International Surgery Journal
Rodrigues ACM et al. Int Surg J. 2023 Apr;10(4):712-714
http://www.ijsurgery.com
pISSN 2349-3305 | eISSN 2349-2902

DOI: https://dx.doi.org/10.18203/2349-2902.isj20230981

Case Report

Desmoid-type fibromatosis of transverse mesocolon

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Received: 13 Februay 2023
Revised: 14 March 2023
Accepted: 18 March 2023

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ABSTRACT

Desmoid tumors represent a rare disease. The recommendations for treatment and approach of these neoplasms are sparse. There are no specific imaging or clinical features which turns the diagnosis difficult. A 53-year-old male patient presented with complaints of peri-umbilical abdominal pain associated with a palpable mass. CT scan revealed a heterogeneous mass with soft tissue density and well-defined contour with approximately 8.5×7×5.3 cm localized in the mesogastric region with apparent origin in the transverse colon. The purposed etiology was of GIST. Colonoscopy was normal. The patient was submitted to surgery which revealed a mass with bright and smooth surface that was in relation with the mid portion of the transverse colon, we performed a right extended hemicolectomy. The postoperative was uneventful. Pathology evaluation revealed a desmoid-type fibromatosis. The accurate etiology of desmoid tumors is still unknown. Desmoid tumors are rare with an annual incidence of 2-4 cases per million people. They typically are slow growing fibrous soft tissue tumors with a benign behavior, however show local aggressiveness with a high ability for recurrence, complete surgical resection with clear margins is crucial.

Keywords: Desmoid-type fibromatosis, Intra-abdominal desmoid tumor, Colon

INTRODUCTION

Desmoid tumors are rare and associated with clinical and radiologic unspecific presentations making preoperative diagnosis difficult and there are few recommendations available for management of these cases.

Desmoid tumor is histologically defined as a fibroblastic monomorphic proliferation of the soft tissues and is classified as intermediate tumors in the WHO 2020 classification. They have a characteristic local aggressiveness being infiltrative and invasive however they have no metastatic ability. The most common site is in the abdominal wall (around 30% of cases), intra-abdominal and mesenteric represent around 20% of cases and around 15-20% are associated with familial adenomatous polyposis. The majority of cases are sporadic, ranging to 90% and linked to a somatic gene activating variant in exon 3 of the CTNNB1 gene. Frequently it manifests as a mass that can sometimes be painful, in rare instances it can be diagnosed after urgent surgery for complications related such as abscess or bowel perforation, intestinal obstruction or ureterohydronefrosis in pelvic locations.[1]

CASE REPORT

A 53-year-old male patient presented with complaints of peri-umbilical abdominal pain associated with a palpable mass, he also refereed periods of constipation. He didn´t had other gastro-intestinal complaints and denied weight loss. Abdominal evaluation revealed a mobile, non-tender

palpable mass in the peri-umbilical region. Laboratory findings were unremarkable. Abdominal CT scan revealed a heterogeneous mass with soft tissue density and well-defined contour with approximately 8.5×7×5.3 cm localized in the mesogastric region with apparent origin in the transverse colon. The mass was near intestinal loops without invading them. It was suggested that it could be a gastrointestinal stromal tumor of the colon. There weren't other findings in the exam. A total colonoscopy was performed without any abnormal findings. The patient was proposed for surgical intervention. Intra-operatively it was identified an oval tumor with bright and smooth surface that was in relation with the mid portion of the transverse colon. We weren´t able to define a safe dissection plane from the mesentery with suspicion of middle colic vessels involvement, so we performed a right extended colectomy (Figure 1 and 2).

