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Case Report

A rare presentation of intraabdominal desmoid tumour

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ABSTRACT

Desmoid tumors are rare deep-seated tumors of mesenchymal origin with 2-4 cases per million. Etiology is multifactorial with variable prognosis. These are highly locally invasive but have no ability to metastasize. A 51-year-old male patient who presented with swelling in the right upper abdomen and heaviness in abdomen. On investigating, there was a mass arising from the anti-mesenteric border of the ileum, caecum and ascending colon. Resection of the tumor with the attached ileum, caecum, ascending colon along with the ileo-colic anastomosis was performed. Post operatively, no chemo-radiotherapy was given and patient was on follow-up since then with no recurrence. Desmoid tumor also commonly known as fibromatosis are large soft tissue tumors of mesenchymal origin are locally aggressive tumors with almost no ability to metastasize. Asymptomatic tumors and small tumors can be managed conservatively with close follow-up. Symptomatic and large tumors are usually managed with surgical resection and radiotherapy. Positive margins lead to higher rates of recurrence. This case, reports the rare presentation of a large intra-abdominal sporadic desmoid tumor arising from the anti-mesenteric border of the bowel. Such cases require multi-disciplinary approach with cautious and meticulous resection to prevent capsular rupture.

Keywords: Desmoid tumor, FAP, Fibromatosis, GIST, Intra-abdominal fibromatosis, Sporadic desmoid tumor

INTRODUCTION

Desmoid tumors are benign rare soft tissue tumors usually associated with FAP/Gardener syndrome, post traumatic status, mostly in woman due to high estrogen levels. Sporadic desmoid tumors are a rare presentation which usually are associated with beta-catenin mutation. Most of the sporadic desmoids are the one's arising from abdominal wall. Most of the intra-abdominal desmoids have their origin from the mesentery involving the vessels or infiltrating the vessels. However, our patient is a case of intra-abdominal sporadic desmoid arising from the bowel is extremely rare. This surgical case report has been reported in line with the SCARE criteria.[1]

CASE REPORT

A 51 years old gentleman presented at surgery OPD with complaint of swelling in the abdomen for last 3 months which was insidious in onset initially of size 6×6 cm, then slowly progressed to present size of 12×10 cm, h/o heaviness in the abdomen and complaint of decreased appetite. Patients had no other complaints. Patients had no prior medical or surgical history. He didn't have any drug allergies. General examination of patient was normal.

On examination, an oval swelling of 15×10 cm swelling was noticed in the right iliac fossa, right lumbar, right hypochondrium and extending into epigastrium. On palpation oval non tender intra-peritoneal swelling with well-defined margins. The swelling was hard in consistency and the mobility of the swelling was restricted in both horizontal and vertical direction. Patient was diagnosed as intra-abdominal soft tissue swelling keeping GIST and Desmoid tumor as differential diagnosis.

All the blood investigations of the patient were within normal range.

On ultrasonography, well-defined heteroechoic mass of size 13×10×13 cm was seen in right iliac fossa with no obvious vascularity.

On CECT, an ill-defined soft tissue mass lesion of approximately size 15×10×13 cm was seen along the terminal ileum and caecum with no evidence of calcification or necrotic components.

The lesion was seen abetting the bowel loop and abdominal wall, right iliac vessels and causing the mass effect on the bowel loops.

