

# Intra-abdominal desmoid tumor: Clinical case and literature review

## *Tumor desmoide intraabdominal. Presentación de caso clínico y revisión de la literatura*

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### Abstract

Desmoid tumors are clinical entities rarely diagnosed at an initial presentation because of its low incidence, they are characterized by a locally aggressive presentation and high rates of local relapse. Its presentation can be intra- or extra-abdominal. We present a clinical case of a female, a 15-year-old patient, with 3 months of abdominal pain, a giant intra-abdominal mass was diagnosed with histologic diagnosis of desmoid tumor. Several surgical procedures were performed, having a R1 resection (focally microscopic margins). In this case, association with pregnancy, abdominal trauma, previous surgeries, and genetic syndromes were discarded.

**Keywords:** Desmoid tumor. Intra-abdominal. Surgery.

### Resumen

Los tumores desmoides son afecciones benignas raramente reconocidas de manera inicial por su baja incidencia. Se caracterizan por un comportamiento local agresivo y altas tasas de recurrencia. Su presentación puede ser extraabdominal o intraabdominal. Presentamos el caso de una paciente de 15 años con cuadro de dolor abdominal de 3 meses de evolución, en la que se documentó una gran masa intraabdominal con diagnóstico histológico de tumor desmoide y fue sometida a múltiples intervenciones quirúrgicas con las que se logró una resección R1 (microscópicamente positivo). Se descartó su asociación con embarazo, trauma abdominal, cirugías previas y síndromes genéticos.

**Palabras clave:** Tumor desmoide. Intraabdominal. Cirugía.

### Introduction

Desmoid tumors are benign neoplasms characterized by a proliferation of myofibroblastic cells. They

possess low metastatic potential, but high local invasion capacity and an aggressive recurrent behavior. Desmoid tumors account for a minimal fraction of all soft-tissue tumors in general, and their most

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**Figure 1.** Contrast-enhanced abdominal tomography (June 2016) showing a heterogeneous density tumor lesion of intra-abdominal location (courtesy of Luis J. Palacios).

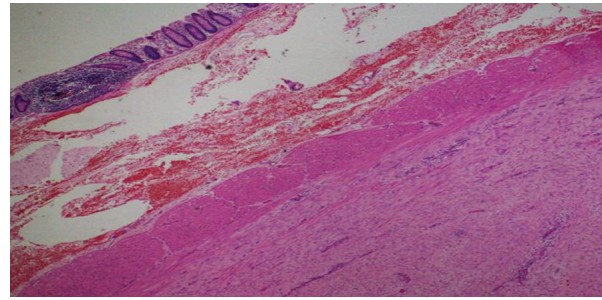
common presentation is in the abdomen. Sometimes, they are associated with familial adenomatous polyposis syndrome or Gardner syndrome.

Herein, we present the case of a 15-year-old female patient, previously healthy, with a 3-month history of abdominal pain, in whom an intra-abdominal desmoid tumor of sporadic onset was documented and who underwent surgical interventions with an R1 resection (microscopically positive) being achieved.

## Case presentation

A 15-year-old female student of mixed race, without medical, surgical, or family history. In August 2016, she attended her clinic for a 3-month history of epigastric pain, not irradiated, without other associated symptomatology. Initially, an upper digestive endoscopy examination was performed by the general medicine department, with "Important extrinsic compression of the gastric body" being described. Contrast-enhanced abdominal magnetic resonance imaging (MRI) showed a solid neoplastic intra-abdominal lesion suggestive of mesenchymal origin (Fig. 1). Subsequently, ultrasound-guided core needle biopsy of the lesion confirmed a desmoid tumor.

In June 2016, the patient underwent intra-abdominal tumor resection, extended right hemicolectomy, and descending colon-ileum anastomosis. On her 3<sup>rd</sup> post-operative day, due to persistent tachycardia and fever associated with acute-phase reactants elevation and findings on contrast-enhanced abdomen tomography consistent with pneumoperitoneum and free intra-abdominal fluid without contrast medium extravasation, performing a new surgical intervention was decided.

