A case of intestinal obstruction in a middle aged woman – a pathological surprise

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Case Report

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Abstract

Background:

Fibromatosis is a tumour of fibroblasts and myofibroblasts with a tendency of local invasion and recurrence but no malignant potential and is classified as superficial or deep. Intra-abdominal fibrosis, a type of deep fibromatosis, is a rare cause of intestinal obstruction. We report intestinal obstruction in a middle aged woman caused by jejunal/mesenteric fibromatosis.

Case presentation:

An early 40s woman presented with intestinal obstruction and a month-long history of abdominal pain. She required emergency surgical intervention which uncovered an obstructive mass involving the jejunum and adjacent mesentery. Histology of the resected tumor with wide surgical margins revealed fibromatosis with positive nuclear â-catenin immunohistochemistry.

Conclusion:

Fibromatosis is a rare cause of intestinal obstruction. Awareness of the condition and recognition of the myriad histological features is important for accurate diagnosis and distinction from its mimickers. Positive nuclear staining of tumour cells for â-catenin is an important clue to diagnosis. Distinction from other causes of intestinal obstruction is paramount for implications on different modalities of management and prognosis.

Background

Intestinal obstruction (IO) is a surgical emergency requiring urgent surgical intervention. IO is commonly caused by fibrous adhesions, hernias, intussusception, intestinal tumours or intraluminal foreign bodies such as gall stones. Fibromatosis is a rare cause of IO and usually not diagnosed until pathological confirmation on excised material from surgery[1–6]. Only sporadic cases were reported. Diagnosis of fibromatosis requires full awareness of the condition even in unusual clinical scenarios, recognition of the myriad morphological patterns and the appropriate application of immunohistochemical and genetic tools. Our patient presented with IO caused by fibromatosis.

Case Presentation

An early 40s woman presented in April 2022 with abdominal pain and lost 15 pounds in the past 4 months. Abdominal ultrasonography in March was normal. Esophagogastroscopy showed mild gastritis. The abdominal pain worsened and in April, colonoscopy revealed long colonic loops and hyperplastic rectal polyps. Computerized tomography showed dilated jejunum to 3.7 cm diameter for a length of 1.8 cm in the right lower abdomen, with oedema of the adjacent mesenteric fat. The clinical diagnosis was intestinal obstruction with possibilities of jejunal tumour, lymphoma, Crohn’s disease or tuberculosis.

Laparotomy was performed in early May, uncovering a mesenteric fibrotic mass infiltrating the jejunum, with kinking and proximal dilation. The fibrotic mass and jejunum were resected en bloc. Post operative recovery was uneventful. The pre-operative haematological blood counts, blood renal and liver function biochemistry, lipid profile, glucose, thyroxine and thyroid stimulating hormone were normal. There was no history of previous abdominal surgery. She was pregnant in 2008 followed by normal spontaneous delivery in September of the same year. There was no history of hormonal therapy, colonic adenomas, familial polyposis (FAP) or Gardner syndrome (GS).

Pathology:

The 28 cm long resected jejunum showed a 7 cm mesenteric tumor with involvement of adjacent jejunum and wide resection margins. The tumour was firm and appeared “fibrotic”, causing severe reduction of the jejunal calibre (Fig. 1). Histologically, the tumour featured proliferation of loosely dispersed bland spindle fibroblasts and myofibroblasts lacking cytological atypia. The stroma was loose, myxoid or fibrotic containing extravasated red cells and inflammatory cells, portraying a nodular fasciitis (NF)-like pattern which accounts for 60% of the tumour (Fig. 2a). The other 40% exhibited frequent staghorn vessels in hyaline or paucicellular fibrous stroma, representing staghorn vessel pattern (Fig. 2b). The tumour extended to the serosa and involved the jejunal muscularis propria and submucosa, with negative jejunal resection margins. Immunohistochemically, the spindle cells are nuclear β-catenin positive (Fig. 2c), equivocally positive for actin and negative for STAT-6, S-100 protein, ALK and cytokeratin. The overall morphological and immunohistochemical findings were those of intestinal/mesenteric fibromatosis causing intestinal obstruction.

