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Mesenteric-Jejunal Desmoid Tumor. Incidental Finding in a Patient with Acute Appendicitis: Case Report

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ABSTRACT

Desmoid tumors, formerly called aggressive fibromatosis and fibrosarcoma grade 1, are fibrous tumors that occur without predilection for any sex, in the third and fourth decade of life. They mainly occur in extremities and abdominal wall.

This case is a 53-year-old female who was diagnosed with an incidental jejunal tumor due to acute appendicitis. The patient was first treated for an attack of acute appendicitis, during the physical examination an epigastric mass became evident. When abdominal CT (computed tomography) scan was performed, the diagnosis of acute appendicitis was confirmed and the presence of a solid tumor was documented, which suggested involvement of the third and fourth portion of the duodenum.

Laparoscopic appendectomy was performed without complications. One week later and as an external patient, a new abdominal CT scan was performed, which suggested "GIST" (Gastrointestinal Stromal Tumor).

Study protocol was completed, and exploratory laparotomy was performed with tumor and jejunal diverticulum resection. Within the surgical findings, we observed a jejunum-dependent tumor with a diameter of 7 cm that was located 10 cm away from the Treitz angle. A block resection and a primary jejunal-jejunal anastomosis were performed. The patient was discharged on her POD 10 without any surgical complications.

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Abbreviations

DT: Desmoid Tumors
FAP: Familial Adenomatous Polyposis
TKIs: Tyrosine Kinase Inhibitors
CT: Computed Tomography
MRI: Magnetic Resonance Imaging
GIST: Gastrointestinal stromal Tumors

Introduction

The term "desmoid" comes from the Greek "desmos" which means "tendon-shaped" and began to be used in 1800 to describe tumors with this consistency.

Desmoid tumors, formerly called aggressive fibromatosis and fibrosarcoma grade 1 are fibrous tumors, most cases are diagnosed in people between 15 and 60 years of age [1,2]. However, the highest incidence is observed in the 30-40 age group. Furthermore, desmoid tumors affect women more frequently than men [3].

Their occurrence is more frequent in extremities and abdominal wall [2]. Although their potential to metastasize is practically nil, they are loco-regionally aggressive tumors, with high recurrence rates. Desmoid tumors in the abdominal cavity represent only 5% of the presentation of these tumors [4-5].

More than 90% of desmoid tumors (DT) develop sporadically and are related to alterations in the CTNNB1 gene, which encodes the β-catenin, the occurrence has been documented in women who use oral contraceptives, as well as in those in the gestational period or post-pregnancy [3]. They have also been associated with a history of trauma or previous surgical procedures, as well as being linked to genetic disorders such as familial adenomatous polyposis (FAP), which occurs in patients with an inherited mutation in the APC gene in 10-15% and Gardner syndrome [3, 6].

TDs may be asymptomatic at the time of diagnosis; when symptoms occur, they are usually related to pain and deformity caused by compression of adjacent organs, neurovascular structures or by erosion and destruction of nearby musculoskeletal components. Its clinical evolution is unpredictable, and it has been reported that up to 20% of cases may experience spontaneous regression [3].

The therapeutic approach to symptomatic and progressive desmoid tumors may require surgical intervention or medical treatment. In particular, desmoid tumors associated with familial adenomatous polyposis (FAP) are often managed more actively due to their more aggressive clinical behavior. Treatment options include antihormonal therapy, tyrosine kinase inhibitors (TKIs) and chemotherapy [3].

The diagnostic process of soft tissue neoplasms should include imaging studies such as ultrasound and computed tomography or magnetic resonance imaging (CT/MRI), which allow determining tumor size and affected structures, facilitating adequate preoperative planning. In particular, desmoid tumors with intra-abdominal location present a broad differential diagnosis that includes gastrointestinal carcinomas, lymphomas, gastrointestinal stromal tumors (GIST), solitary fibrous tumors, inflammatory myofibroblastic tumors, sclerosing mesenteritis and retroperitoneal fibrosis. In cases of complicated aggressive fibromatosis, the differential diagnosis becomes even more challenging, as it can be confused with more frequent causes of acute surgical abdomen, making early identification and proper management difficult [7].

Case Presentation

A 53-year-old female was diagnosed with an incidental jejunal tumor following acute appendicitis. The patient was first seen for an attack of acute appendicitis, and an epigastric mass became evident during the physical examination. An abdominal computed axial tomography (CT) scan (Figure 1) confirmed the diagnosis of acute appendicitis and documented the presence of a solid tumor, which suggested involvement of the third and fourth portions of the duodenum. The findings were discussed with the patient and her family, and the patient was scheduled for appendectomy by laparoscopy, which was performed in that hospitalization without complications, having informed them that, except for exceptional transoperative findings, the tumor diagnosed preoperatively, would be studied extensively in the postoperative period to define definitive management in a second time. The postoperative evolution was satisfactory and she was discharged after 48 hours.

