Radiology Case Reports 17 (2022) 2910–2914

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journal homepage: www.elsevier.com/locate/radcr

Case Report

Retroperitoneal desmoid tumor in a patient with familial adenomatous polyposis: A case report☆,☆☆

Fathia Aidid, MD[a], Narjisse Aichouni, MD[a], Ihssane Afilal, MD[a,], Widad Abbou, MD[a], Rachid Jabi, MD[b], Nadir Miry, MD[c], Amal Bennani, MD[c], Imane Kamaoui, MD[a], Siham Nasri, MD[a], Imane Skiker, MD[a]*

[a] *Department of Radiology, Faculty of Medicine, Mohammed VI University Hospital, University Mohammed First, BP 4806 Oujda University 60049, Morocco*

[b] *Department of General Surgery, Faculty of Medicine, Mohammed VI University Hospital, University Mohammed First, BP 4806 Oujda University 60049, Morocco*

[c] *Department of Pathology, Faculty of Medicine, Mohammed VI University Hospital, University Mohammed First, BP 4806 Oujda University 60049, Morocco*

ARTICLE INFO

Article history:
Received 18 April 2022
Revised 30 April 2022
Accepted 5 May 2022

Keywords:
Retroperitoneal
Desmoid tumor
Familial adenomatous polyposis
Rare tumor

ABSTRACT

Desmoid tumors are benign fibroblastic neoplasms, with locally invasive features and a tendency of recurrence. They are considered an aggressive non-metastatic fibromatosis. The retroperitoneal location is extremely rare. Their exact mechanism of occurrence is still controversial, but could be related to a genetic predisposition, hormonal factors or traumatic factors, including surgery. This entity faces management difficulties due to its rarity, the variable circumstances of its discovery, and the non-specific clinical manifestations. Their sensitivity to chemotherapy and radiotherapy is limited and surgery remains the only curative treatment in symptomatic cases, however observational waiting could consist the most appropriate management in selected asymptomatic patients, moreover it could avoid unnecessary morbidity from surgery or radiotherapy, which makes the management of this condition a multidisciplinary decision and should be adapted to fit the patients individually. We report a case of a retroperitoneal desmoid tumor in a 31-year-old woman with a history of familial adenomatous polyposis, through which we will discuss this extremely rare neoplastic entity.

☆ Competing Interests: No conflict of interest exists. We wish to confirm that there are no known conflicts of interest associated with this publication and there has been no significant financial support for this work that could have influenced its outcome.

☆☆ Funding: No funding was received for this work.

* *Corresponding author.*
E-mail address: ihssane.afilal@gmail.com (I. Afilal).

