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CASE REPORT

Diagnostic imaging and CEUS findings in a rare case of Desmoid-type fibromatosis. A case report

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Abstract
Desmoid-type fibromatosis (DF), also known as aggressive fibromatosis, is a locally aggressive benign fibroblastic neoplasm that can infiltrate or recur but cannot metastasize. It is rare, with an estimated annual incidence of two to four new cases per million people. Most DFs occur sporadically, but it may also be associated with the hereditary syndrome familial adenomatous polyposis. Treatment is necessary when the disease is symptomatic, especially in case of compression of critical structures. When possible, surgical resection is the treatment of choice; however, recurrence is common. Due to the high rate of recurrence, imaging plays an important role not only in diagnosis, but also in the management of DF. Although there are a number of studies describing CT and MRI findings of DF, there is no description of contrast-enhanced ultrasound findings.

Keywords Desmoid-type fibromatosis · Aggressive fibromatosis · CEUS · Ultrasound

Sommario
La fibromatosi di tipo desmoide (DF), anche conosciuta col nome di fibromatosi aggressiva, è una neoformazione benigna localmente aggressiva con la capacità di infiltrare o recidivare ma incapace di metastatizzare. È rara con un'incidenza annuale stimata di 2-4 nuovi casi per milione, costituendo approssimativamente lo 0.03% di tutte le neoplasie e meno del 3% dei tumori dei tessuti molli. La maggior parte delle DF sono sporadiche ma possono anche essere associate con la sindrome della poliposi familiare (FAP). Il trattamento è necessario quando la malattia è sintomatica, specialmente in caso di compressione di strutture critiche. Quando possibile, la resezione chirurgica è il trattamento di scelta, sebbene la recidiva sia comune. Visto l'elevato tasso di ricadute, l'imaging gioca un ruolo importante non solo nella diagnosi ma anche nella gestione della DF. Sebbene vi siano un certo numero di studi che descrivono le caratteristiche della DF in TC e RMN, non c'è alcun lavoro che descriva la patologia nell'ecografia con mezzo di contrasto (CEUS).

Introduction

Desmoid-type fibromatosis (DF), also known as aggressive fibromatosis, is a locally aggressive benign fibroblastic neoplasm that can infiltrate or recur but cannot metastasize. DF can arise anywhere in the body. It is rare, with an estimated annual incidence of two to four new cases per million people, accounting for approximately 0.03% of all neoplasms and less than 3% of all soft tissue tumors [1, 2]. Most DFs occur sporadically, but it may also be associated with the hereditary syndrome familial adenomatous polyposis (FAP). The combination of FAP and DF is known as Gardner's syndrome [3]. DF is classified, based on the location, as either extra-abdominal or intra-abdominal. Intra-abdominal DF may occur sporadically when associated with FAP as Gardner's syndrome; it typically manifests as slow-growing masses and may present with a series of complications, including intestinal obstruction and bowel ischemia.

Treatment is necessary when the disease is symptomatic, especially in the case of compression of critical structures. When possible, surgical resection is the treatment of choice [4]; however, recurrence is common (19–77%) [5, 6]. Non-surgical treatment options include radiation (as an adjuvant

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treatment to reduce the risk of local recurrence) and systemic therapy (cytotoxic agents, molecular-targeted agents and anti-estrogen therapy).

Due to the high rate of recurrence, imaging plays an important role not only in diagnosis, but also in the management of DF. Although there are a number of studies describing CT and MRI findings of DF [7–13], there is no description of contrast-enhanced ultrasound (CEUS) findings.

