Diagnosis Challenge and Treatment Options in Intra-abdominal Fibromatosis: A Case Report

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Abstract

Desmoid-type Fibromatosis is a relatively rare disease with diagnosis challenge and differential treatment options. In this study, we summarized and analyzed the diagnosis and treatment of a case of intra-abdominal desmoid- type fibromatosis occurring in the transverse mesocolon, aiming to improve the clinical understanding of the disease and reduce the occurrence of misdiagnosis and missed diagnosis. Based on our diagnosis and treatment experience of the case, and consulting the relevant literature in recent years, we conclude and arise the clinical awareness on this disease.

Keywords

Transverse Mesocolon; Intra-abdominal; Desmoid-type Fibromatosis; Surgery.

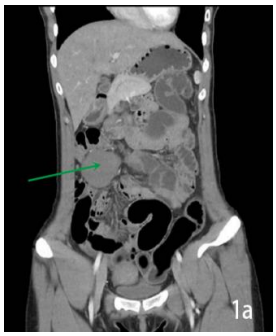
1. Introduction

Desmoid- type Fibromatosis (DF) is a relatively rare mesenchymal tumor occurring mainly due to the proliferation of fibroblasts and myofibroblasts in clinic. Although DF exhibits the appearance and morphology of a benign tumor, it is classified as a moderately malignant tumor due to its local invasion and a high recurrence rate after surgical resection [1]. According to the location of the tumor, it can be divided into extra-abdominal (60%), abdominal wall type (25%) and intra-abdominal type (8%–15%) [2]. The intra-abdominal type is the most rare, which mainly occurs in the mesentery, and a few in the transverse mesocolon [3]. Aggressive fibromatosis of the abdominal wall had been reported, noticed DF it is easily misdiagnosis even on the surface of the abdominal wall. We report a case on the most difficult differentiate diagnosis anatomic location of intra-abdominal DF occurring in a young woman, highlighting the process and methods of differential diagnosis and treatment of the intra-abdominal transverse mesocolon DF.

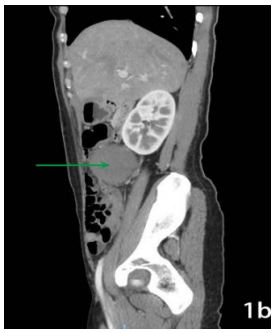
2. Case Introduction

A 25-year-old woman was admitted to our hospital on 22 January 2021 because a progressive enlargement of a right middle abdominal mass was found for one month. The patient occasionally had abdominal bloating, no fever, nausea, vomiting and other symptoms, and his stool and urine were normal. Physical examination: A mass about 5.5cm in diameter was palpable in the right middle abdomen. The mass was medium in nature, with a clear boundary, poor activity, and no tenderness. Tumor markers, such as CEA, CA199, CA125, and AFP, were within normal limits. Total abdominal contrast-enhanced CT revealed a solid lesion in the right middle abdomen, measuring about 6.5cm ×4.3cm ×5.5cm, with a clear boundary and uniform density. After enhancement, mild enhancement was performed, and the lesion was diagnosed as mesenchymal tumor (Figure 1), which was initially considered as gastrointestinal stromal tumors (GIST). She got a preoperative colonoscopy and no intestinal polyps were found. A right hemicolectomy and partial duodenal resection were performed on 27 January 2021. During the operation, a mass with size of about 9cm ×6cm×6cm located in the transverse mesocolon, with a smooth surface and infiltrative growth, medium quality, poor activity, and unclear boundary. It surrounded the horizontal part of duodenum upward, and had severe adhesion to the transverse colon, transverse mesocolon and the ileum (Figure 2). Postoperative

pathological diagnosis was desmoid-type fibromatosis of the mesentery (Figure 3). Immunohistochemistry showed β-catenin(nuclear,+), but CD117/DOG-1/CD34/desmin/ALK/SOX-10 were negative. The patient recovered well after surgery and was discharged after his condition was stable. At a follow-up about six months after surgery, total abdominal contrast-enhanced CT showed no significant evidence of tumor recurrence and intestinal function recovered essentially same as pre-surgery.



(a)



(b)

Figure 1. CT showed a parenchyma mass in the right midabdomen, clearly demarcated from the surrounding tissue.

