guizhou, China.

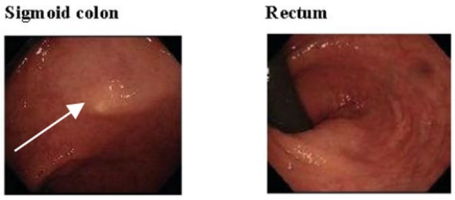

Fig. 1 A submucosal elevation measuring approximately 0.4 cm in diameter is observed 15 cm from the anal verge, appearing slightly yellowish with indistinct borders, as indicated by the arrow

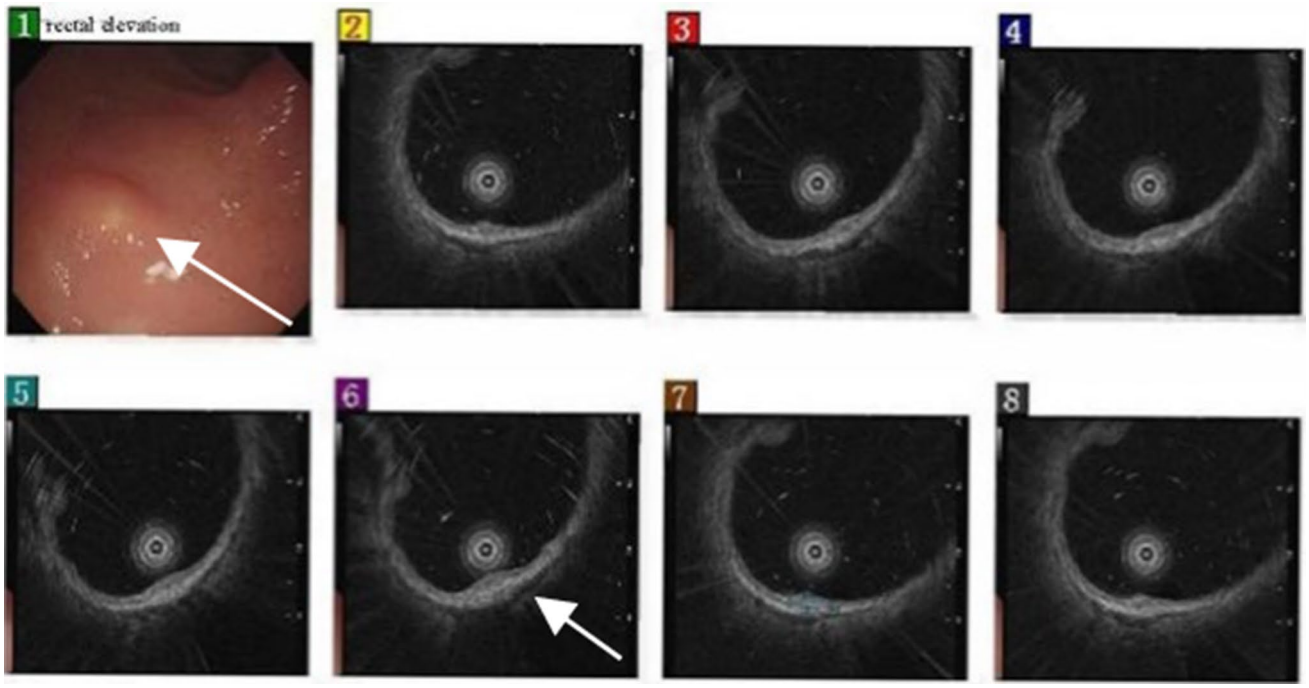

Fig. 2 Endoscopic Ultrasound Findings: White light observation with an endoscope revealed a flat elevation in the rectum approximately 15 cm from the anal verge. The water immersion technique was employed for ultrasound examination at a frequency of 20 MHz, indicating that the rectal elevation originated from a hypoechoic mass in the second layer (submucosa)

2.2 History of present illness

The patient had experienced symptoms for five years, with a marked exacerbation in the past two months. The primary manifestations included intermittent abdominal pain, accompanied by mild nausea and reflux. There were no reports of constipation, hematochezia, or abnormal bowel movements. Due to recurrent upper gastrointestinal discomfort and associated abdominal pain, the patient underwent esophagogastroduodenoscopy (EGD) and colonoscopy. EGD revealed chronic non-atrophic gastritis with antral erosion, while colonoscopy identified multiple polyps and an abnormal submucosal rectal elevation (Fig. 1). To further evaluate the nature, size, and relationship of the rectal submucosal lesion with surrounding structures, endoscopic ultrasound (EUS) was performed. EUS demonstrated a hypoechoic mass located within the mucosal layer, leading to an initial suspicion of neuroendocrine tumor (NET) (Fig. 2).