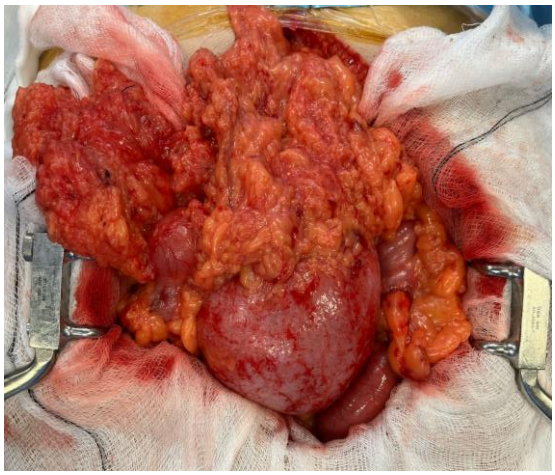

Figure 1: The tumor in its position with bright and homogenous appearance.

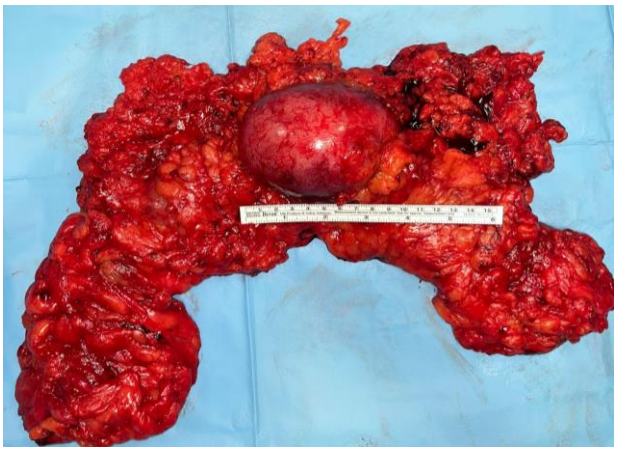

Figure 2: The resection specimen of right colectomy containing the tumor. It can be seen a rule below the lesion.

The post-operative period was uneventful. The pathologic report showed a tumor with 11×7 cm consisting of long fascicles of slender spindle cells, with ovoid nuclei and without cytological atypia or mitosis (Figure 3). The lesion had infiltrative contour with entrapment of mature adipose tissue and delicate vessels (Figure 4). Colonic mucosa was uninvolved. Immunochemistry (Figure 5) revealed: AE1AE3 -, desmin-, S100-, CD117-, DOG1-, CD34-, beta-catenin+(nuclear expression), ciclin D1+, CDK4-, estimated proliferative index (Ki-67) <1%.

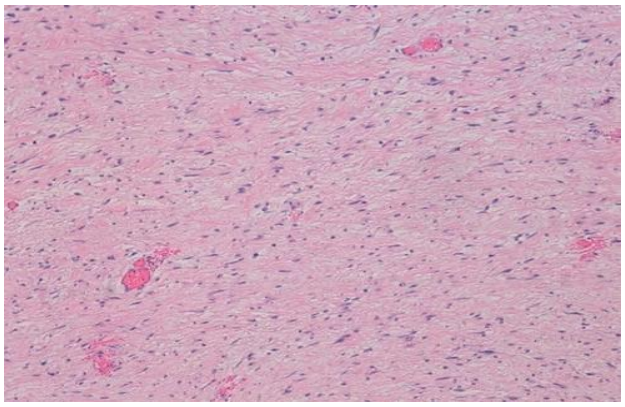

Figure 3: The tumor consists of long fascicles of slender spindle cells, without cytological atypia or mitosis (H and E 100X).

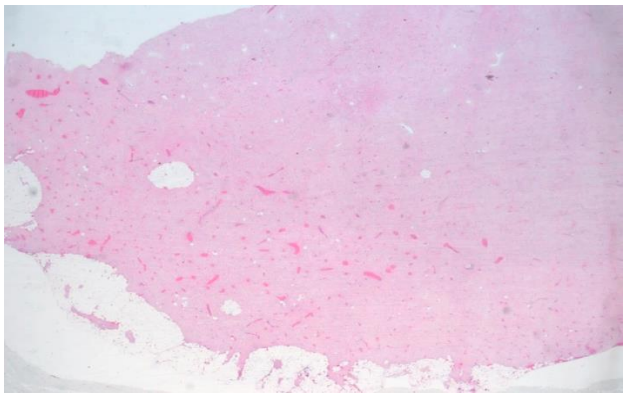

Figure 4: Low power H and E image of the tumor, with peripheral soft tissue infiltration. Presence of delicate vessels.

