CASE REPORT

Desmoid-type fibromatosis difficult to distinguish from GIST : A case report

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Abstract : Background : Desmoid-type fibromatosis is a very rare disease that has no characteristic image findings, so it is often difficult to differentiate from gastrointestinal stromal tumor (GIST). A case of desmoid-type fibromatosis that was difficult to differentiate from GIST is reported. The decisive factor in the diagnosis was positive nuclear immunostaining for β-catenin nucleus. Case presentation : A man is his 30s had no significant past medical history, including no abdominal surgery. A medical check-up found a large tumor in the right lateral abdomen. After some examinations, a preoperative diagnosis of GIST was made, and open ileocecal resection was performed. However, the final diagnosis based on the pathological findings was desmoid-type fibromatosis. Conclusions : We should consider desmoid-type fibromatosis when we find a large abdominal mass, but it may be difficult to diagnose based only on imaging findings. Immunohistochemical examination of the specimen may make the diagnosis. J. Med. Invest. 67 : 375-377, August, 2020

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BACKGROUND

Fibromatosis is classified as deep or superficial depending on the site of its occurrence, and the deep type is called desmoid-type fibromatosis. The prevalence of desmoid-type fibromatosis is 2-4/1,000,000 persons. It often occurs in women in their 20s to 40s. It shows locally aggressive growth and a strong tendency for local recurrence. Preoperative diagnosis is often difficult, and differentiation from gastrointestinal stromal tumor (GIST) is particularly challenging (1). A rare case of desmoid-type fibromatosis that was difficult to differentiate from GIST is presented.

CASE PRESENTATION

A man in his 30s was aware of a large abdominal mass. A medical check-up found a large right lower abdominal tumor. He had no past medical history. On physical examination, there was a large mass in the right lower abdomen. All laboratory data were within normal ranges, and tumor markers (AFP, CEA, CA19-9, CA125, sIL-2R) were not elevated. Abdominal ultrasonography showed an iso-low and homogeneously echoic large tumor. There was also blood flow from the margin to the inside of the tumor [Fig. 1 (A)]. Small bowel fluoroscopy showed an exclusion image of the colon and intestine by the tumor [Fig. 1 (B)]. Plain computed tomography (CT) showed a large homogeneous tumor at the ileocecal mesentery, $12 \times 12 \times 9$ cm^3, with weak enhancement. On contrast-enhanced CT, the tumor was gradually enhanced, and the border between the mesentery and tumor was unclear. A branch from the SMA flowing into the tumor was observed [Fig. 2 (A)]. On MRI, the tumor showed low intensity on T1WI, iso-high intensity on T2WI, and low intensity on diffusion-weighted imaging (DWI) [Fig 2 (B)]. Based on these findings, the preoperative diagnosis was GIST, and malignant lymphoma and liposarcoma, etc. were considered in the differential diagnosis.

Ileocecal resection was then performed. The tumor was located at the mesentery of the ileocecum, and it was elastic hard and easy to move. The ascending colon was mobilized from the ileocecal region to the hepatic flexure, and the ileocecal artery was ligated and dissected. The mesentery was resected with a line that dissected the swollen lymph nodes near the ileocecal region. The patient was discharged after a good postoperative course on postoperative day 7.

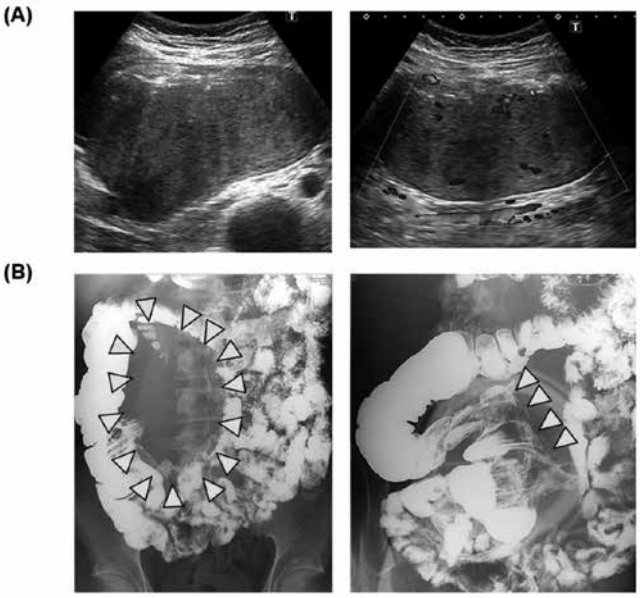

Fig 1. (A) Abdominal ultrasonography shows an iso-low, homogeneously echoic large tumor. Blood flow is confirmed.
(B) Colon and intestine are excluded on small bowel fluoroscopy.