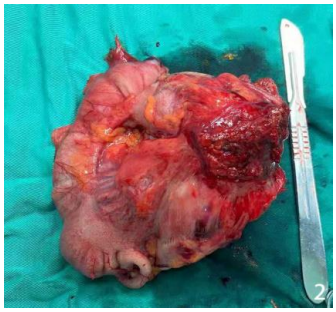


Figure 2. The gross specimen of the tumor removed included part of the ascending colon, appendix, duodenal intestine and mesentery.

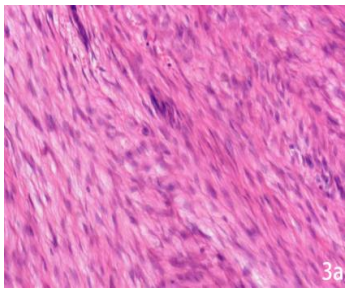


Figure 3(a). Postoperative pathological images showed a large number of myofibroblasts. (HE×200)

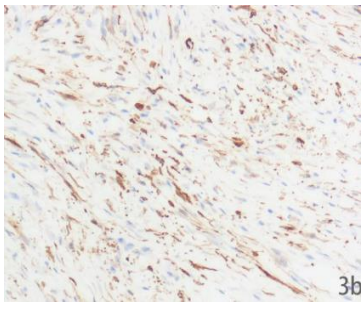


Figure 3(b). Immunohistochemistry of a DF with characteristic β-catenin staining.

3. Discussion

Desmoid-type fibromatosis, also known as aggressive fibromatosis, is a myofibroblastic tumor that is prone to developing in deep soft tissues. DF is rare in clinical, with an incidence of 2–4/1 million, accounting for about 3% of soft tissue tumors, and is common in females and young adults, especially in those aged 10–40 years old. The ratio of females to males is 3: 1 [3]. The cause of DF has not been clarified, but DF has been previously reported being associated with abdominal trauma, such as surgery and pregnancy, estrogen disorders, and familial adenomatous polyposis (FAP) [3–4]. The myofibroblasts forming DF are similar to the proliferative phase of wound healing in histology, so trauma such as surgery and pregnancy may be important factors leading to DF. In patients with FAP, the incidence of DF is 1000 times of ordinary people, 5% to 30% of DF patients also suffer from FAP, and FAP-associated DF represents about 2% of DF cases, which may be related to a mutation in APC gene [4].

The clinical symptoms of intra-abdominal DF are nonspecific. The main manifestations of DF are painless abdominal mass and mild abdominal bloating, and a few of them are intestinal obstruction, intestinal ischemia,necrosis, and gastrointestinal perforation due to compression of the tumor. Intra-abdominal DF is extremely rare in clinic and non-specific in imaging examinations such as CT and MRI. Therefore, preoperative diagnosis is difficult and it is easy to be misdiagnosed as gastrointestinal stromal tumor (GIST). In a retrospective investigation, it was found that the incidence of intra-abdominal DF first misdiagnosed as GIST by preoperative imaging examination was 56.6%, and the proportion of misdiagnosis to GIST by intraoperative frozen section pathology was still as high as 42.9% [5]. In our case, the patient was also misdiagnosed as GIST preoperatively and intraoperatively, so the differentiation between the two diseases had important clinical significance. GIST is the most common mesenchymal tumor in the abdominal cavity, and its histological morphology is similar to that of DF. The differential diagnosis of GIST and DF needs to be confirmed by immunohistochemistry: Most cases of GIST express CD117 and DOG1 at the same time, and cases without simultaneous expression of CD117 and DOG1 can be diagnosed with the help of detecting c-kit or PDGFRA gene [6]; Relatively, Muscle-specific actin(MSA) and smooth muscle actin (SMA) staining can be observed instead of CD117 and DOG1, β-catenin expression is visible in tumor nucleus of 70%-75% DF cases, and cases without the expression of β-catenin can be diagnosed with the help of detecting CTNNB1 gene [8,9]. In addition, DF should be differentiated from myofibroma, fibrosarcoma, Gardner fibroma, etc. In our case, the patient had no obvious pathological signs and no intestinal polyps were found through preoperative colonoscopy. Therefore, she was also empirically misdiagnosed as GIST preoperatively and intraoperatively. However the proliferation of myofibroblasts under microscope, no mitotic pathology was observed and the immunohistochemical staining showed β-catenin (nuclear,+) and CD117/DOG-1were negative, which helped us to diagnose intra-abdominal DF.