2.3 Past medical history

The patient had a 10-year history of hypertension, which was well controlled with regular antihypertensive medication. Ten years ago, the patient underwent radical surgery for clear cell renal cell carcinoma (ccRCC) of the right kidney. Postoperative follow-up showed no evidence of recurrence. The patient had no significant family history and denied smoking or alcohol consumption.

2.4 Physical examination

Upon examination, the patient appeared to be in good general condition. Abdominal examination revealed mild tenderness in the left lower quadrant, with no hepatosplenomegaly noted. Bowel sounds were normal, and digital rectal examination was negative.

2.5 Laboratory tests

Laboratory tests revealed a white blood cell (WBC) count of 6.04×10^9/L, hemoglobin (Hb) level of 143 g/L, and platelet (PLT) count of 245×10^9/L. Serum creatinine (Cr) was 93 μmol/L, alanine aminotransferase (ALT) was 38.5 U/L, and aspartate aminotransferase (AST) was 37.9 U/L.

2.6 Imaging studies

Abdominal contrast-enhanced computed tomography (CECT) demonstrated localized thickening of the rectal wall with unclear differentiation from surrounding soft tissue, raising suspicion for a submucosal lesion.

2.7 Final diagnosis

Postoperative pathological examination revealed proliferation of spindle cell clusters and fibrofatty vascular tissue. Immunohistochemical results indicated that the spindle cell clusters were S100 (-), Desmin (-), Caldesmon (-), CD117 (-), CD34 (-), DOG-1 (-), and Ki-67 (<1% +). Special staining results showed elastin fibers (+), Masson's trichrome (+), AB-PAS (-), and PAS (-), confirming the diagnosis of EF (Figs. 3 and 4).

2.8 Treatment

On the second day after admission, the patient underwent endoscopic submucosal dissection (ESD). Endoscopic examination revealed a flat, elevated lesion approximately 15 cm from the anal verge, with a smooth, slightly yellowish surface and indistinct margins (Figure 5).