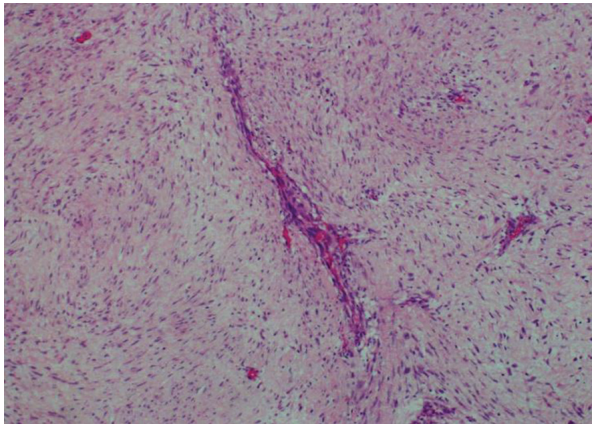


**Figure 2.** Representation of the colonic wall (upper right), in close contact with the tumor (lower left). H&E x 40 (courtesy of Alfredo Martelo).

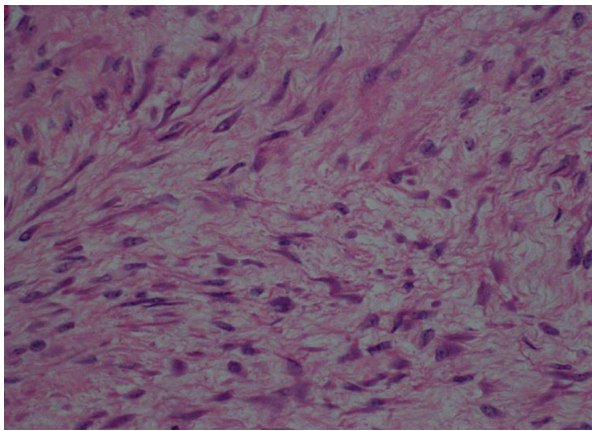
Unscathed anastomosis, without necrosis and without macroscopic areas of leakage, was confirmed. Deferred surgical wound closure was performed given the presence of clinical signs of infection. On the 17<sup>th</sup> post-operative day, the patient was discharged. Final pathology report indicated desmoid tumor-type fibromatosis measuring 30 cm on its largest diameter, right hemicolectomy with free resection margin pT-2bpN0 (Fig. 2). No further treatment was provided, due to the report of negative pathological margins, R0 surgery.

The patient continued on quarterly post-operative follow-up with abdominal tomography, with no signs of local recurrence and negative colonoscopy for intestinal polyps. In March 2018, she reported a non-painful mass sensation on the epigastrium, which prompted the performance of a contrast-enhanced abdominal tomography, with a lesion of soft-tissue density of approximately 21 × 50 mm being observed located toward the epigastric region, adhered to the anterior abdominal wall, and without a cleavage separation plane. Core needle biopsy showed results consistent with a desmoid tumor (Figs. 3-5). In May of same year, resection of the abdominal wall tumor and eventration was performed, with a desmoid tumor being identified on the abdominal wall, in the subxiphoid supraumbilical midline, measuring 10 × 10 cm, with an involvement of the anterior fascia of the rectus abdominis and the skin, and toward deeper planes, involvement of the left hepatic lobe, segment 2, distal end. A definitive pathology report of the lesion and the margins was obtained, which indicated a lesion consistent with desmoid tumor recurrence, 9 cm in size, with findings extended to the right lateral distal margin in contact with the tumor in a 2 mm area.

Being this the case of a young female patient who in the last procedure underwent a complex abdominal wall reconstruction (Fig. 6), and who had a slowly



**Figure 3.** Tumor with collagenized stroma with small blood vessels and fusiform cells proliferation. H&E x 100 (courtesy of Alfredo Martelo).

