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Case Report

CASE REPORT

An oncological curiosity of a male patient with a huge leiomyoma of the terminal ileum

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Abstract

Leiomyoma is a smooth muscle tumour that can arise in any part of the body especially the uterus. Even though it is traditionally linked with hormonal influence, it can also develop in extrauterine organs with a slight female predominance. It is indistinguishable with gastrointestinal stromal tumour (GIST) histologically. We report a case of a 30-year-old gentleman who presented with a huge painful mass in the right iliac fossa. Computed tomography revealed a 10 × 10 cm homogeneous mass arising from the terminal ileum; he subsequently underwent an open right hemicolectomy. Histology showed a well-circumscribed lesion composed of interlacing bundles of smooth muscle fibres of the submucosa with positive smooth muscle actin and H-Caldesmon stains but negative for DOG-1 and CD117 (c-kit) stains which were consistent with leiomyoma. Despite its rarity, this hormone-related tumour needs to be considered regardless of gender. Immunohistochemistry is paramount as it is histologically identical to GIST.

INTRODUCTION

Leiomyoma is a benign tumour that can arise in any soft tissue sites most commonly in the uterus [1]. The extrauterine cause is rare with gastrointestinal leiomyoma most often found in the oesophagus, stomach and colon [2]. Leiomyoma of the duodenum is the rarest among all gastrointestinal leiomyomas which is then followed by the ileum. This tumour is classified as mesenchymal neoplasms of gastrointestinal tract which are divided into two groups: gastrointestinal stromal tumours (GISTs) and other smooth muscle tumours. The latter group consists of a spectrum of diseases such as lipomas, liposarcomas, leiomyomas, leiomyosarcoma, desmoid tumors, schwannomas and peripheral nerve sheath tumors [3]. There has been a shift in nomenclature to separate GISTs from the smooth muscle tumours due to the discovery of expression of an KIT protein in GISTs by immunohistochemistry in the late 1990s [4]. Smooth muscle tumours are most commonly encountered in middle-aged patients and have a slight female predominance (ratio 1.3:1) [3]. We report a 30-year-old male who presented with a painful right iliac fossa mass, diagnosed as an intestinal leiomyoma following surgical resection.

CASE REPORT

A 30-year-old gentleman with a history of laparoscopic appendicectomy 7 years ago presented to a surgical clinic with a

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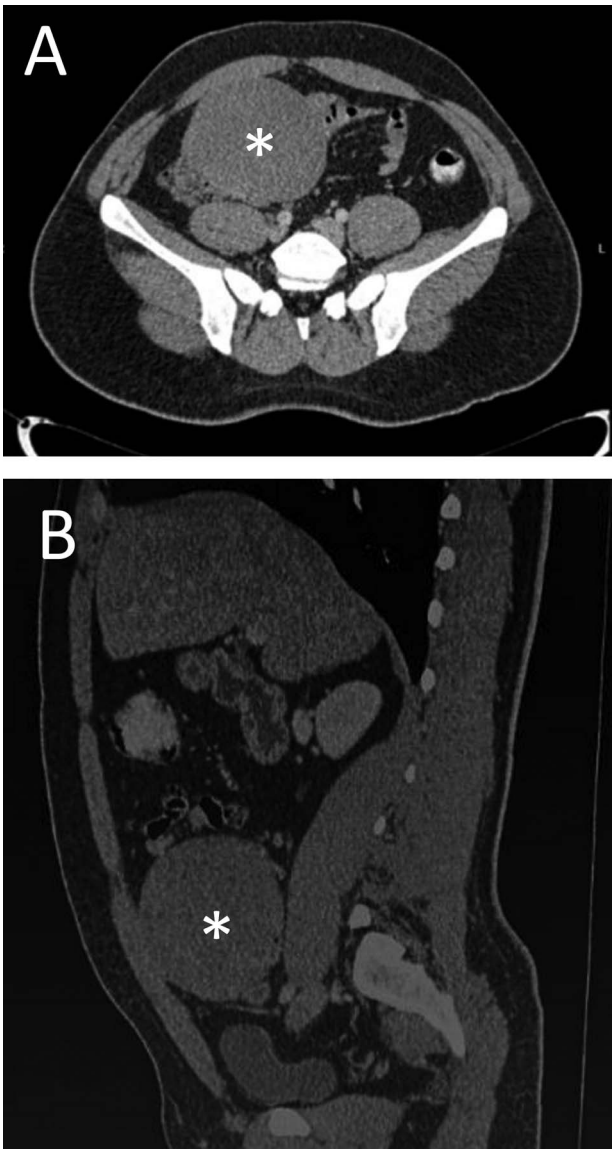

Figure 1: (A) Axial CT image shows homogenous mass (*) extending anteriorly abuting the posterior margin of the right rectus abdominis muscle. (B) Sagittal CT image of the homogeneous soft tissue mass (*) in the right iliac fossa with specks of fat within it.

painful right iliac fossa mass for 3 weeks. He was otherwise well and denied any bowel or constitutional symptoms. On examination, there was a 10 × 10 cm tender, firm, mobile, globular mass in the right iliac fossa with no overlying skin changes. No lymph node enlargement was appreciated.