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The inside of the tumor was solid and white, and the tumor measured 12 × 12 × 8 cm^3 [Fig. 3]. On microscopic examination, the tumor developed mainly from the subserosal tissue of the intestine and consisted of spindle cells and a collagenous background. Increased cell density and nuclear fission images were not noticeable. Immunohistochemically, the tumor cells were positive for nuclear β-catenin and negative for c-kit, CD34, DOG-1, δ-desmin, and S-100. The Ki67 index was less than 5% [Fig 4]. Based on these findings, the final diagnosis was not GIST, but desmoid-type fibromatosis. In addition, there were no

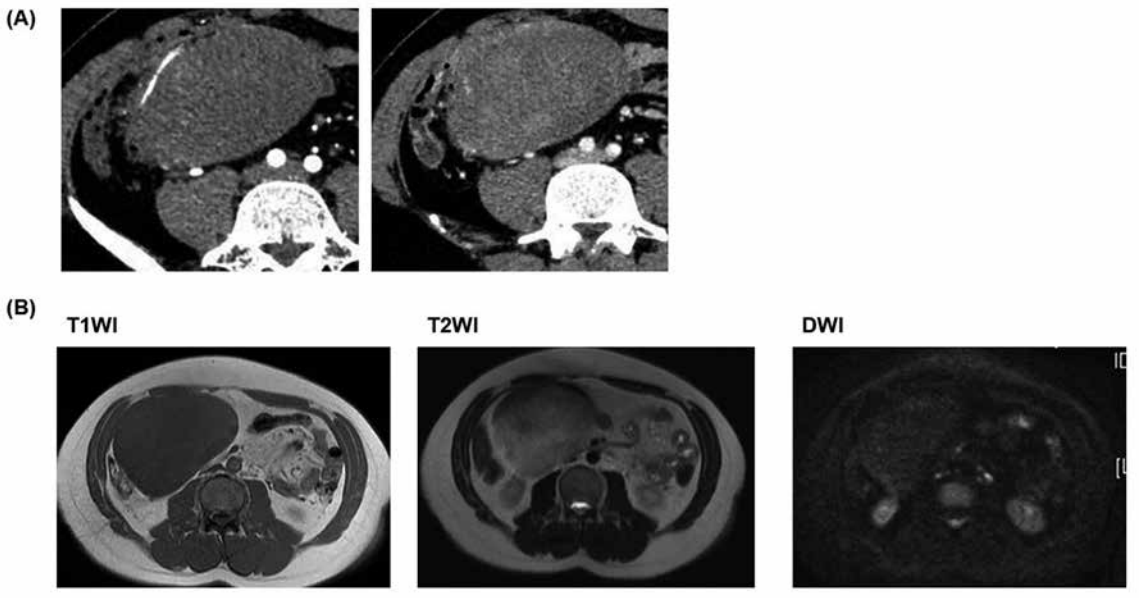

Fig 2. (A) Abdominal CT shows a large, 12-cm mass. The artery is compressed by tumor in artery phase. Enhancement is very weak and homogeneous in venous phase.
(B) MRI shows a T1WI low, T2WI high, and DWI low-intensity mass.

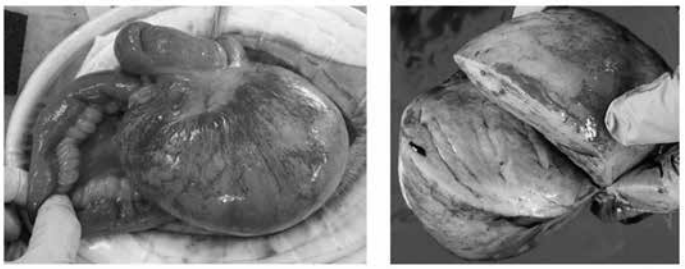

Fig 3. The resected specimen shows a whitish and solid tumor. There is no necrotic part inside the tumor.