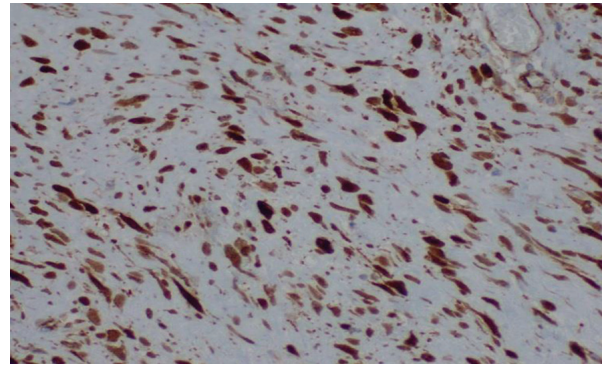


**Figure 4.** Detail of tumor cells, with characteristics of myofibroblasts. H&E x 400 (courtesy of Alfredo Martelo).

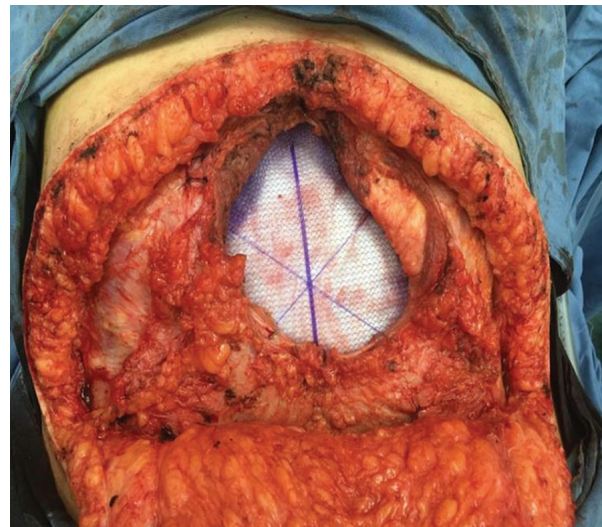
progressing disease, a meeting of institutional specialists was held, where it was decided, for the moment, not to subject the patient to any surgical intervention or additional medical treatments, based on institutional treatment guidelines, where R1 resections are also acceptable for providing adjuvant management, although on alert for further validation studies.

## Discussion

Desmoid tumors or desmoid-type fibromatosis are a rare and benign condition characterized by fibroblastic/myofibroblastic proliferation that can occur on any location of the body. The absence of capsule and presence of infiltration of adjacent tissues in a multifocal manner generate a high rate of local recurrence, which makes of their treatment a real medical challenge<sup>1</sup>.



**Figure 5.** Immunohistochemistry. The  $\beta$ -catenin marker shows strong and diffuse nuclear positivity in tumor cells. Immunohistochemistry staining ( $\beta$ -catenin marker) x 400 (courtesy of Alfredo Martelo).



**Figure 6.** Trans-operative image (April 2018). Supraumbilical defect without achieving closure on the midline, which required placement of a 30 x 30 cm component spacer mesh (courtesy of Luis J. Palacios).

Desmoid tumors have an incidence of 2-4 cases/million/year, accounting for 0.03% of all neoplasms and < 3% of soft-tissue tumors. These neoplasms are more common in women, with a 2:1 ratio with regard to men, and in the age group of 25-35 years<sup>1</sup>.

Most cases (80%) occur sporadically, but 2-15% of patients with desmoid tumors have associated familial adenomatous polyposis or Gardner syndrome; typical anatomical location of the lesion in these patients is the mesentery of the small intestine<sup>2-4</sup>. The *Adenomatous polyposis coli (APC)* gene has been shown to be involved in the control of  $\beta$ -catenin levels in the cytoplasm. The APC mutation present in familial adenomatous polyposis does not allow the degradation of  $\beta$ -catenin, which generates its translocation to the

nucleus and affects gene transcription.  $\beta$ -catenin is a protein encoded by the *CTNNB1* gene that regulates cell adhesion in the cytoplasm, as well as gene transcription in the nucleus. It is part of the cadherin protein complex and acts through Wnt intracellular signaling pathway. *APC* mutations are not present in most sporadic cases, but *CTNNB1* mutations are described in 85-88% of them<sup>5,6</sup>.

Desmoid tumor etiology is not fully established. It may appear in scars from previous surgeries, mainly of abdominal procedures, especially in patients with familial adenomatous polyposis. The most common site of occurrence in sporadic cases remains the abdominal wall<sup>7</sup>. An increased risk for post-pregnancy desmoid tumor is described, but a direct link between estrogen levels and the size of the lesion has not been demonstrated. Expression of sexual steroid receptors by the tumor is inconstant; according to this, and in the absence of randomized clinical trials, isolated hormone therapy has not been standardized in this disease<sup>8</sup>.