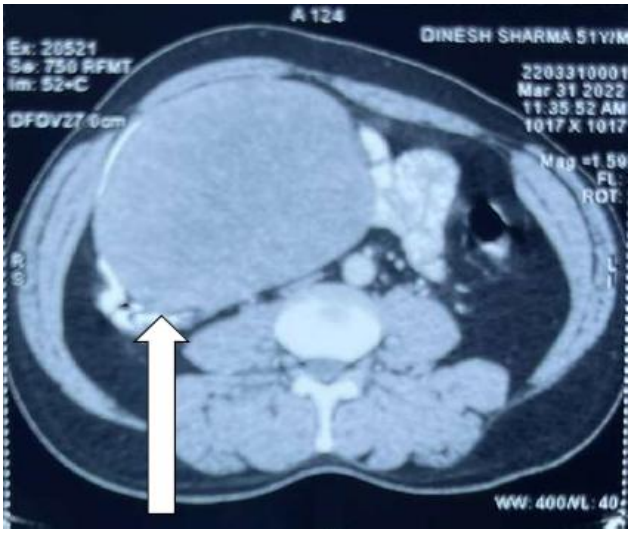

Figure 1: Contrast-enhanced CT showing a large intra-abdominal mass arising from the terminal ileum and caecum.

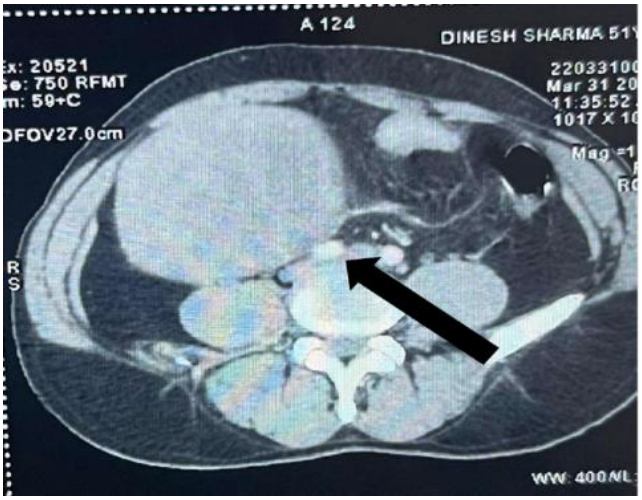

Figure 2: CECT image demonstrating the tumor abutting adjacent iliac vessels and anterior abdominal wall.

On diagnostic laparoscopy an intra-abdominal mobile mass, arising from the anti-mesenteric border of ileum, caecum and ascending colon without involving the vessels, or the peritoneum was seen.

Patient was managed with exploratory laparotomy with resection of the tumor along with terminal ileum, caecum and part of ascending colon. Intestinal resection was done with 10 cm of tumor free margin.

An ileo-ascending colon anastomosis was done to restore the bowel continuity. Post-operative period was un-eventful and patient was discharged on POD 6.

Patient is still in active follow-up with no recurrence or developed any new lesion for the last 4 years.

HPE report-gross pathological specimen examination-16×12×9 cm large growth, arising from the serosa of ileum, caecum, ascending colon with resected margins being tumor free.