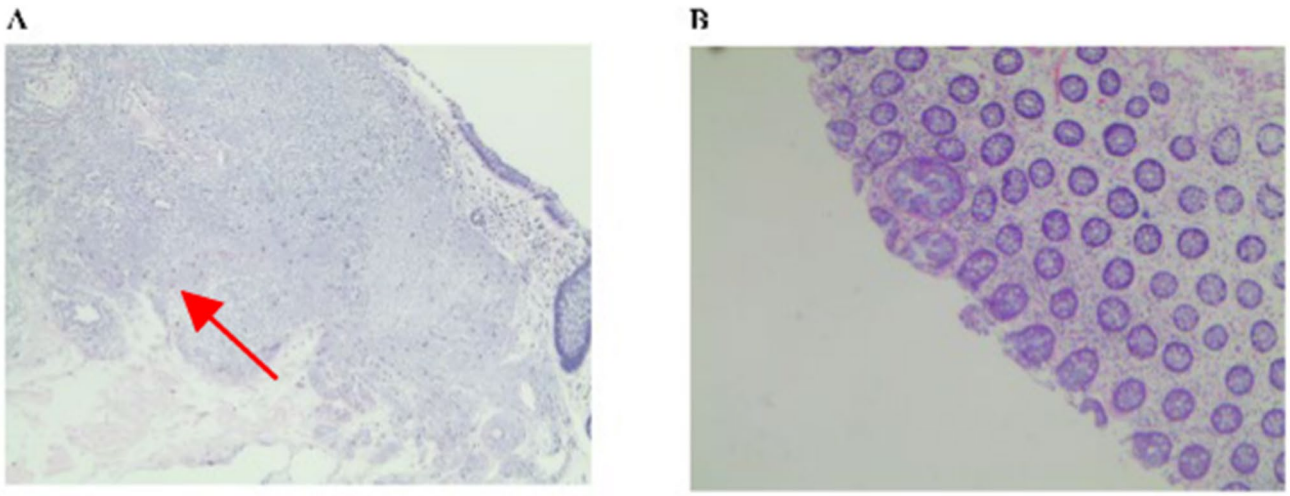

Fig. 3 In **A**, the arrow indicates a dense proliferation of spindle cells, arranged in bundles or interwoven patterns. In **B**, normal glandular structures are observed, with well-organized epithelial cells, and no evident dysplasia or neoplastic changes

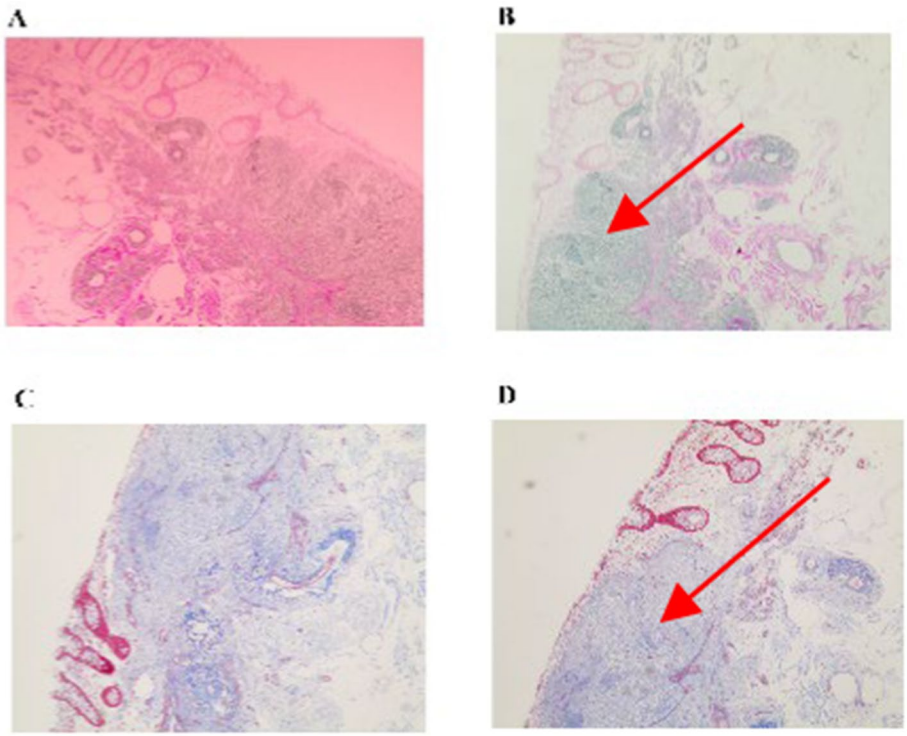

Fig. 4 In the figure, the green-stained area (indicated by the arrow) represents elastin fiber staining, showing a positive result, indicating an abundant presence of elastin fibers in the tissue. The blue-stained area corresponds to Masson's trichrome staining, also positive, demonstrating a large amount of collagen fibers. These staining results support the rich presence of both elastin and collagen fibers within the tissue