The appearance of a desmoid tumor at surgical scar sites reflects an alteration in cell repair process and makes  $\beta$ -catenin involvement evident in this phenomenon, which transiently rises in fibroblasts during healing processes. Although desmoid tumor cells are similar to fibroblasts with abundant collagen matrix, fibroblasts have not been described as their predecessor cells. Surface markers have been identified that coincide with aberrantly activating mesenchymal cells, which could be desmoid tumor progenitor cells<sup>8</sup>.

The disease has a silent course of slow and painless growth, with symptoms associated with infiltration or compromise of adjacent organs. Cases of multifocal occurrence are most often reported in the extremities or by post-operative recurrences<sup>8</sup>. Our patient initially presented with an insidious process of abdominal pain secondary to gastric compression. The imaging study of choice is contrasted abdominal tomography, which allows an adequate evaluation of the tumor relationships with intra-abdominal organs and reveals an isodense lesion to skeletal muscle with heterogeneous density within that corresponds to collagen or myxoid areas. In cases of desmoid tumors on extremities, MRI evaluation is indicated, with better performance with regard to abdominal MRI<sup>8</sup>.

Desmoid tumors are composed of fibroblast-like fusiform cells with no malignancy characteristics. These cells are surrounded by a dense collagen matrix and neovascularization of various calibers. They have positive  $\beta$ -catenin immunohistochemistry with higher

nuclear expression and vimentin.  $\beta$ -catenin nuclear expression is not specific to desmoid tumors, since it may be present in other mesenchymal neoplasms, such as synovial sarcoma, among others. *CTNNB1* mutation analysis in sporadic cases is a diagnostic tool in unclear situations<sup>8</sup>.

The reference method remains surgical resection of the lesion. The need to ensure negative microscopic margins is currently under discussion, since case series with similar recurrence rates is reported in R0 and R1 groups, which is why administration of complementary therapies with adjuvant purposes is being evaluated. Depending on the tumor site, the need for multi-visceral resections and the morbidity associated with the procedure should be taken into account; our case required, on its second intervention, wall reconstruction with component separating prosthetic material, given the large defect after tumor resection<sup>8</sup>.

On the other hand, regarding management with systemic therapies, the use of nonsteroidal anti-inflammatory drugs, such as sulindac, and hormone therapy with anti-estrogenic effect, such as tamoxifen or raloxifene, is indicated<sup>9</sup>. Systemic therapies as first-line treatment in intra-abdominal tumors associated with familial adenomatous polyposis show 77% of effectiveness, in comparison with patients who received post-recurrence therapy with 50% effectiveness. Other systemic therapies include chemotherapeutic agents such as anthracyclines, methotrexate, vinblastine, and cisplatin, with effectiveness rates of up to 79%. Therapies that have been also documented include tyrosine kinase inhibitors and antiangiogenic drugs that are in Phase II studies, showing favorable results<sup>10</sup>.

Finally, radiotherapy as an adjuvant to surgery in cases of desmoid tumor when microscopically positive margins (R1) persist is indicated as single therapy in case of surgical unresectability of the lesion, with local control and growth stability of 70-80% being obtained<sup>4</sup>. According to certain series of studies, achieving a local control of 78% is possible with radiotherapy alone and 75% with radiotherapy plus surgery, in comparison with 61% with surgery alone<sup>11</sup>. For intra-abdominal desmoid tumors, the use of radiotherapy is controversial due to its abdominal effects, such as actinic enteritis<sup>12</sup>.

## Conclusion

Intra-abdominal desmoid tumors suspicion and identification allow timely and appropriate management of

a pathology that, although rare, has a locally aggressive behavior. In our case, an R1 resection (microscopically positive) was achieved, which is acceptable for adjuvant management, although on alert for further validation studies. In addition, association with pregnancy, abdominal trauma or surgery, genetic syndromes such as Gardner syndrome, and familial adenomatous polyposis were ruled out in this patient.

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## Conflicts of interest

The authors declare that they have no conflicts of interest.

## Ethical disclosures

**Protection of human and animal subjects.** The authors declare that no experiments have been performed on humans or animals for this research.

**Confidentiality of data.** The authors declare that they have followed the protocols of their work center on the publication of patient data.

**Right to privacy and informed consent.** The authors have obtained informed consent from the patients and/or subjects referred to in the article. This document is in the possession of the corresponding author.

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