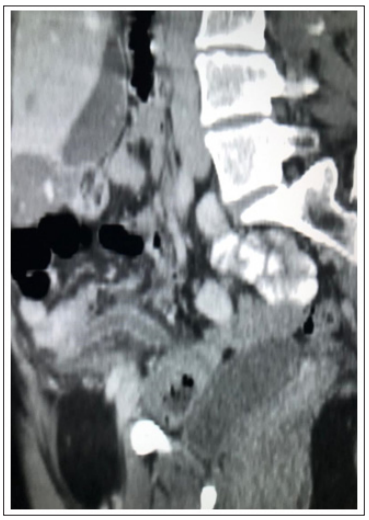

Figure 1: Axial and Coronal Computed Tomography. Acute Appendicitis and Carce Adjacent Free Fluid

One week later and as an outpatient, a new abdominal CT scan was performed suggesting as diagnosis; tumor of the junction of the third and fourth portions of the duodenum, compatible with "GIST" to rule out other tumor origin (Figure 2). Extended panendoscopy was performed, being possible to visualize the first 45 cm posterior to the pylorus, without visualizing endoluminal tumor or extrinsic compressive effect. Tumor markers were taken including CEA, Alpha-fetoprotein, DHL, Beta 2 microglobulin, Ca 19.9, Ca 125 and Ca15.3, all reporting normal parameters. A PET-CT scan was performed (Figure 3) which reported a xerophytic mural tumor lesion of the small intestine (at the level of the duodeno-jejunal transition), hypermetabolic in relation to tumor activity of origin to be determined, the rest of the study without other relevant findings and without data of metastatic lesions. A date was set for a new surgery and exploratory laparotomy was chosen.

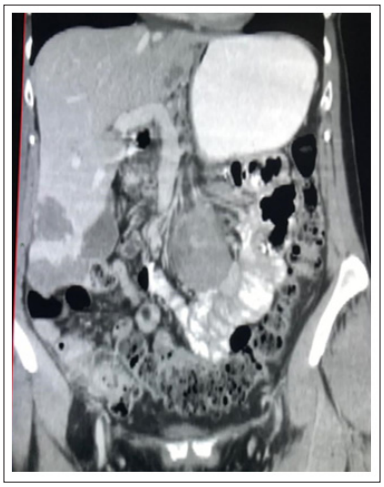

Figure 2: Abdominal Computed Tomography (Coronal Section). Mesenteric Lesion and Multiple Hemangiomas. It Shows a Mesenteric Lesion with Central Contrast Filling, Transit of the Distal Jejunum

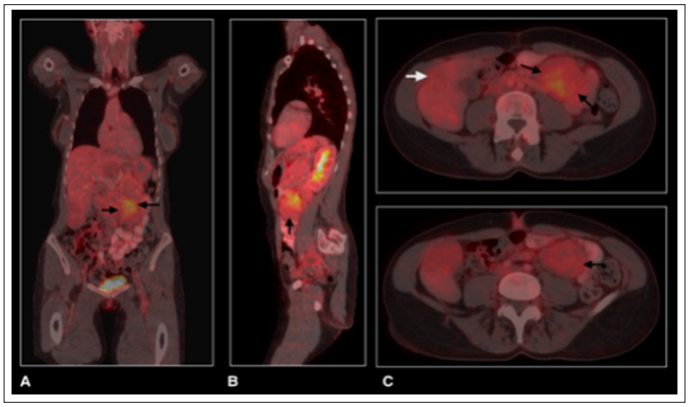

Figure 3: PET-CT. A) Coronal, B) Sagittal, C) Axial views Showing Mesenteric Lesion (Black Arrows) with 4.6 SUV and Liver Lesions without FDG Uptake (White Arrow)

Exploratory laparotomy was performed with resection of the tumor and jejunal diverticulum. Among the surgical findings, we observed a 7 cm diameter jejunum-dependent tumor 10 cm from the Treitz angle. A traction jejunal diverticulum was found attached to the tumor lesion, which was resected en bloc with a total length of 49 cm with an extensive and thorough dissection including mesentery in the surgical specimen, achieving oncologic resection (Figure 4 and 5) and with a primary jejunal-jejunal jejunal reconstruction. The rest of the abdominal cavity was thoroughly reviewed without finding any other lesion or pathology.