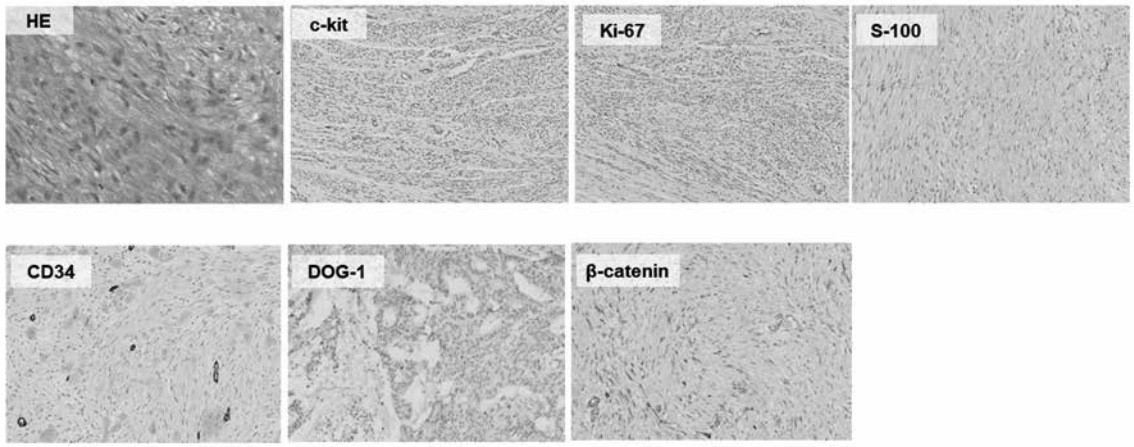

Fig 4. Hematoxylin-eosin (HE) staining shows spindle cells and a collagenous background. On immunohistochemical analysis, nuclear β-catenin staining is positive, and other stromal markers are negative.

lymph node metastases. About 1 year has passed since the operation, but no recurrence has been observed.

DISCUSSION

Fibromatosis is classified into 2 types according to the site of its occurrence : superficial fibromatosis and deep fibromatosis (desmoid-type). Furthermore, desmoid-type fibromatosis is classified by the site of occurrence into extra-abdominal (49%), abdominal wall (43%), and intra-abdominal cavity (8%) (2,3). Surgical resection is the first-choice treatment, but the recurrence rate is high (25-76%) (4). In addition, it is said to be associated with familial adenomatous polyposis or a history of abdominal surgery and trauma.

The typical CT findings of desmoid fibromatosis are a homogeneous and solid tumor showing weak enhancement. On MRI, it shows iso-low intensity on T1WI and low density on T2WI and DWI. On the other hand, in the case of GIST, CT shows homogeneous inside enhancement when the tumor is small, but as the tumor grows, the inside becomes necrotic and bleeding, the margin shows enhancement, and the center shows low enhancement. Furthermore, on MRI, GIST shows low intensity on T1WI and high intensity on T2WI and DWI (5-8). The findings of some imaging modalities are summarized in Table 1.

Histopathologically, the tumors are moderately cellular, infiltrative, and composed of spindled cells with only mild nuclear atypia. On immunohistochemical staining, desmoid-type fibromatosis is characteristically nuclear β-catenin positive (9), and ≥ 80% of desmoid tumors show at least focal nuclear staining for β-catenin. Desmoid-type fibromatosis is usually negative for KIT and CD117 and lacks reactivity for CD34, DOG-1, and S-100. On the other hand, GIST often expresses CD117, CD34, and S-100 and is usually negative for β-catenin (10). In this case, since some markers of gastrointestinal stromal tumors were negative, and nuclear β-catenin was positive on immunohistochemical staining, desmoid-type fibromatosis was diagnosed.

This case was difficult to differentiate from GIST. Differentiating desmoid-type fibromatosis and GIST is sometimes difficult because they overlap clinically, macroscopically, and histologically (10). Other differential diagnoses include lymphoma, inflammatory pseudotumor, and metastasis, etc. There was no history of FAP, trauma, and surgery in the present case. Furthermore, since intra-abdominal fibromatosis is rare, desmoid-type fibromatosis was not initially considered. In the present case, despite the formation of a large, 12-cm mass, there were no necrotic findings inside the tumor, and DWI showed high intensity ; thus, the diagnosis of GIST was incorrect in retrospect. Immunological staining appeared to be the decisive factor in making the diagnosis.

Table 1.

	CT	MRI	US
Fibromatosis	Homogeneous Solid Weak enhancement	T1 : Iso-Low T2 : Low DWI : Low	Low
Small GIST	Homogeneous enhancement	T1 : Low T2 : High DWI : High	Low
Large GIST	Peripheral- enhancement Central necrosis Internal bleeding		
Our case	Homogeneous Weak enhancement	T1 : Low T2 : Iso-high DWI : Low	Low

CONCLUSION

Desmoid-type fibromatosis should be considered in patients with a mesenteric mass, but its diagnosis is difficult by diagnostic imaging alone. Immunohistochemical evaluation can provide the diagnosis of desmoid-type fibromatosis.

COMPETING INTERESTS

The authors declare no conflict of interests for this article.

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None

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