https://doi.org/10.1016/j.radcr.2022.05.013

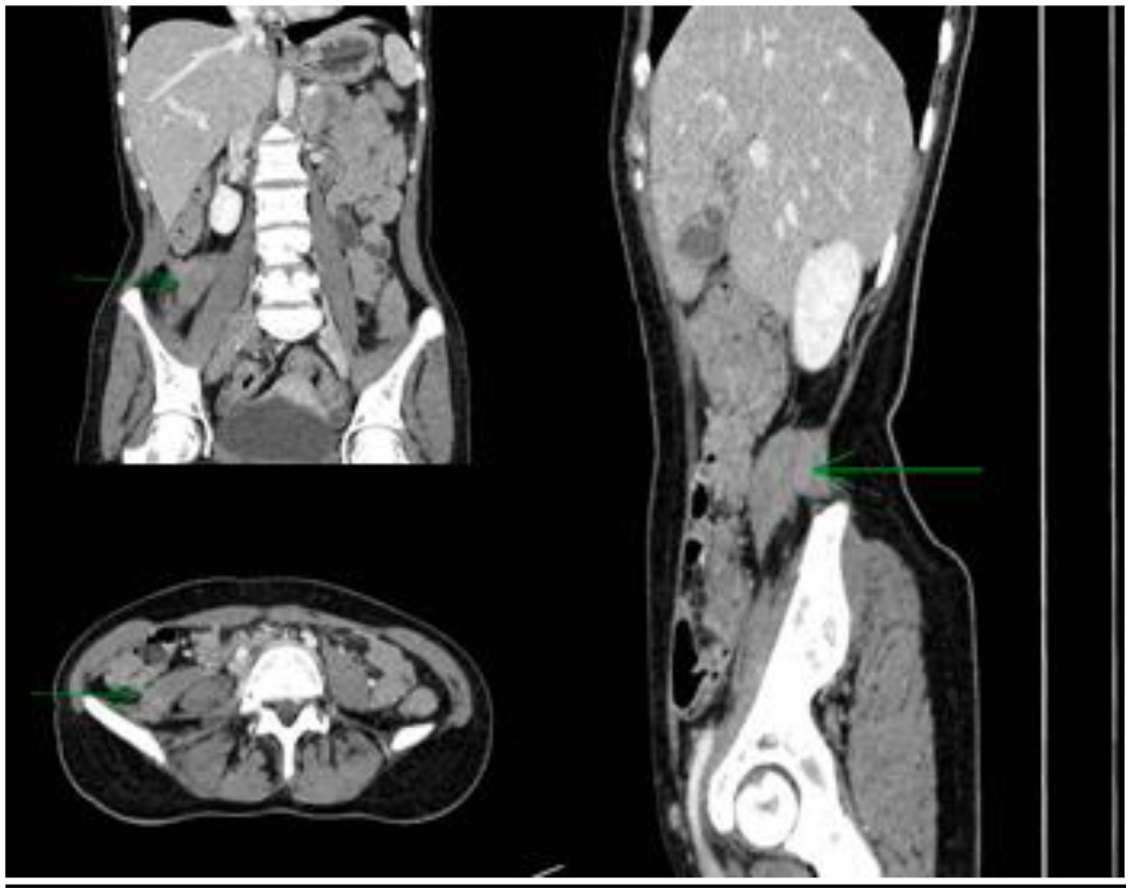

Fig. 1 – Coronal, axial, and sagittal sections of postcontrast CT scan, showing a well-circumscribed retroperitoneal mass, demonstrating soft tissue density and homogeneous enhancement after contrast, confirmed histologically to be a desmoid tumor (arrow).

Introduction

Desmoid tumors are defined as rare, locally aggressive, benign fibrous neoplasms that arise from the mesenchymal cell lines. This neoplastic entity is characterized by a high tendency of local recurrence even after complete surgical resection, but lacks metastatic potential. Though the exact etiology is still unknown, it has been reported that traumatic injury including surgical operations could be incriminated. On the molecular level, desmoids are related to mutations in β-catenin gene, CTNNB1, or the adenomatous polyposis coli (APC) gene [1]. It presents as an insidiously growing and locally invading fibroblastic proliferation with a variable and often unpredictable clinical course. The natural evolution of this tumor is variable and ranges from spontaneous regression, to stability for a long period of time, to invasion of local organs and neuro-vascular structures. Optimal management remains a controversial issue. Treatment options include observation and surveillance, surgery, cytotoxic chemotherapy, radiotherapy and NSAIDs as well as antiestrogens that have been proven to reduce the risk of recurrence [2]. However, it is a clinical challenge to distinguish recurrent malignant neoplasms from desmoid tumors and surgical resection remains the first line therapy, depending on the anatomical location, which makes the basis of care for these patients; a multidisciplinary concertation with multimodal management.

We report a case of retroperitoneal desmoid tumor that was fortuitously diagnosed in a 31-year-old woman, with a history of familial adenomatous polyposis, presenting to the emergency department with recurring abdominal pain, through which we will discuss this extremely rare neoplastic entity.

Patient observation

We report a case of 31-year-old woman, with a history of psoriasis and familial adenomatous polyposis. The patient benefited from a colonoscopy that showed a degenerating aspect of her familial adenomatous polyposis with a positive histological result in endoscopic biopsies for adenocarcinoma. A CT scan was ordered to evaluate the extension of the adenocarcinoma, revealing a well-circumscribed mass, in the right retroperitoneal cavity, demonstrating soft tissue