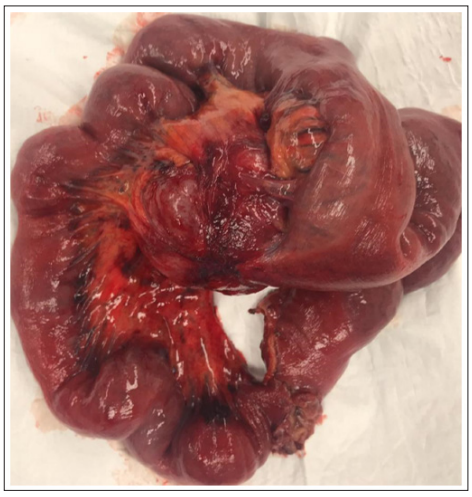

Figure 4: Resection of Mesenteric Tumor and Jejunal Diverticulum. Intestinal Retraction within the Mesenteric Lesion

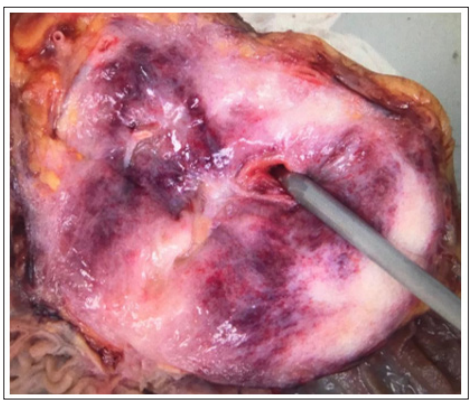

Figure 5: Macroscopic Pathologic Section. "Desmoid Tumor of Mesentery"

Results

The specimen sent to pathology consisted of a segment of intestine 48.7 cm long and in whose mesentery a nodular lesion of 8.2x6.3x4.4 cm could be identified. During dissection it was possible to identify that the lesion infiltrated the mesentery and extended irregularly retracting the intestinal wall of two segments. In a slice it was identified that the lesion invaded the mucosa of one segment resembling a diverticulum. The surface of the lesion was heterogeneous, alternating gray areas with diffuse areas of hemorrhagic stippling.

The final pathology report describes: Mesenteric fibromatosis desmoid type. 8.2 x 6.3 x 4.4 cm. Lesion infiltrates up to jejunal submucosa. Mitotic index of 3 mitoses per field. No necrosis, lymphovascular or perineural invasion. Negative borders for lesion. Nine mesenteric nodes negative for neoplasia.

The patient had an uneventful postoperative course, except for a gastroparesis that required management with prokinetics and parenteral nutritional support, leaving the hospital on postoperative day 10.

Discussion

Desmoid tumors of the mesentery represent only 10-15% of patients with FAP [8,9], however, they are loco-regionally aggressive tumors with a high incidence of recurrence despite appropriate surgical resection [10]. Up to 30% of patients with desmoid tumors report a history of trauma, particularly surgical trauma in patients with FAP following colon resections [11].

The etiology of these tumors is not entirely clear; however, they exhibit chromosomal changes that suggest a neoplastic origin. However, as already mentioned, there is a percentage that could be linked to a deregulation in the healing process not only in these tumors, but also in other types of tumors of fibroblastic origin [12].

Most of these tumors present as asymptomatic or minimally painful masses with slow growth. However, some of these tumors may become manifest when presenting with intestinal occlusion, anemia, ischemia or deterioration in the functionality of a previous anastomosis (ileoanal) in patients with FAP who required total colectomy [11].

They can occur in any anatomical region but predominate in the trunk and extremities, abdominal wall and intra-abdominal wall, the latter being the predominant in patients with FAP.

Imaging studies are required to completely delimit and characterize this type of tumors. CT is recommended for intra-abdominal tumors; however, for tumors in extremities, MRI is recommended, and ultrasound can be particularly useful for tumors in the thoracic or abdominal wall [12,13].

Histologically, they are tumors composed of fibroblasts and myofibroblasts in a dense stroma, rich in collagen with few mitotic figures. They show Beta-catenin expression which confirms that mutation of the CTTNB1 gene is linked to the expression of this molecule. Lazar et al. have described this mutation in 85% of desmoid tumors and attributed them to three different mutations in the 41A, 45F and 45P gene with ranges of 59%, 33% and 8%. Being the 45F mutation the one with the highest risk for recurrence at 5 years [14].

In the histologic sections of our case, a proliferative lesion of mesenchymal origin was identified, with a growth pattern in

long criss-crossing bundles. The cells are spindle-shaped without cytologic atypia and are surrounded by fibrous-like stroma between which there are occasional thin-walled blood vessels with a curvilinear appearance (Figure 6). Likewise, occasional perivascular lymphocytes were identified, as well as up to 3 mitoses per 50 high power fields.

