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A Rare Case of Caecal Desmoid Tumor

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Abstract

Desmoid tumors (DTs) are rare, benign fibroblastic neoplasms with local aggressiveness but no metastatic potential. We report a 16-year-old woman with chronic right lower quadrant pain and an abdominopelvic mass, initially suspected to be a gastrointestinal stromal tumor. Surgical excision confirmed a caecal desmoid tumor. DTs can mimic malignancies, requiring MRI, histopathology, and immunohistochemistry for diagnosis. Treatment varies, with surgery for symptomatic cases and emerging systemic therapies as alternatives. This article aims to raise awareness of these rare tumors in unexpected locations. It highlights the importance of considering DTs in the differential diagnosis of abdominal masses, particularly in young patients, and emphasizes the need for genetic counseling and multidisciplinary management.

Keywords: Desmoid Tumor, Caecal Desmoid Tumor, Caecal Tumor, Right Hemicolectomy, Colorectal Surgery

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Introduction

Desmoid tumor (DT) is a benign yet locally aggressive tumor with no metastatic potential. It develops from musculoaponeurotic structures and accounts for 0.03% of all malignancies and less than 3% of all soft tissue tumors (Master et al., 2024). DTs are considered mesenchymal neoplasms with infiltrative behavior, also known as aggressive fibromatosis, deep fibromatosis, or musculoaponeurotic fibromatosis. Despite their non-metastasizing nature, DTs can cause significant morbidity and mortality due to local invasion.

There is no standardized treatment for DTs, and their management requires a multidisciplinary approach. The World Health Organization (WHO) classifies them as "clonal fibroblastic proliferations that arise in deep soft tissues, characterized by infiltrative growth and local recurrence but no metastatic capability" (Ganeshan et al., 2019). Recent therapeutic advancements include systemic treatments, with the FDA approving several new drugs for DT management.

The precise cause of DTs remains unclear. Most cases are sporadic, with 85% exhibiting mutations in the CTNNB1 gene encoding β-catenin (Cassidy et al., 2020). The three key mutations identified are 41A, 45F, and 45, with 45F associated with a higher recurrence risk (Nieuwenhuis et al., 2011). Recurrence-free survival rates differ by mutation: 23% for 45F, 57% for 41A, and 65% for patients without mutations.

Desmoid tumors occur with increased frequency in familial adenomatous polyposis (FAP) due to APC gene mutations. FAP-associated DTs often develop at prior surgical sites, making them a significant morbidity factor post-prophylactic colectomy. Other risk factors include pregnancy, high estrogen states, and abdominal wall trauma (Fiore et al., 2016).

Desmoid tumors are rare, with an incidence of 2 to 4 cases per million and accounting for 0.03% of all neoplasms (Timbergen et al., 2018). They most commonly affect women between 15-60 years, peaking at 30-40 years. Approximately 5-10% of cases are associated with FAP syndrome (Desmoid Tumor Working Group, 2020).

Aim of the Article

We report a rare case of a 16-year-old woman presenting with a symptomatic abdomino-pelvic mass, mimicking a caecal gastrointestinal stromal tumor, which was surgically treated with en bloc excision. The pathology confirmed a caecal desmoid tumor. This article aims to raise awareness of these rare tumors that can arise in unexpected locations.

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Case Presentation

A 16-year-old woman with no known comorbidities presented with abdominal pain localized to the right lower quadrant, along with chronic constipation evolving over 8 months. There were no symptoms of intestinal obstruction or gastrointestinal bleeding. She had a normal appetite and no history of weight loss or recent changes in bowel habits. General physical examination was normal. Abdominal examination revealed a 10 cm well-defined mass in the right lower quadrant with limited mobility. Colonoscopy and blood investigations, including carcinoembryonic antigen (CEA) and cancer antigen 125 (CA125), were normal.

On abdominopelvic MRI, there was a 126x117x68 mm midline abdominopelvic mass, with a T1 isosignal, a T2 hypersignal, and heterogeneous enhancement after contrast injection. A provisional diagnosis of a gastrointestinal tumor was made (Figure 1).

The patient was prepared for surgery, and on intraoperative exploration, a 15 cm tumor was found arising from the caecum with invasion of the terminal ileum. The liver, remaining small bowel, large bowel, peritoneum, and ovaries appeared normal. A right hemicolectomy with ileocolic side-to-side anastomosis was performed. Macroscopic examination showed a large encapsulated solid mass measuring 16x12x7 cm (Figure 2).

Histopathology and immunohistochemistry revealed positivity for nuclear ß-catenin and negativity for CD117, smooth muscle actin, and S-100. The diagnosis was confirmed as a desmoid tumor.

The postoperative course was uneventful, and the patient was discharged on postoperative day 6 in stable condition. After one year of follow-up, the patient remains well.