Blood investigations were unremarkable. Abdominopelvic computed tomography (CT) showed a homogeneous oval-shaped soft tissue mass (HU 50-80) arising from the terminal ileum (Fig. 1). The mass was 9.8 × 9.0 × 10.4 cm in size with specks of fat within and without calcification. It extended anteriorly and abutted the posterior margin of the right rectus abdominis muscle. The radiological features were suggestive of ileal GIST.

There was no evidence of intraluminal lesion, especially a central umbilication upon colonoscopy. However, random biopsies of the terminal ileal wall were obtained and the histopathological report was normal. Terminal ileal GIST was suspected and the patient was prepared for limited right hemicolectomy via a laparoscopic approach. Conversion to open surgery was done due to dense adhesions between the tumour and the anterior and lateral abdominal wall. Intra-operatively, a solid tumour of approximately 8 × 8 cm in size arising from the terminal ileum was resected together as a limited right hemicolectomy with ileocolic anastomosis (Fig. 2). Post-operatively, he developed an anastomotic leak 5 days after his surgery and subsequently underwent a re-exploration surgery with the formation of a double-barrel stoma. He was discharged and was well at 1 month follow-up.

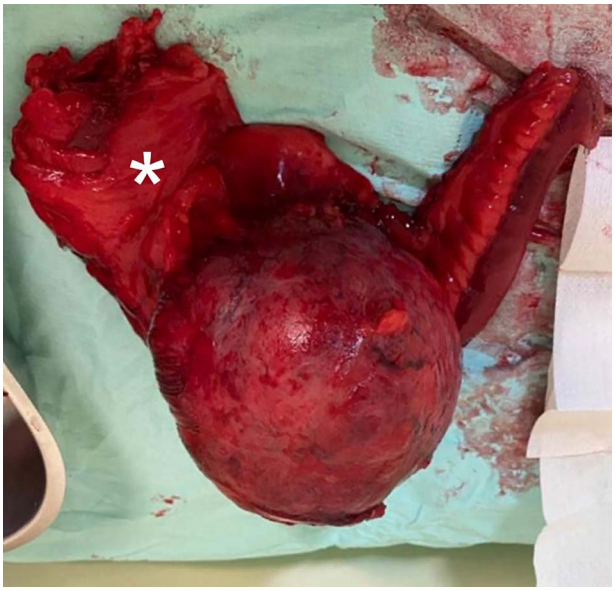

Figure 2: Hemicolectomy specimen with 8 × 8 cm tumour at the terminal ileum. Caecum (*) was cut open.

The tumour was 105 × 100 × 70 mm in size, in which microscopy showed submucosal leiomyoma displaying a well-circumscribed lesion composed of interlacing bundles of smooth muscle fibre extending to the serosa layer. There was no increment in mitotic activity or cytological atypia. Immunohistochemistry (IHC) was negative for CK AE1, CK AE3, DOG-1 and CD117. It was positive for smooth muscle actin (SMA) and H-Caldesmon (Fig. 3). The final histopathological diagnosis of the tumour was reported as leiomyoma of the terminal ileum.

DISCUSSION

This case demonstrates an unusual cause of huge abdominal mass and the difficulty to distinguish GIST from leiomyoma without immunohistochemical analysis. Clinical presentation depends on size and site of the lesion. Around 44–50% of leiomyomas are symptomatic and can present as intestinal obstruction, gastrointestinal haemorrhage or intussusception [5]. Smaller lesions are usually found incidentally during surgery, radiological studies or endoscopy. The clinical presentation is similar to GISTs. Although 50% of these tumours are usually less than 5 cm, around 5% can grow to more than 15 cm [5, 6].