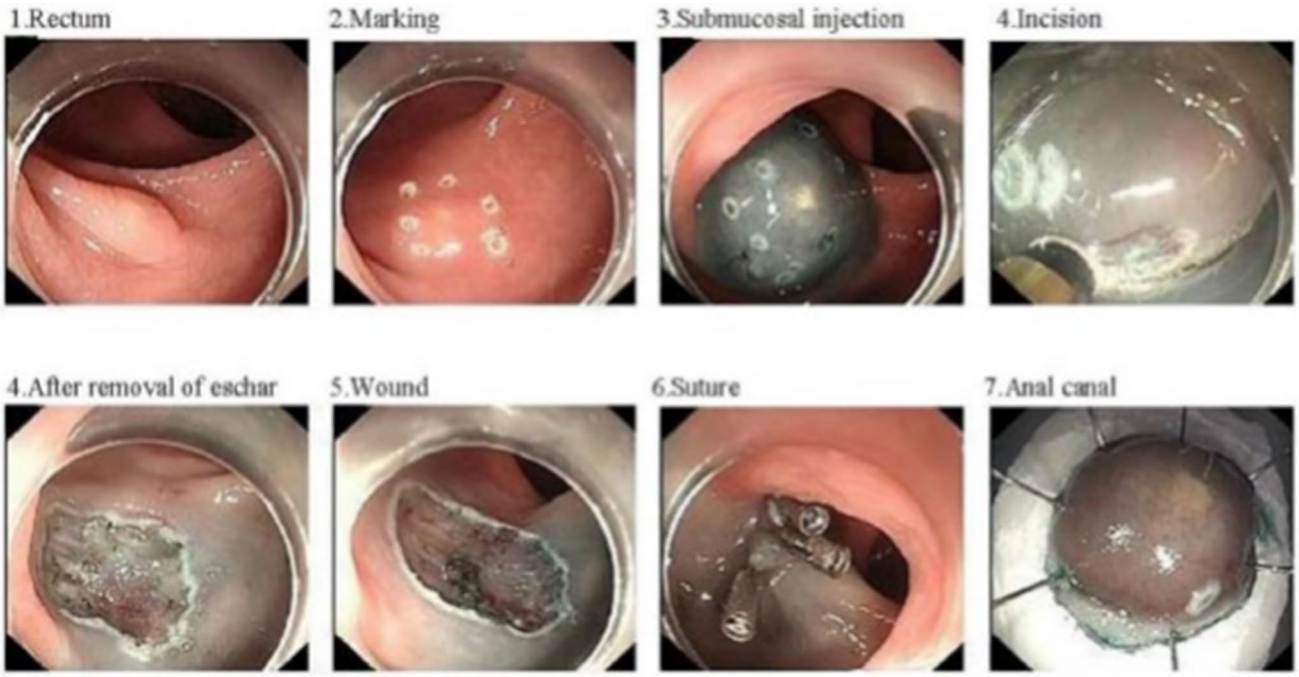

Fig. 5 Electronic Proctoscopy Findings and Endoscopic Submucosal Dissection (ESD) Procedure for Rectal Submucosal Tumor

2.9 Results and follow-up

The patient had an uneventful postoperative recovery, with no reported discomfort. The patient gradually resumed a normal diet and was discharged on postoperative day 3. Follow-up to date has shown no evidence of recurrence or associated symptoms.

3 Discussion

EF is a rare benign soft tissue tumor primarily composed of abnormally proliferated elastic fibers [6]. Compared to normal skin, the elastin in EF exhibits increased resistance to trypsin digestion, with its content reaching twofold and threefold that of normal tissue in wet and dry weight measurements, respectively. Under electron microscopy, EF is predominantly composed of fibroblasts exhibiting ultrastructural features such as rough endoplasmic reticulum and intermediate filaments [7].

Further biochemical analysis has revealed an abnormal amino acid composition in EF elastin, particularly a significant increase in cross-linking-related amino acids, such as desmosine, isodesmosine, and lysine [7]. Altered elastin metabolism, particularly increased cross-linking-related amino acids, may contribute to the pathogenesis of EF.

In the gastrointestinal tract, EF primarily presents as fine granular or fibrous accumulations of elastic fibers, occasionally involving the submucosal layer and/or muscularis mucosae [8]. However, the precise pathogenesis of gastrointestinal EF remains unclear. Some researchers hypothesize that it may be associated with elastic degeneration of submucosal blood vessels followed by fibrosis [9]. Some studies have identified deletions at 1p, 13q, 19p, and 22q, as well as losses of *CASR*, *GSTP1*, and *BRCA2*, along with gains of *APC* and *PAH*. These genomic alterations may be implicated in the pathogenesis and progression of EF [10]. Additionally, fibroblasts within EF have been found to overexpress transforming growth factor-β (TGF-β) and basic fibroblast growth factor (bFGF) [11]. These factors promote fibroblast proliferation and may play a crucial role in tumor development. Given the unique histopathological characteristics of elastofibroma, various staining techniques have been employed for its differentiation, including elastin staining, Masson's trichrome staining, Verhoeff-Van Gieson (VVG) staining, and Orcein staining. Elastin staining is specifically designed to highlight elastic fibers, enabling precise visualization of elastin-rich structures within tissues. Masson's trichrome staining is primarily utilized to differentiate collagen fibers, muscle fibers, and cytoplasmic components, facilitating the distinction between fibrotic and muscular tissues. VVG staining is a specialized histochemical technique that accurately demonstrates elastic fibers, aiding in the identification of elastic tissue abnormalities and distinguishing them from other fibrotic or connective tissue disorders. Orcein staining selectively stains elastic fibers and serves as a valuable tool for excluding other connective tissue diseases, ensuring more precise histopathological differentiation [12–15].