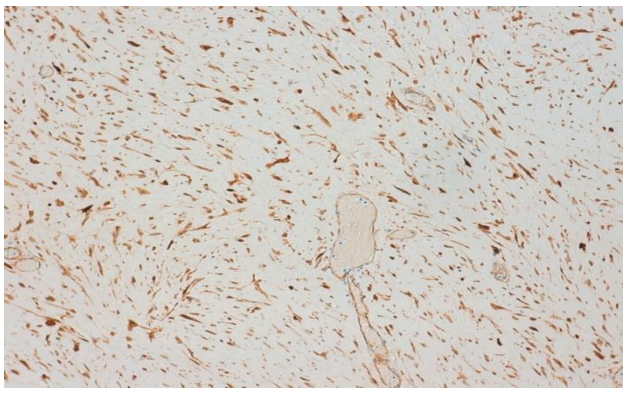

Figure 5: Nuclear β-catenin expression (100X).

The findings were compatible with a diagnosis of desmoid-type fibromatosis localized in the subserosa with clear margins, there were 38 lymph nodes isolated with reactive features. The patient is now under surveillance, without signs of recurrence after 12 months of surgery.

DISCUSSION

The accurate etiology of desmoid tumors is still unknown, however they seem to be associated with abnormalities in genes involved in tissue repair. Desmoid tumors seem to develop when stimulating factors develop together with these gene abnormalities. Female gender, estrogen exposure, trauma, such as laparotomy or abdominal injury, adenomatous polyposis gene abnormalities such as familial adenomatous polyposis and Gardner syndrome all seem to be risk factors for development of desmoid tumors.[2,3]

Desmoid tumors are rare with an annual incidence of 2-4 cases per million people. They typically are slow growing fibrous soft tissue tumors with a benign behavior. Nevertheless they are locally aggressive and have the ability to infiltrate nearby organs, therefore they have a high potential for recurrence, described at 19-77%. Hence, complete surgical resection with clear margins is crucial.[2-4]

At the time of diagnosis, they usually achieve a size of 5.5 cm. The main manifestation is by a mass syndrome that can sometimes be painful. The evolution is unpredictable and within 2 years after the diagnosis about 30-50% may show spontaneous regression, about one third may have progression and the other third may have a stabilization.[1]

In familial adenomatous polyposis cases intra-abdominal location is very common, however in sporadic cases this site is very rare accounting for 5-10%. Mesentery is the most common site for intra-abdominal presentation and usually the diagnosis is delayed because of sparse symptoms and subtle development. One of the diagnostic hypotheses when evaluating these tumors is GIST that in immunochemistry shows positivity to CD117, CD34, DOG1 meanwhile desmoid-type fibromatosis is characterized by positive nuclear staining of beta-catenin and lack of CD34 expression.[3]

Although desmoid tumors have no characteristic features in imaging exams, it is important to evaluate preperatively some elements namely the size of the tumor, relationship with mesentery and retroperitoneal vessels and infiltration of adjacent viscera. When there is uncertainty of diagnosis resection is a valid option.[1-5]

Our patient had a suspicion of GIST on imaging report however we couldn´t obtain a biopsy due to the location of the tumor, and so after a peer discussion and a dialog with the patient we proposed him for surgery.

CONCLUSION

Desmoid tumors are rare and albeit they have a negligible rate of distant dissemination there is an important local aggressive behavior. Differential diagnosis with other types of neoplasms is important and a multidisciplinary team discussion is essential for an adequate treatment.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Rodrigues ACM, Cardoso M, Alves DG, Fernandes S, Caires AG, Sousa MO, et al. Desmoid-type fibromatosis of transverse mesocolon. Int Surg J 2023;10:712-4.