Diagnostic workup includes an endoscopic assessment of the gastrointestinal tract. However, as 63% of these tumours are extra-luminal and difficult to assess using conventional endoscopy, it may yield negative results as demonstrated in

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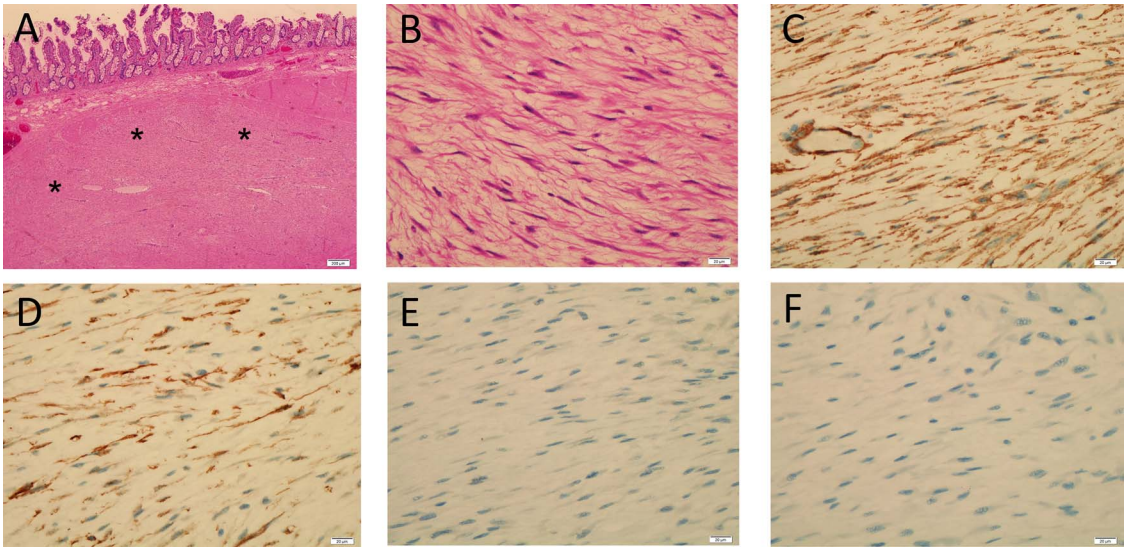

Figure 3: (A) Histopathological assessment showing a submucosal leiomyoma (*) displaying a well-circumscribed lesion composed of interlacing bundles of smooth muscle fibres adjacent to a small bowel (×10 magnification). (B) Higher magnification showing interlacing bundles of smooth muscle fibres (×40 magnification). (C) IHC showed a positivity against H caldesmon (×40 magnification). (D) Positive IHC of SMA (×40 magnification). (E) Negative IHC of CD117 (c-kit) (×40 magnification). (F) Negative IHC of DOG-1 (×40 magnification).

our case [5]. CT has been successful in detecting 89.5% of leiomyomas, and the tumours are usually homogeneous, low-attenuation masses with an endoluminal growth pattern [5]. Magnetic resonance imaging enteroclysis and enterography are gaining acceptance as the initial imaging modality to investigate small-bowel diseases as the soft-tissue contrast is superior and the morphological information and functional information can also be obtained. It is difficult to differentiate GISTs and leiomyoma radiologically as their characteristics overlap as well as the anatomical origin. Other differential diagnoses include leiomyosarcoma, malignant fibrohistiocytoma and schwannomas [7].

Treatment should be guided by the pathological diagnosis; however, this is difficult to be obtained pre-operatively in practice. If the tumour histology is unknown, an intraoperative pathological diagnosis is recommended via frozen section [8]. Nevertheless, any suspicious colonic or terminal ileal pathology warrants a surgical resection following oncological outcomes.

Grossly, both leiomyoma and GISTs are typically well circumscribed tumours. Microscopically, both exhibit spindle cell morphology although a minority of GISTs have epithelioid morphology [4]. Although in our case there were no cytological atypia and no increase in mitosis, smooth muscle tumours of the small bowel larger than 10 cm have been found to have 33% risk of progressive disease warranting a follow-up of at least 2 years [3]. It is clinically important to differentiate leiomyoma from GISTs as the latter is 40–50% malignant and these patients may benefit from tyrosine kinase inhibitors. KIT (CD117) and GIST-1 (DOG-1) are established GISTs markers with 95% sensitivity. The discovery of KIT as the main proto-oncogene in GISTs necessitated the use of KIT inhibitors such as imatinib in the setting of metastatic or unresectable GISTs, or adjuvant therapy for high-risk disease, thus significantly improving recurrent free survival and overall survival [9]. In this case, the immunohistochemical tests were negative for both and were positive for the smooth muscle markers (actin and H-Caldesmon), which strongly suggest a leiomyoma [6, 10].

Terminal ileum leiomyoma is a rare cause of acute abdomen. CT is a helpful tool to diagnose this entity. However, histopathological examination with adjunct immunohistochemistry is of paramount importance to distinguish between GIST or leiomyoma and benign or potential malignant diseases to guide further treatment.

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CONFLICTS OF INTEREST STATEMENT

The authors have no conflict of interests to declare.

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ETHICAL APPROVAL

No ethical approval was required for this report.

CONSENT

Written informed consent was obtained from the patient. A copy of the written consent is available for review by the editor of this journal.

GUARANTOR

Firdaus Hayati

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