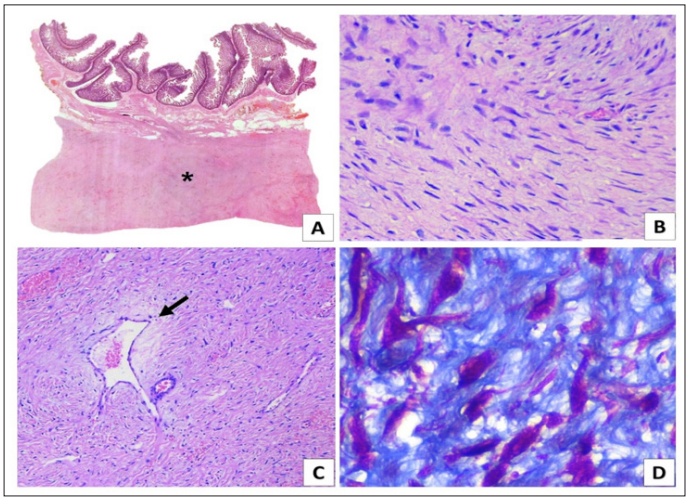

Figure 6: Pathological anatomy. Lesion Composed of Fibroblasts and Myofibroblasts, which form Long Bundles that Crisscross Each other, Irregularly Infiltrate the Mesentery and Extend into the Submucosa of the Intestine. No Necrosis or High Mitotic Index is Identified

Given the morphologic appearance of a low-grade spindle-shaped lesion, immunolabeling was performed with the following antibodies: smooth muscle actin, desmin, DOG-1, CD117, Vimentin, B-catenin and S100, of which B-catenin (nuclear) and Vimentin were positive; the rest of the antibodies were negative (Figure 7). The morphology together with the immunohistochemical result supported the diagnosis of mesenteric fibromatosis of the desmoid type and ruled out other neoplasms that can present the same histological image such as gastrointestinal stromal tumor, leiomyoma, schwanomma, neurofibroma and low-grade fibromyxoid sarcoma.

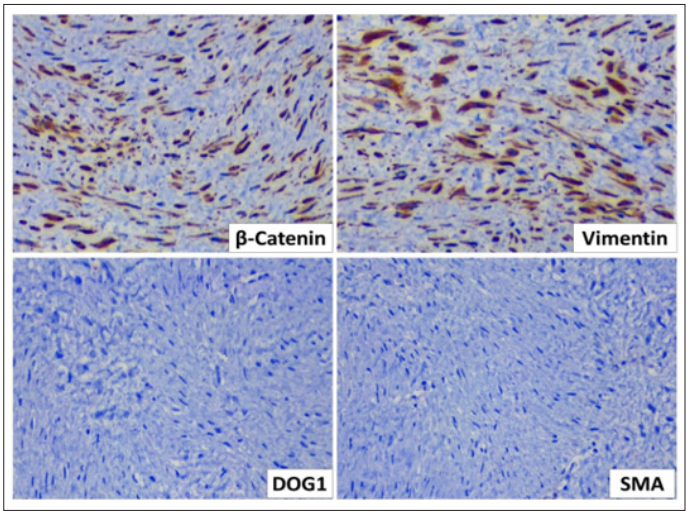

Figure 7: Immunohistochemistry. Beta-Catenin and Vimentin were Diffusely Positive. DOG1 is Weakly Expressed, so it is Complete with CD-117, S-100 Negative

Surgical resection is the only effective therapeutic option for the treatment of desmoid tumor and achieves very good results if there is the possibility of achieving R0. In any case, it will be necessary to continue surveillance exclusively [10]. In cases of R2, radiotherapy is indicated with 50-56 Gy in patients not previously irradiated and 50 Gy in patients with previous radiation.

Surgical therapeutic options exist for tumors previously considered unresectable involving large vessels of the mesentery. Intestinal autotransplantation is one of these, achieving good results. Initially described by Tzakis in 2003 and later by Quintini and Tzvetanov, in which bleeding and ischemia, responsible for perioperative morbi-mortality, are prevented [15 -18].

In our case, the tumor presented as an incidental finding and was confirmed radiologically. Given the prematurity of acute appendicitis, it was treated surgically in a first event. Subsequently, a diagnostic protocol was completed and a resection with negative margins (R0) was performed. She presented good postoperative evolution. At the moment she is under follow-up with surveillance only [19,20].

Conclusion

Desmoid tumors of the mesentery represent a rare entity. They are associated and related to PAF. In particular, this type of tumors may have a history of trauma, surgical or otherwise.

The presentation is generally asymptomatic with slow growth. The protocol for the study of these tumors should be complete and supported by imaging studies. The treatment of choice is surgical, so if it does not occur in the setting of an acute abdominal condition, it will require the greatest scrutiny and the best possible conditions for the patient before undergoing surgical resection. There are specific cases that merit highly specific procedures such as intestinal transplantation.

The use of technology has allowed us early diagnosis and characterization of different pathologies; however, the clinic remains our most useful tool as surgeons.

Conflict of Interest

The authors declare that there is no conflict of interest regarding the publication of this case report.

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