Discussion And Conclusions

Fibromatosis is a rare fibroblastic or myofibroblastic tumor of uncertain aetiology though genetic, hormonal factors, irradiation and trauma have been ascribed to its predisposition[7–8]. The incidence is 4–6/1,000,000 population per year, accounting for 0.03% of all tumours and 3% of soft tissue tumours[8]. There is increased association of fibromatosis with FAP and GS, associated with abnormalities in the *APC* gene. *CTNNB1* mutations are observed in 90% of sporadic cases. Hormonal contribution is suggested by increased incidence of fibromatosis during and after pregnancy. Fibromatosis developing after previous surgery, irradiation or trauma is also observed[7–8]. Our patient is a sporadic case with no history of FAP or GS. Her last pregnancy was in 2008, fourteen years earlier than the episode of intestinal obstruction and is unlikely a contributing factor in the development of fibromatosis. There was also no history of previous abdominal surgery, trauma or irradiation. There were apparently no predisposing causes or antecedent events in our patient.

Fibromatosis is classified as superficial or deep[7–9]. It is a recurrent disease without metastatic potential. Deep fibromatosis is more prone to recurrence and regarded as a borderline tumour. It includes extra-abdominal, abdominal and intra-abdominal types. Among intra-abdominal types, mesentery fibromatosis is more common than that of the ileocecal ligament, omentum or retroperitoneum[8]. Fibromatosis grows

rapidly initially followed by slow progression to involve nearby structures, causing symptoms related to the anatomical involvement. Intra-abdominal fibromatosis is mostly asymptomatic or may present as abdominal distension or mass. Intestinal obstruction, intestinal perforation with acute peritonitis or symptoms related to compression of adjacent anatomical structures such as ureteric obstruction are less common presentations of fibromatosis[1–2, 8]. Our patient presented as intestinal obstruction, an uncommon presentation.

The histology of fibromatosis is myriad, requiring full recognition for diagnosis. There are seven histological patterns[10]; conventional, hypocellular/hyalinized, staghorn vessel, myxoid, keloidal, nodular fasciitis-like and hypercellular. Most fibromatosis manifest several patterns with the mean number of two. The conventional pattern is the most common and is present in almost all cases. In all patterns, the spindle-shaped fibroblasts and myofibroblasts have bland pale-staining nuclei, inconspicuous nucleoli with or without mild cytological atypia and minimal mitotic activity. The histology of our case was unusual, lacking the conventional pattern and exhibits prominent NF-like and staghorn vessel patterns. This poses NF and solitary fibrous tumour (SFT) the two most important differential diagnoses. Fibromatosis is distinguishable by nuclear β-catenin expression and genetic changes related to β-catenin/Wnt signalling pathway[1–8,11–13]. Though β-catenin immunopositivity is not entirely specific for fibromatosis[7], its morphologic differences from the mimickers and the different immunophenotypic and genotypic features of these various conditions enable their distinction. NF, a major differential diagnosis in our patient, is negative for nuclear β -catenin staining immunohistochemically. Further, NF lacks staghorn vessels present in our case. NF is also characterized by *USP* gene rearrangement[14]. SFT may be more problematic as it morphologically may also manifest keloidal fibres, staghorn vessels and that 40% may be nuclear β-catenin positive. It is however distinctive in STAT-6 positive immunophenotype[15]. Other differential diagnoses of fibromatosis encompass various malignant/borderline spindle cell tumours (spindle cell carcinoma and melanoma, leiomyosarcoma, malignant peripheral nerve sheath tumour, synovial sarcoma, angiosarcoma, fibrosarcoma and gastrointestinal stromal tumour). These tumours could be easily distinguished by their characteristic morphology, immunophenotypes and genotypes. Differential diagnoses[7] from reactive fibroblastic/myofibroblastic proliferations, myxoma, keloid, sclerosing mesenteritis/idiopathic retroperitoneal fibrosis (Ig4-related disease) and inflammatory myofibroblastic tumour (IMT) can usually be made by differences in morphology, immunophenotypes and nuclear β-catenin positivity in fibromatosis. IMT may morphological range from bland to atypical spindle cells with frequent ALK immunopositivity and *ALK* gene rearrangement, allowing its distinction from fibromatosis[7].