Case description

A 66-year-old woman was admitted to the emergency department of our hospital for abdominal pain in the left flank. The patient described accidental trauma 10 days before and worsening pain over the last 3 days. A FAST examination showed no peritoneal fluid, and an abdominal X-ray showed subocclusion. Contrast-enhanced CT was performed, showing a large, poorly enhanced mass in the left abdomen, suggesting an intestinal tumor (Fig. 1a, b). A similar situation was observed using MRI after injecting gadolinium contrast agent (Fig. 2a, b). The patient's condition deteriorated during the night and the surgeon requested an ultrasound (US) examination. The US showed a predominantly solid hypoechoic mass with a cystic portion and poor color Doppler signal (Fig. 3a, b). Contrast-enhanced US (CEUS) was performed using a low mechanical index (MI) (range 0.04–0.1) after the administration of SonoVue (Bracco™). SonoVue is a blood pool US contrast agent consisting of microbubbles. A total of 4.8 ml was administered in two intravenous bolus doses of 2.4 ml. The second dose was injected 15 min after the first, and both were followed by a 5-ml saline flush [14]. The CEUS showed hyper-enhancement in the arterial phase with later washout after 110 s. The contrast agent also showed persistence in the late phase after 3 min (Fig. 4a, b). The patient underwent video-assisted laparoscopy and the final diagnosis was DF (Fig. 5a–d).

Discussion

DF is an uncommon disease that is benign, but aggressive due to its local infiltration capacity. The disease originates from an irregular proliferation of well-differentiated fibroblasts resulting from the abnormal healing of previous tissue damage. Onset is associated with hormonal factors, genetic predisposition and atypical connective tissue synthesis [15, 16]. The highest incidence is recorded in young adults (20–40 years), with no difference between genders

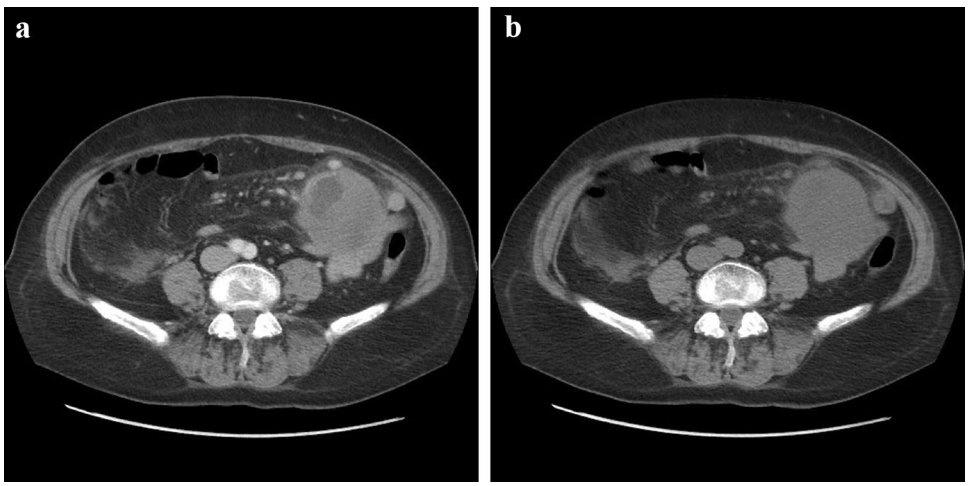

Fig. 1 **a**, **b** Contrast-enhanced CT: **A** venous phase; **B** late phase. Early enhancement of the mass and persistent enhancement within the mass in the late phase

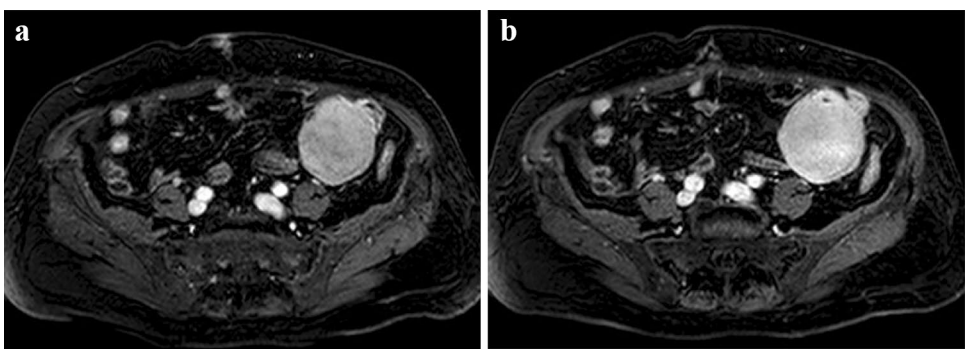

Fig. 2 **a**, **b** Contrast-enhanced MRI: **A** venous phase; **B** late phase. Early enhancement of the mass and persistent enhancement within the mass in the late phase