NETs originate from neuroendocrine cells that are widely distributed throughout the gastrointestinal tract and pancreas [16]. NET cells are typically small to medium in size, round or oval in shape, with relatively large nuclei, often exhibiting lobulated or cord-like arrangements. The cytoplasm is scant, transparent, and presents a "foamy" appearance. The cellular arrangement commonly appears in nest-like, island-like, or trabecular patterns, with occasional honeycomb-like structures [17]. Histopathologically, NETs exhibit round to oval tumor cells with characteristic "salt-and-pepper" nuclear chromatin [18].

Endoscopic ultrasound (EUS) is a crucial imaging modality for NETs, offering high resolution and excellent tissue differentiation. On EUS, NETs typically appear as small, well-defined hypoechoic nodules, a characteristic that correlates with their densely packed cellular arrangement and minimal stromal components on histology [19]. Based on these imaging features, NETs were considered in the initial differential diagnosis of this case.

Regarding treatment, the approach depends on tumor location, size, and grading. For NETs ≤1 cm without invasive features, endoscopic resection (ESD/EMR) is recommended. However, for larger lesions or those with invasive potential, surgical resection should be considered [17]. Consequently, hospital admission for lesion excision was recommended, and the patient consented to the procedure.

Histopathological analysis demonstrated a proliferation of interwoven spindle cells, which lacked the characteristic nuclear features of NETs, such as "salt-and-pepper" chromatin. This finding suggested a possible differential diagnosis, including leiomyoma, gastrointestinal stromal tumor (GIST), schwannoma, or neurofibromatosis, all of which are spindle cell-derived neoplasms or lesions [20–23]. Therefore, further immunohistochemical (IHC) and special staining analyses were performed for precise differentiation.

Immunohistochemical analysis demonstrated that the spindle cell component was negative for S100, Desmin, Caldesmon, CD117, CD34, and DOG-1, with a proliferation index (Ki-67) of < 1%.Special staining results showed positive staining for elastin fibers and Masson's trichrome, while AB-PAS and PAS were negative.

S100 protein is a widely used marker for diagnosing neurological and certain soft tissue tumors [24]. Desmin is a muscle-specific intermediate filament protein, and Caldesmon is an actin-binding protein, both of which are typically expressed in leiomyomas and rhabdomyogenic tumors [25, 26]. CD117 (c-KIT) and DOG-1 are specific markers for GIST [27]. CD34 serves as a marker for stem cells and certain types of vascular endothelial cells [28].

The negative expression of these markers indicates that this tumor does not belong to common neurogenic tumors, myogenic tumors, or GIST.

In the analysis of special staining, elastin staining was positive, Masson's trichrome staining was positive, while AB-PAS and PAS were both negative. Based on the combined immunohistochemical and special staining results, the lesion does not exhibit features consistent with common gastrointestinal spindle cell tumors, including neurogenic, myogenic, and GISTs. The positive elastin staining and the abundant collagen fibers within the tissue are highly characteristic of EF [29]. Therefore, the lesion is diagnosed as EF.

Although rare in the gastrointestinal tract, EF should be considered in the differential diagnosis of abnormal submucosal lesions. Previous reports on gastrointestinal EF indicate a favorable prognosis, with no documented cases of recurrence [30]. Nevertheless, regular follow-up and close surveillance are recommended to monitor for any potential complications and ensure optimal long-term outcomes.