The treatment of asymptomatic fibromatosis could be surveillance. In emergencies such as intestinal obstruction, treatment is surgical intervention. Recurrence occurs in about 50% of deep fibromatosis, and is related to age (younger age being a risk factor), tumour location and resection margin status[8]. Wide negative resection margins are associated with a lower recurrence rate, compared with positive margins (10% vs 80%)[7–8]. Negative resection margins in our patient portend an expected lower recurrence rate.

Other treatment modalities are radiotherapy or systemic therapy (hormonal, cytotoxic drugs, targeted therapy)[8].

To conclude, we reported a rare case of intestinal obstruction caused by intra-abdominal intestinal/mesenteric fibromatosis which required urgent surgery. Wide surgical excision with negative resection margins in our case should confer a lower recurrence rate. The tissue diagnosis was challenging due to the prominent NF-like and staghorn vessel patterns, but was confirmed by bland-looking fibroblastic proliferation with infiltrative border, β-catenin nuclear staining and negative immunostaining for markers of its mimickers. Distinction from various other spindle cell tumours and malignancies is important as the post-surgical management and prognosis are materially different from fibromatosis.

Declarations

Ethics approval and consent to participate: not applicable. This is a case report and the patient's consent for publication has been obtained.

Consent for publication: Consent obtained and approved by the patient.

Data and material availability statement: Data available from corresponding author on reasonable request.

Competing interests: None

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Authors' contribution: CSN and WC researched on the topic and reviewed the paper. CSN conceived and drafted the paper. DYSK contributed to the clinical summary and reviewed the paper. All authors have read and approved the final manuscript.