At present, the treatment options for intra-abdominal DF are not unified, and the consensus is that the option of "watching and waiting [7]" is a preferred treatment for asymptomatic intra-abdominal DF. Recent studies have shown that growth stagnation or even spontaneous regression may occur in the natural course of asymptomatic DF, and the incidence of spontaneous regression is 20%–30% [8]. Through follow-up observation, more than 50% of patients have a progression-free survival(PFS) of more than five years [9]. For patients with tumor progression or obvious symptoms, extensive surgical resection is still the most important treatment [8,9]. Based on the high recurrence rate and aggressive growth of DF, some scholars believe that the pursuit of RO resection with negative margins under the microscope can effectively reduce the risk of disease recurrence and improve the disease-free survival rate. Bertain E [10] observed 62 postoperative patients with DF and found that the 5-year cumulative recurrence rates of patients with negative incisal margin (R0) and positive endoscopic incisal margin (R1) were 7. 1% and 46. 4% respectively. This result demonstrates the necessity of performing the R0 resection. However, it is not easy to achieve RO resection with negative endoscopic margins due to the non-encapsulated invasive growth of DF. Moreover, surgery is also a

form of trauma, which can lead to the generation of various inflammatory and growth factors. These factors are very important for wound healing, however, they may also stimulate the proliferation of DF-related cells which lead to tumor recurrence [4]. At the same time, extensive invasion of the tumor necessitating extended resection of the affected organ may result in severe organ dysfunction. Therefore, intra-abdominal DF surgery should follow the principle of maximum tumor resection and maximum possible preservation of the morphology and function of the surrounding important structural organs. In addition, radiotherapy is considered as the second-line treatment after surgery, mainly for patients who cannot be surgically removed, who have tumor recurrence, and who have gotten palliative tumor resection (R2) [11]. However, whether postoperative adjuvant chemotherapy should be performed in DF patients with RO and R1 resection is still controversial [12]. Chemotherapy is the third-line treatment for DF after surgery and radiotherapy. The combination of methotrexate and vinblastine was one of the earliest widely used chemotherapy options for DF, with the effective rate ranging from 30% to 50% [4]. In recent years, research has been carried out on the treatment of unresectable DF, including non-steroidal drugs combined with hormones, imatinib and other drugs, but the clinical effect is still not clear [13].

In our case, considering the continuous growth of the tumor might lead to compression symptoms such as intestinal ischemia and intestinal obstruction, surgical treatment was selected. Meanwhile, in combination with enhanced CT judgment, we believed that it was possible to remove the tumor completely and the intestinal tube and adjacent organs did not need to be resected. During the operation, the tumor was seen to arise from the transverse mesocolon and spread aggressively around, involving the hepatic region of the colon, the terminal ileum and part of the horizontal portion of the duodenum. We failed to separate the tumor intact, and eventually we opted for right hemicolectomy with partial duodenal resection. Postoperative pathology showed negative bilateral margins, so we did not perform postoperative adjuvant radiotherapy and chemotherapy to the patient as complementary therapies. The patient's intestinal function basically recovered 3 days after surgery and was discharged after 7 days without stitches. In the follow-up for nearly 6 months after surgery, the patient's intestinal function returned to normal, and the abdominal bloating rarely recurs. She got a reexamination of the abdominal contrast-enhanced CT and found no sign of tumor recurrence. The treatment outcome was satisfactory.

4. Conclusion

Intra-abdominal DF is a rare mesenchymal tumor. Although its tumor morphology and appearance are similar to benign tumors, DF is characterized by aggressive growth and a high recurrence rate, which make the diagnosis and treatment of intra-abdominal DF more complicated. Preoperative colonoscopy and imaging examination are helpful in its diagnosis, but it is still difficult to distinguish DF from abdominal tumors such as GIST, myofibroma, fibrosarcoma, and Gardner fibroma, and the final diagnosis still depends on immunopathological examination. Considering that intra-abdominal DF is rare in clinic and there is no unified treatment plan at present, the author believes that RO surgical resection without affecting the functions of adjacent organs may be an effective treatment option.

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