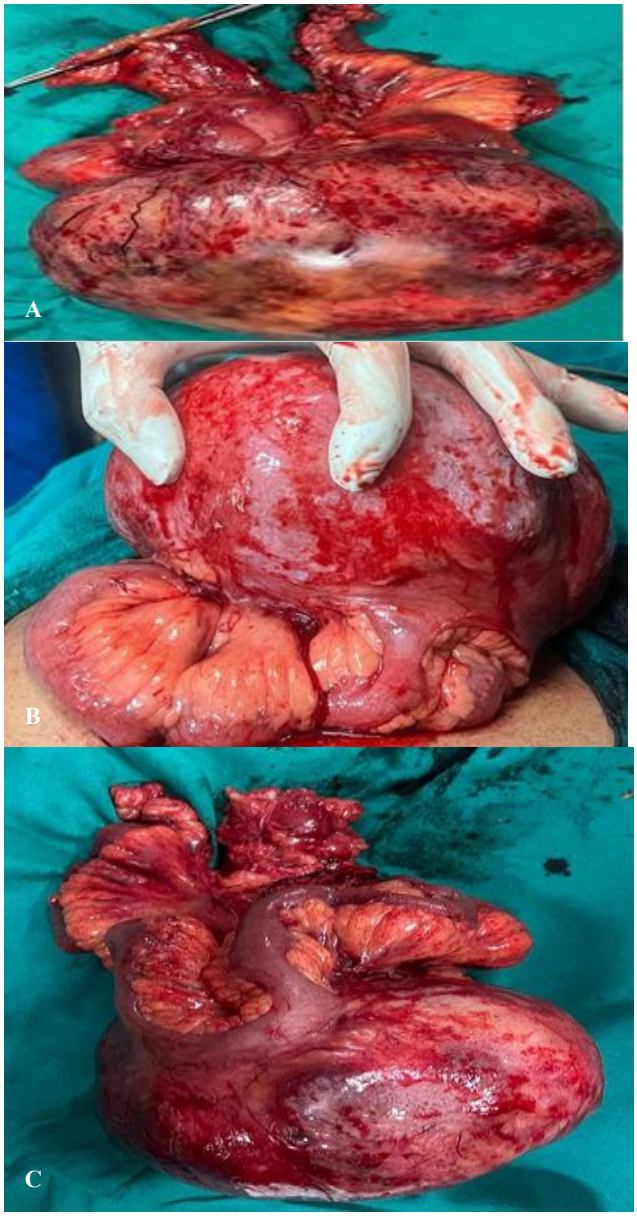

Figure 3 (A-C): Specimens showing desmoid tumor arising from the anti-mesenteric border of the ileum, caecum, ascending colon.

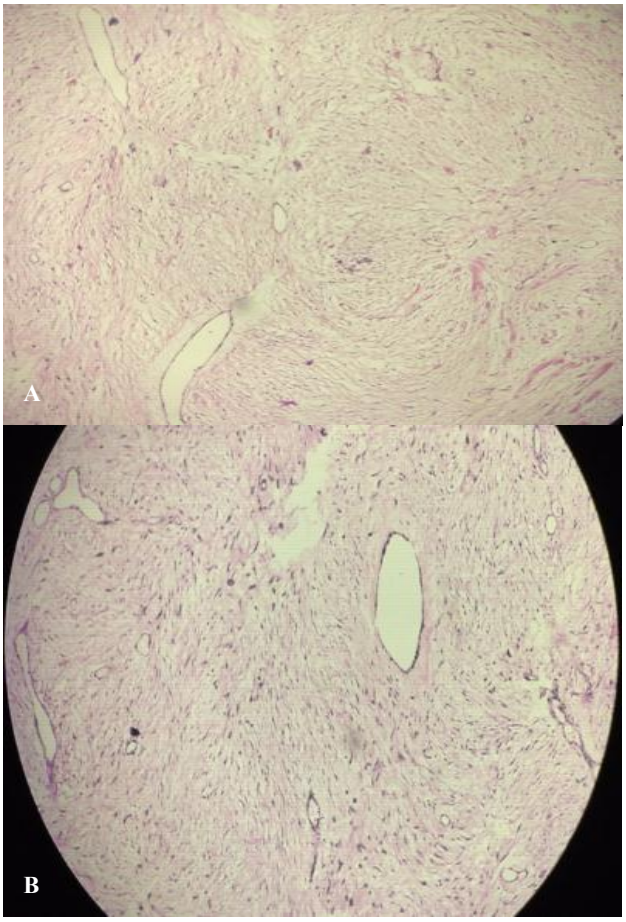

Figure 4 (A and B): Microscopic examination.

On microscopic examination-partially encapsulated tumor, showing fibroblasts and myofibroblasts.

Resected margins tumor free.

On immunohistochemistry Vimentine and Beta Catenino were positive in both cytoplasm and nucleus confirming it as intra-abdominal desmoid tumor.

SMA, CD-117 was negative.

During follow up, patient underwent colonoscopy for colonic polyps to rule out FAP. Colonoscopy showed no intra-luminal abnormalities making the final diagnosis intra-abdominal sporadic desmoid tumor arising from anti-mesenteric border.