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Figures

Figure 1

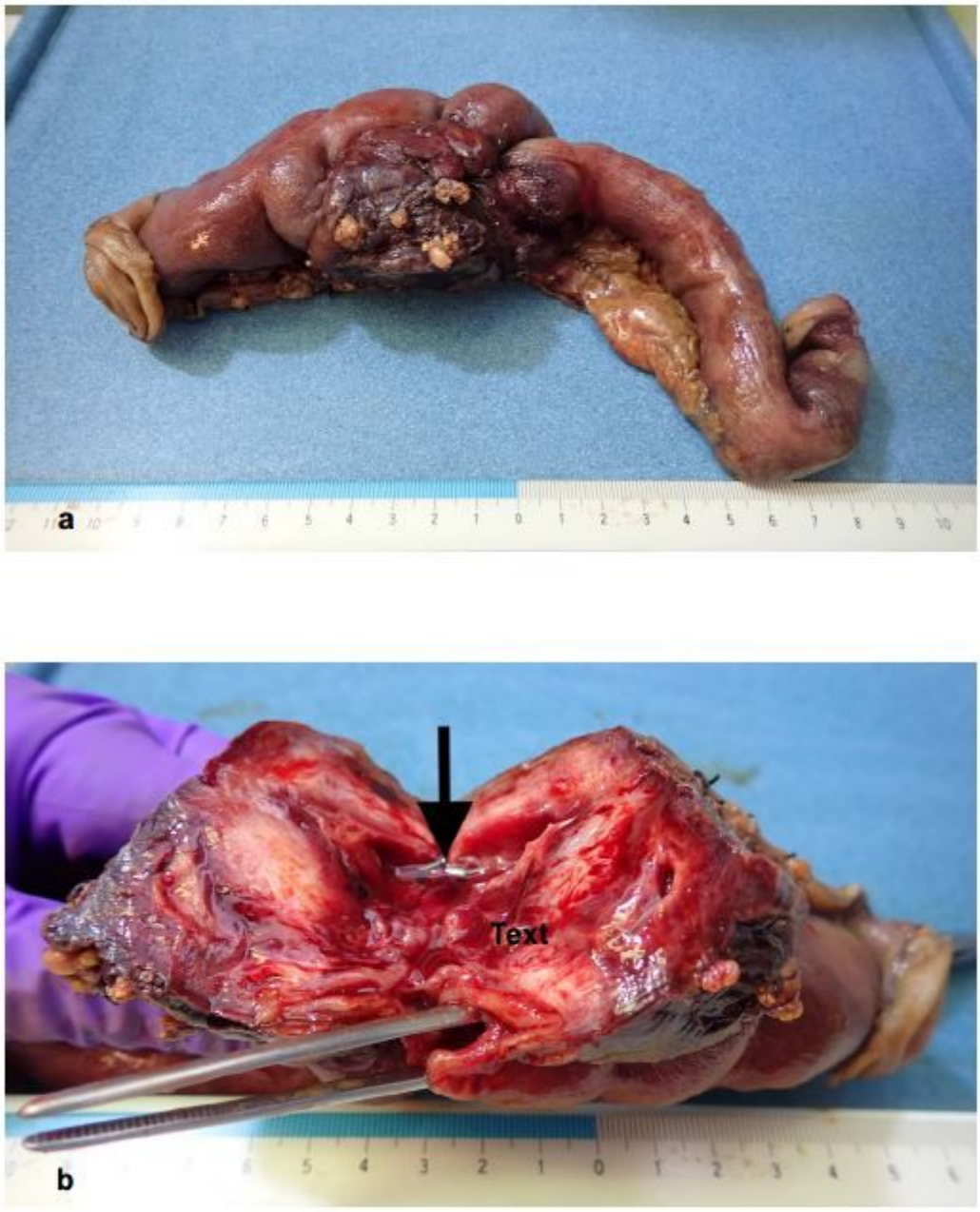

Figure 1

(a) Resected jejunum with a tumour at almost the middle involving the intestine and the adjacent mesentery. (b) Cut surface of tumour uncovered firm “fibrotic” tissue. The tumour significantly reduced the jejunal calbre, as indicated by a silvery probe (marked with arrow).

Figure 2

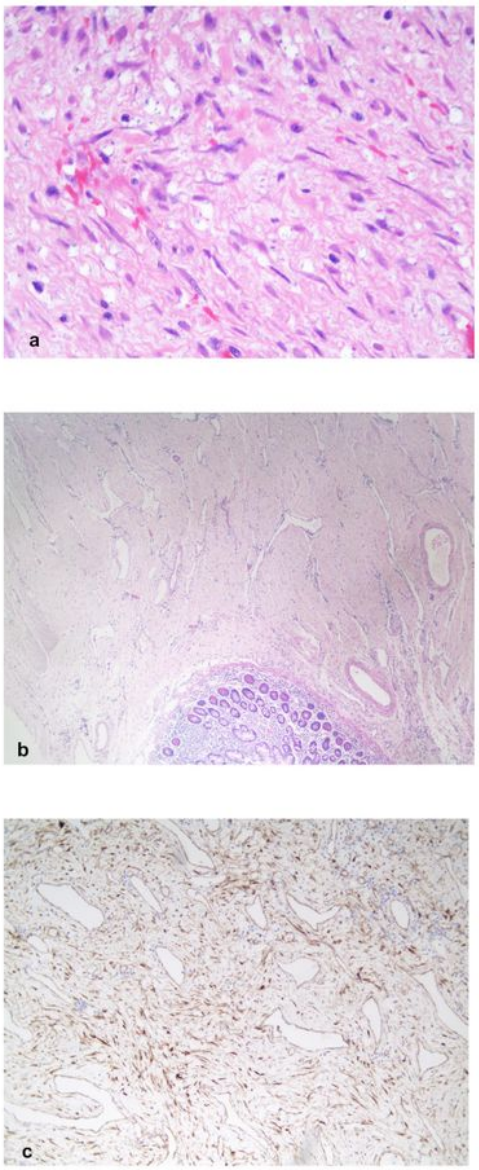

Figure 2

(a) NF-like pattern. Bland spindle fibroblasts and myofibroblasts in loose stroma with extravasated red cells portraying “tissue culture-like” pattern (H&E x 400). (b) Staghorn vessel pattern. Many thin walled staghorn vessels in paucicellular stroma. There is invasion of the jejunal muscularis propria and submucosa (H&E x 100). (c) Positive nuclear â-catenin immunostaining of tumour cells (Immunostaining x 200).