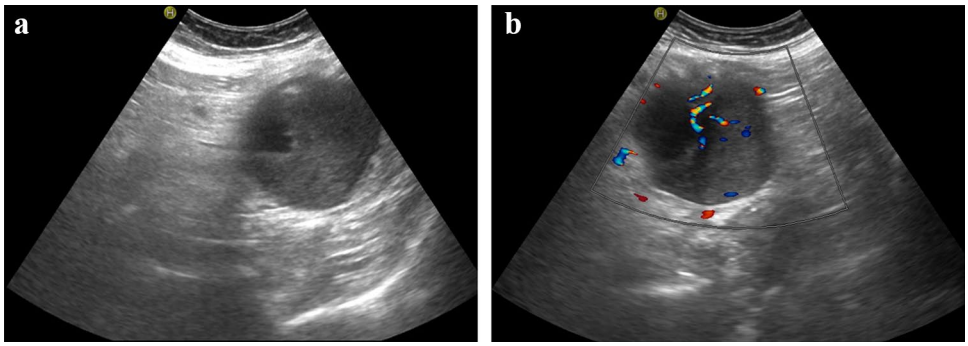

Fig. 3 **a**, **b** US: **A** grayscale; **B** US color Doppler shows a solid mass with a liquid area. Color Doppler shows the presence of vessels within the mass