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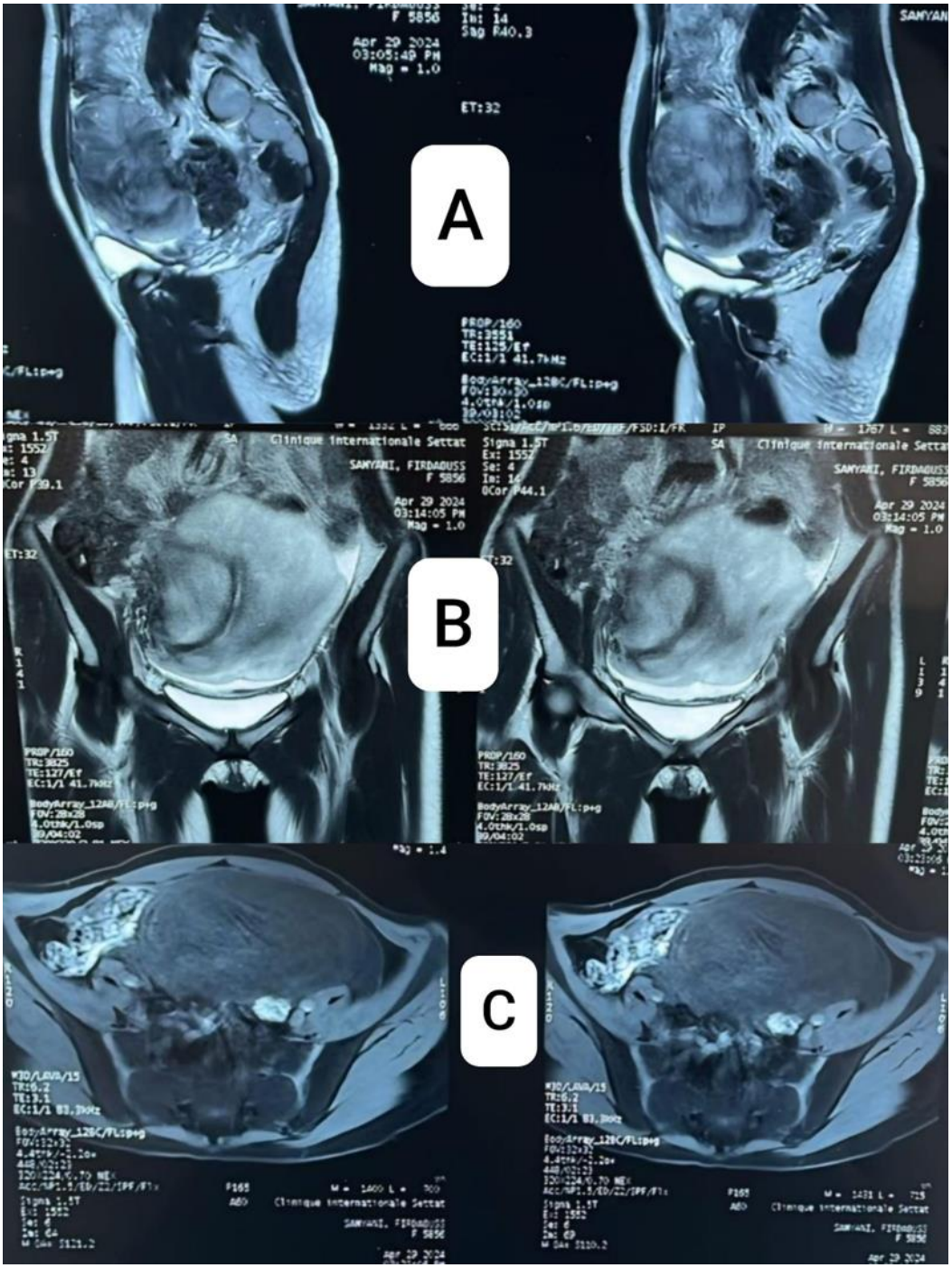

Figure 1: (A) Sagittal MRI image showing a heterogeneous abdominopelvic mass with irregular contours. The tumor is not separable from the caecum. (B) Coronal MRI image showing the tumor invading the small intestine. (C) Axial MRI image showing the caecal tumor with invasion of the small intestine and displacement of ileal loops