4 Conclusion

This study presents a rare case of rectal EF and analyzes its clinical, pathological, and imaging characteristics. Although EF is a benign lesion, its morphological overlap with GISTs, neurogenic tumors, and smooth muscle tumors can lead to diagnostic challenges. Histopathology and special staining play a crucial role in EF diagnosis. In this case, positive elastin fiber staining and Masson's trichrome staining confirmed the presence of abundant elastin and collagen, effectively differentiating EF from other mimicking lesions. Given the exceptional rarity of EF in the gastrointestinal tract, clinicians should consider it in the differential diagnosis of submucosal rectal lesions. Further research should focus on elucidating the molecular mechanisms of EF and developing standardized diagnostic criteria to enhance diagnostic accuracy and clinical management.

Author contributions Chen-Xin Xiang and Yu Long were responsible for image acquisition and manuscript drafting; Fei Huang and Yun Ke were responsible for data interpretation and manuscript revision; Yi-Ning Xiang provided pathological analysis; Yi-Ran Yao was responsible for clinical evaluation and manuscript review; Yun-Huan Zhen, PhD, supervised the research and finalized the manuscript. All authors contributed to the manuscript and approved the final version for submission.

Funding This research was supported by the National Natural Science Foundation Incubation Project of Guizhou Medical University Affiliated Hospital (No. gyfynsfc [2022]-5) and the Key Projects of the Basic Research Program of the Science and Technology Department of Guizhou Province (No. Qian Ke He Ji Chu-ZK [2023] Key 043).

Declarations

Ethics approval and consent to participate The research was ethically approved by Guizhou Medical University. Prior to participation, participants were duly informed of her rights and responsibilities and provided explicit written consent. The study was conducted in agreement with the guidelines governing research involving human participants, as outlined by the Ethics Committee of Guizhou Medical University.

Consent to publication The authors have obtained written consent to publish the images/details of the individual(s) included in this manuscript.

Competing interests The authors declare no competing interests.