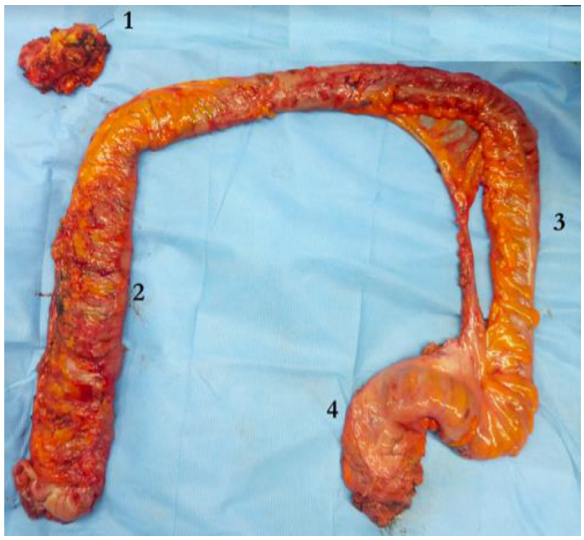

Fig. 2 – Macroscopic view of the excised specimens. 1. The retroperitoneal mass. 2. The ascending colon. 3. The descending colon. 4. The sigmoid colon.

density and homogeneous enhancement after contrast, associated with fat stranding all around (Fig. 1). This mass appeared to attract and infiltrate the inferior duodenal flexure. Bowel wall thickening of the sigmoid colon and the ascending colon were also noted. Considering the history of familial adenomatous polyposis and the context of malignancy, the main diagnostic differential for the mass was a carcinomatous mass or a desmoid tumor even though the retroperitoneal location was extremely rare. According to these findings, a total colectomy with ileo-anal anastomosis and removal of the retro-peritoneal mass infiltrating the inferior duodenal flexure (Fig. 2). Histological examination of the mass shows a diffuse and well-circumscribed proliferation arranged in multiple, long fascicules, arising in the duodenal wall. Tumor cells are spindle shaped, with no necrosis or mitotic figures; the cytoplasm is pale and scarce. The stroma is scant and fibrous, and contains numerous mast cells and prominent thin-walled blood vessels (Fig. 3). Immunohistochemical staining shows characteristic nuclear positivity of tumor cells for Beta-catenin; the tumor is also positive for SMA but negative for CD34.

According to these pathology findings the diagnosis of a desmoid tumor was retained. This case emphasizes the importance of frequent follow-up of patients with familial adenomatous polyposis. Desmoid tumor should be considered in the differential diagnosis of retroperitoneal masses in patients with a history of familial adenomatous polyposis. Although not metastatic, desmoid tumors can be locally aggressive, and complete excision is required to minimize the risk of recurrence. In some cases, desmoid tumors can be the primary lesion to indicate a diagnosis of familial adenomatous polyposis. Patients with such predisposition should be followed up closely after surgical excision of desmoid tumors.

Discussion

Desmoid tumor is a rare and heterogeneous entity, representing 0.03% of all neoplasms and less than 3% of soft tissue tumors [3]. Their prevalence reaches 13% in patients with familial adenomatous polyposis coli [4]. Although it is a locally aggressive fibroblastic proliferation, it is considered a benign neoplasm histologically with lack of metastasizing proprieties. The exact etiology is still debatable. However, its likely pathogenesis includes hormonal factors; including high estrogen states such as pregnancy, trauma, surgical interventions and genetic abnormalities. On the molecular level, desmoids are related to mutations in the beta-catenin gene, CTNNB1, or the adenomatous polyposis coli (APC) gene [1]. Moreover, histologically, desmoids are characterized by a monoclonal fibroblastic proliferation appearing as small bundles of spindle cells in an abundant fibrous stroma, with low cellularity and no features of malignancy. It presents as an insidiously growing and locally invading fibroblastic proliferation with a variable and often unpredictable clinical course. Physicians should keep in mind the possibility that a postoperative desmoid tumor may mimic a recurrence of the initial neoplasm, especially when the tumor presents at the previous surgical site. Multifocal presentations are exceptional but possible and recurrence can be local or regional, but never metastatic [5]. The most common locations include the abdominal wall, trunk and rarely extremities, peritoneal cavity as well as the retroperitoneal cavity. Computed tomography and magnetic resonance are indicated for diagnosis and follow-up. Imaging determines the extension of the tu-