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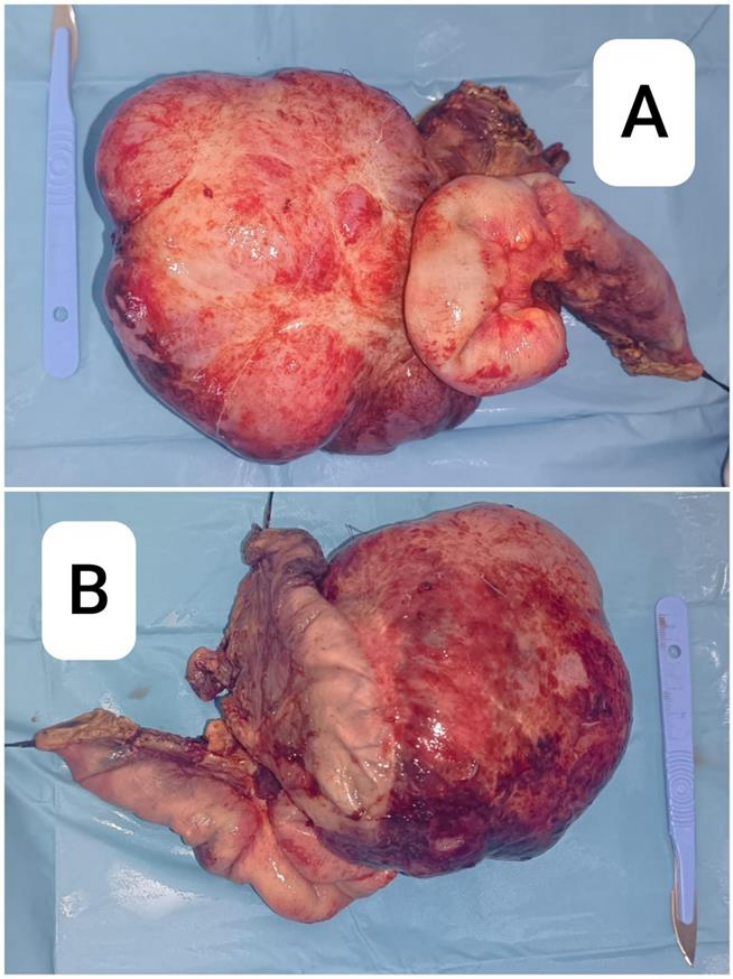

Figure 2: Post-operative specimen (A: Front view, B: Rear view)
Note: The ileocecal appendix was engulfed by the tumor

Discussion

Desmoid tumor (DT) is a rare benign tumor with local aggressiveness. It arises from fibroblasts along fascial planes. DT occurs more frequently in females, especially during or after pregnancy, potentially due to high estrogen states and abdominal trauma (Master et al., 2024). Pregnancy-associated DTs have shown favorable outcomes (Drabbe et al., 2023).

The etiology of DT is unclear; most occur sporadically, with 85% harboring mutations in the CTNNB1 gene, encoding the ß-catenin pathway. The three distinct mutations are 41A, 45F, and 45P. Mutation 45F is associated with a high recurrence risk, with a 5-year recurrence-free survival rate of 23% compared to 57% for 41A and 65% for no mutations (Bektas et al., 2023). DT can also occur in familial adenomatous polyposis (FAP), requiring thorough

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evaluation with colonoscopy and genetic assessment (Pathology Outlines - Fibromatosis-desmoid, n.d.).

Common locations include the retroperitoneum, mesentery, abdominal wall, chest wall, and cervicofacial region (Master et al., 2024). Intra-abdominal DTs often present with chronic abdominal pain and constipation. Complications typically arise from local compression, and symptoms vary by affected region. Imaging with MRI can suggest the diagnosis, showing tumors with spiculated, expansive, and retractile margins infiltrating surrounding tissues. Signal characteristics vary based on the tumor's activity phase, appearing hyperintense in active tumors and hypointense in chronic, fibrous ones (Ben Haj Amor et al., 2020).

Histology confirms the diagnosis, characterized by spindle cell proliferation resembling myofibroblasts amidst collagenous stroma. Features such as hyperchromasia and atypia are typically absent. On immunohistochemistry, DT stains positive for nuclear ß-catenin, vimentin, COX-2, PDGFRB, androgen receptor, and estrogen receptor beta, and negative for desmin, S-100, h-caldesmon, CD34, and c-KIT. Nuclear ß-catenin positivity supports DT diagnosis (Pathology Outlines - Fibromatosis-desmoid, n.d.).

There is no standard treatment for DT, though surgery with negative margins is often recommended for symptomatic patients. Radiation is primarily used for cases with positive surgical margins or when surgery is infeasible (Master et al., 2024). Emerging treatments include immunotherapy and hormone therapy, showing promise for the future.

Conclusion

Intra-abdominal desmoid tumors rarely develop from the digestive tract. This case report highlights the possibility of DT presenting in the gastrointestinal tract. Given the diagnostic and treatment challenges of DT, awareness of genetic counseling and screening colonoscopy, particularly in adolescents and young adults, is essential.

Conflict of Interest: The authors reported no conflict of interest.

Data Availability: All data are included in the content of the paper.

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Ethical Approval

Ethical approval for this study was waived by the Head of the Department of General Surgery at Hassan II University of Casablanca because it is a retrospective case report that does not involve experimental intervention,

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patient randomization, or identifiable personal data. This study was conducted in accordance with the Helsinki Declaration as revised in 2013. As per international or university standards, written ethical approval has been collected and preserved by the authors.

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