References

1. Jarvi O Fau - Saxen E, Saxen E. Elastofibroma dorse.
2. Çevik HB, Girgin AB, Gökçe A, Kurtuluş B. Isolated Elastofibroma of the Thigh: A Case Report. (2168–8184 (Print)).
3. Haneke E. Subungual Elastofibroma. (2296–9195 (Print)).
4. Yenigün BM, Yildiz O Fau - Yüksel C, Yüksel C Fau - Enön S, Enön S Fau - Kayi Cangir A, Kayi Cangir A Fau - Kutlay H, Kutlay H Fau - Akal M, et al. [Elastofibroma dorsi: report of 11 cases analysis and review of the literature]. (2980–3187 (Electronic)).
5. Goldblum JR, Beals T Fau - Weiss SW, Weiss SW. Elastofibromatous change of the rectum. A lesion mimicking amyloidosis. (0147–5185 (Print)).
6. Oliva MS, Smimmo A, Vitiello R, Meschini C, Muratori F, Maccauro G, et al. Elastofibroma dorsi: what's new? Orthopedic Rev. 2020;12(Suppl 1):8708.
7. Nakamura Y, Okamoto K, Tanimura A, Kato M, Morimatsu M. Elastase digestion and biochemical analysis of the elastin from an elastofibroma. Cancer. 1986;58(5):1070–5.
8. Ishida M, Iwai M, Kagotani A, Iwamoto N, Okabe H. Elastofibromatous change of the intestine: report of four lesions from three patients with review of the literature. Int J Clin Exp Pathol. 2014;7(5):2291–7.
9. Märkl B, Kerwel TG, Langer E, Müller W, Probst A, Spatz H, et al. Elastosis of the colon and the ileum as polyp causing lesions: a study of six cases and review of the literature. Pathol Res Pract. 2008;204(6):395–9.
10. Hernández JL, Rodríguez-Parets JO, Valero JM, Muñoz MA, Benito MR, Hernandez JM, et al. High-resolution genome-wide analysis of chromosomal alterations in elastofibroma. Virchows Archiv: An Int J Pathol. 2010;456(6):681–7.
11. Imanishi A, Imanishi H, Yoshida Y, Okabayashi A, Tateishi C, Ikushima H, et al. Upregulation of TGF-β1 and basic FGF in elastofibroma: an immunohistochemical analysis. Med Mol Morphol. 2016;49(2):83–8.
12. Rodrigues Rodrigues R, de Maia Queiroz MS, da Silveira ÉJD, Freitas RA, de Souza LB, de Andrade Santos PP. Identification of elastofibroma and elastofibroma-like lesions in cases diagnosed as oral fibromas. Biotech Histochem. 2021;96(8):608–15.
13. Bosisio MB, Schmid C, Schiaffino E. Elastofibroma. Case report. Tumori. 1981;67(5):501–5.
14. Neagoe O, Faur CI, Ionică M, Baderca F, Folescu R, Gurgus D, et al. Elastofibroma dorsi, a rare condition, with challenging diagnosis. Case report and literature review. Medicina. 2021. https://doi.org/10.3390/medicina57040370.
15. Tsutsumi A, Kawabata K, Taguchi K, Doi K. Elastofibroma of the greater omentum. Acta Pathol Jpn. 1985;35(1):233–41.
16. Sorbye H, Grande E, Pavel M, Tesselaar M, Fazio N, Reed NS, et al. European neuroendocrine tumor society (ENETS) 2023 guidance paper for digestive neuroendocrine carcinoma. J Neuroendocrinol. 2023;35(3):e13249.
17. Ito T, Masui T, Komoto I, Doi R, Osamura RY, Sakurai A, et al. JNETS clinical practice guidelines for gastroenteropancreatic neuroendocrine neoplasms: diagnosis, treatment, and follow-up: a synopsis. J Gastroenterol. 2021;56(11):1033–44.
18. Ichikawa Y, Kobayashi N, Takano S, Kato I, Endo K, Inoue T. Neuroendocrine tumor theranostics. Cancer Sci. 2022;113(6):1930–8.
19. Marc B, Monino L, Rimbas M. EUS-guided intra-tumoral therapies. Best Pract Res Clin Gastroenterol. 2022;60–61:101817.
20. Kaur G, Gondal R. Oral leiomyoma. J Oral Maxillofac Pathol: JOMFP. 2011;15(3):361–2.
21. Miettinen M, Sarlomo-Rikala M, Lasota J. Gastrointestinal stromal tumours. Ann Chir Gynaecol. 1998;87(4):278–81.
22. Lin CS, Hsu HS, Tsai CH, Li WY, Huang MH. Gastric schwannoma. J Chin Med Assoc: JCMA. 2004;67(11):583–6.
23. Moghadam EA, Navabi Shirazi MA, Mirzaaghayan MR, Nikoufar M, Ghamari A. [Not Available]. Archives de pediatrie : organe officiel de la Societe francaise de pediatrie. 2018;25(1):39-41
24. Allgöwer C, Kretz AL, von Karstedt S, Wittau M, Henne-Bruns D, Lemke J. Friend or foe: S100 proteins in cancer. Cancers. 2020. https://doi.org/10.3390/cancers12082037.
25. Hirota S. Differential diagnosis of gastrointestinal stromal tumor by histopathology and immunohistochemistry. Transl Gastroenterol Hepatol. 2018;3:27.
26. Bayçelebi D, Kefeli M, Yıldız L, Karagöz F. Comprehensive immunohistochemical analysis based on the origin of leiomyosarcoma. Polish J Pathol. 2022;73(3):233–43.
27. Parab TM, DeRogatis MJ, Boaz AM, Grasso SA, Issack PS, Duarte DA, et al. Gastrointestinal stromal tumors: a comprehensive review. J Gastrointest Oncol. 2019;10(1):144–54.
28. Hassanpour M, Salybekov AA, Kobayashi S, Asahara T. CD34 positive cells as endothelial progenitor cells in biology and medicine. Front Cell Dev Biol. 2023;11:1128134.
29. Kakudo N, Morimoto N, Ogawa T, Hihara M, Koseki R, Kusumoto K. Elastofibroma dorsi: a case report with an immunohistochemical and ultrastructural studies. Med Mol Morphol. 2016;49(1):42–7.
30. Hobbs CM, Burch DM, Sobin LH. Elastosis and elastofibromatous change in the gastrointestinal tract: a clinicopathologic study of 13 cases and a review of the literature. Am J Clin Pathol. 2004;122(2):232–7.