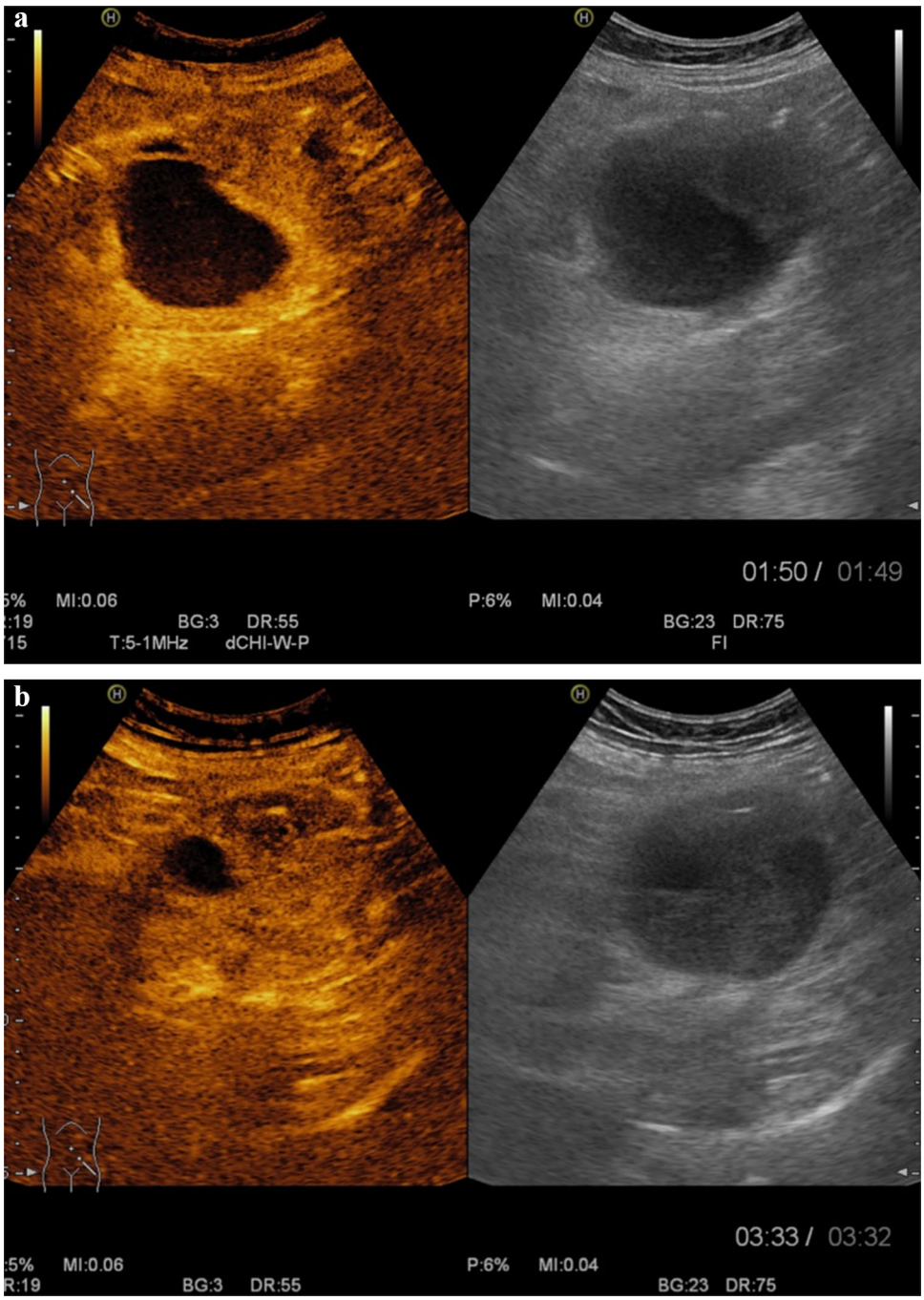

Fig. 4 **a**, **b** CEUS: **A** venous phase; **B** late phase. Enhancement during the venous phase (110 s) and presence of contrast agent in the late phase (210 s)