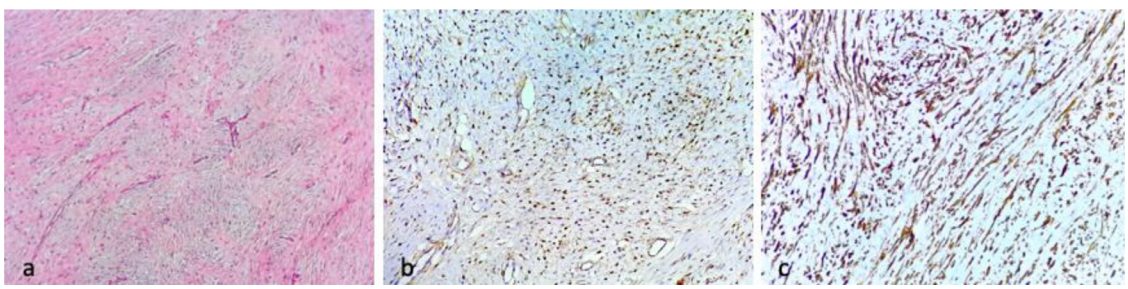

Fig. 3 – Histological findings in the desmoid tumor. (a) The stroma is scant and fibrous, and contains numerous mast cells and prominent thin-walled blood vessels (hematoxylin and eosin, original magnification ×100). (b) Immunohistochemical staining shows characteristic nuclear positivity of tumor cells for Beta-catenin. (c) The tumor is also positive for SMA.

mor, the infiltrated organs, and guides the surgical intervention plan. On CT scan, desmoids usually appear as a well-circumscribed homogeneously enhancing soft tissue density masses, although in some cases they may appear more aggressive with ill-defined margins [6]. On MRI T1-weighted sequences, desmoid tumors are hypo or iso-intense compared to the muscle, and hyperintense on T2-weighted sequences with gradual postcontrast enhancement [7]. Treatment options for desmoid tumors can be divided into 3 categories: asymptomatic resectable tumors, symptomatic resectable tumors, and unresectable and recurrent tumors. For the first category, most reviews of literature recommend that the observation and surveillance approach constitutes a reasonable option for small tumors in non-threatening locations. In case of progression a definitive therapy such as surgery or radiotherapy could be indicated [4]. For the second category, surgical resection is the main treatment option. Recurrence depends on the following risk factors: the size of the tumor, the location of the mass, the patient age, genetic predispositions, and the resection margins. It should be considered that extra-abdominal desmoids have a higher recurrence risk than intra-abdominal sites. Studies have shown that postoperative radiotherapy seems to reduce the risk of recurrence after a complete surgical resection [8]. For the third category, treatment options include antihormonal therapy, NSAIDs, Tyrosine kinase inhibitors, cytotoxic chemotherapy, radiotherapy and close observation [2]. The treatment response in this category is very slow, but with satisfying local control rates [9]. Follow-up depends on the location of the desmoid tumor and is usually based on clinical examination and appropriate imaging studies every 6 months for the first 3 years, then every year for the next 3 years, and then once every 2 years [3].

Conclusion

Desmoid tumor is a benign fibroblastic proliferation and the retroperitoneal location is extremely rare. This neoplastic entity is particularly seen in patients with familial adenomatous polyposis and it has a tendency for local invasion and recurrence, as well as variable topography and evolution, making the diagnosis, treatment and follow-up a multidisciplinary care team issue. Management should be individualized to optimize local tumor control and preserve the patients' quality of life.

Patient consent

I confirm that I have obtained written consent from the patient's guardian to publish: Retroperitoneal desmoid tumor in a patient with familial adenomatous polyposis: A case report

I informed the patient of the following:

- Although the patient's name will not be published, details of her case may mean that complete anonymity cannot be guaranteed.
- This article will be published in a medical journal, which is distributed electronically and on paper to doctors, nurses, and other medical personnel.
- The article, in whole or in part, may be available on websites accessible to members of the public.
- Nothing contained in the article will be used for commercial purposes or used out of context.
- The signed consent form will be retained by the corresponding author of the paper; copies will not be sent to anyone else involved in the publication or distribution of the article.

I will retain a signed copy of the patient consent form.

Intellectual property

We confirm that we have given due consideration to the protection of intellectual property associated with this work and that there are no impediments to publication, including the timing of publication, with respect to intellectual property. In so doing we confirm that we have followed the regulations of our institutions concerning intellectual property.

Research ethics

We further confirm that any aspect of the work covered in this manuscript that has involved human patients has been conducted with the ethical approval of all relevant bodies and that such approvals are acknowledged within the manuscript.

Written consent to publish potentially identifying information, such as details or the case and photographs, was obtained from the patient(s) or their legal guardian(s).

Authorship

The International Committee of Medical Journal Editors (ICMJE) recommends that authorship be based on the following four criteria:

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2. Drafting the work or revising it critically for important intellectual content; AND
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