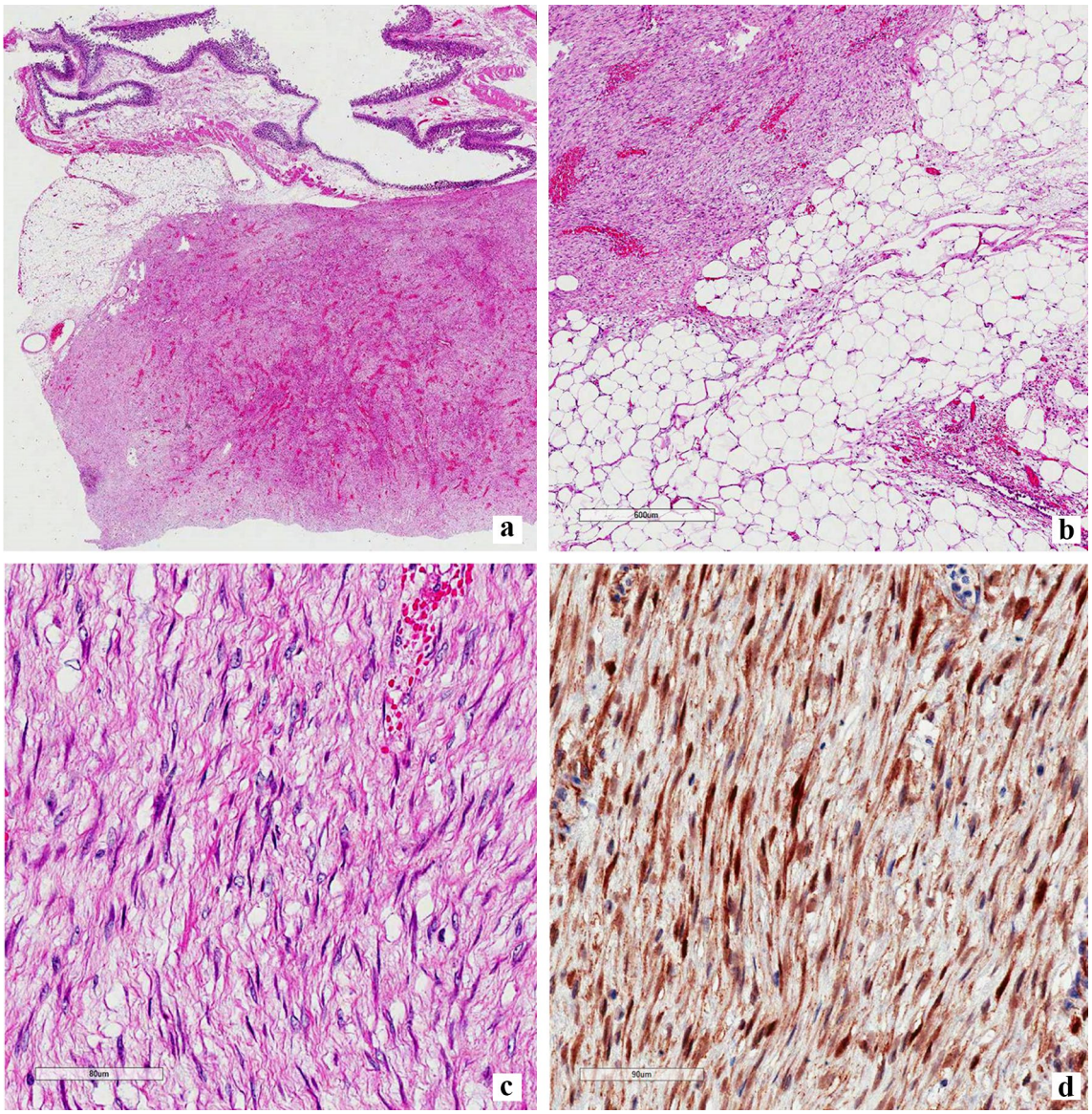

Fig. 5 **a–d** Histological examination showing: **A** panoramic view of the bowel wall with extramural fibroproliferative process (hematoxylin–eosin); **B** infiltration of the perivisceral adipose tissue (magnification ×50, hematoxylin/eosin); **C** details showing low-grade fusocellular proliferation and delicately fibrillated cytoplasm; the karyokinetic process is insignificant. There is characteristic leakage of erythrocytes (magnification ×200, hematoxylin/eosin); **D** strongly positive for β-catenin (magnification ×200)

[15]. The clinical features of DF depend on the localization of the lesion, since this neoplasm is usually asymptomatic until it compresses or infiltrates nearby structures.

In the described case, the patient experienced accidental trauma 10 days before admission. Trauma has been reported to have a role in triggering the proliferation of a neoplastic mass [17–19]. This is probably true, but it cannot explain our case, since the time interval between the trauma and the onset of symptoms was too short. It is more likely that the neoplastic mass had been growing slowly over a long period of time and that the clinical signs resulted from the trauma.

All diagnostic images were performed immediately after hospital admission. The CT scan and MRI suggested a diagnosis of intestinal tumor.

The US and CEUS examinations were performed in emergency.

CEUS has previously been proven effective for several organs [20–24]. However, to the best of our knowledge, this is the first report of the use of CEUS in the diagnosis of DF. The good temporal resolution of CEUS allows the best assessment of enhancement and washout times. Very early enhancement is characteristic of a benign mass, meaning a vascularization from the peripheral vessel, while later

enhancements could indicate different mesenchymal origins. Washout times could also be meaningful. In a recent study, early washout (/120 s) was found to be characteristic of malignant tumors [9]. Using CEUS, we were able to depict a well-vascularized tumor, with late wash-in and early washout, all of which suggested a malignant tumor [9, 10]. In our case, after early enhancement of the contrast agent, we relieved the very long washout: a typical aspect of benign lesions probably due to the presence of fibrotic tissue.

CEUS can depict very well vascular patterns and the relation to adjacent major blood vessels. However, the general principles of CEUS examination may be applicable to DF, thus making the diagnosis more accurate.

Compliance with ethical standards

Conflict of interest The authors declare no competing financial interest.

Ethical approval Ethics committee approval was obtained before beginning any research involving human subjects or clinical information. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000.

Informed consent All patients provided written informed consent for enrollment in the study and for the inclusion in this article of information that could potentially lead to their identification.

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