DISCUSSION

"Desmoid", the word is derived from the Greek word "desmos" which meant "tendon like or band" described first by MacFarlane in 1832.[2] Desmoid type fibromatosis previously known as aggressive fibromatosis. These are rare slow growing benign deep seated monoclonal myo-fibroblastic mesenchymal tumors. These are highly locally aggressive tumors with high recurrence rate but lack malignant potential. These tumors have their origin from fascial or musculo-aponeurotic stromal elements.[3]

Desmoid tumors occur mostly in fertile women. This may be due to higher levels estrogen and estrogen associated changes after child birth. Desmoid tumors are also associated with trauma. Incidence of desmoid tumors at previous surgery sites is noticed.[4,5] Most of the desmoid tumors occur at abdominal region, but they can occur other places like shoulder girdle, chest wall, inguinal region and intra-abdominal. Desmoid tumors can be of 3 types-spontaneous, FAP/Gardener syndrome related and hereditary/familial. Spontaneous desmoids are rare and usually occur at extremities.[6]

The incidence of the desmoid tumor is 2-5 cases per million people per year. They account for almost 0.03% of all neoplasms, and 3% of all-soft tissue tumors.[3-5]

As per the literature, etiology is multifactorial, yet the established genetic factors associated with the desmoid tumor are APC gene mutation related FAP, beta catenin gene (CTNNB1) mutation. According to the lazar and co, 98% of the sporadic desmoid tumors occur in patients with CTNNB1 mutation.[7]

The calculated incidence of the sporadic intra-abdominal desmoid tumors that occur in general population is about 3.42 per million person years, while that in FAP patients have 2784 per million person years. The incidence in FAP patients is approximately 850 times that of the general population. Spontaneous desmoids are rare and usually occur at extremities. Presence of APC mutation significantly increases the risk of desmoid tumor by 800 fold in FAP patients. About 10-15% of the FAP patients are prone to have intra-abdominal desmoid tumors.[3-6] The demonstration of mutations in the Wnt-APC, beta-catenin pathway implicates beta-catenin as the key factor in the pathogenesis of aggressive fibromatosis. Regardless of the mutations, all the desmoid tumors had stable production of the beta-catenin in the cytoplasm and nucleus of the tumor cells.[7,8]

Most of the patients with desmoid tumors are asymptomatic. Rarely they might cause symptoms like ureteric obstruction, hydronephrosis, intestinal obstruction, fistula. Life threatening complications like sepsis, hemorrhage, peritonitis may occur, requiring emergency laparotomy.[9,10]

Contrast enhanced CT is the main modality of non-invasive investigation that is used for the assessment of intra-abdominal soft tissue tumors. In case of trans-abdominal and extra-abdominal desmoid tumors where local invasion is suspected, MRI gives better differentiation. Differentiating GIST from IAF (Intra-abdominal fibromatosis) is very important for planning the management protocol as GIST have higher potential for malignancy and even metastasis, whereas metastasis

is almost never expected in IAF patients. Unless indicated IAF patients are not taken up for surgery.

As per the study conducted by Zhu et al they have described 8 feature to differentiate GIST from desmoid. All these 8 features on CT can be used to distinguish a GIST from IAF. If any 3 of these 8 features on CT scan are combined the specificity in distinguishing IAF (Intra-abdominal fibromatosis) from GIST using these criteria 89.5%.[11]

CT imaging features

Extra-gastrointestinal location, ovoid or irregular lesion contour, homogeneous contrast enhancement, absence of intra-lesional necrosis, degree of enhancement during arterial phase ≤40.5 HU, degree of enhancement during portal venous phase ≤46.5 HU, lesion-to-aortic attenuation ratio during arterial phase ≤0.315 and lesion-to-aortic attenuation ratio during portal venous phase ≤0.525.

Diagnosis of desmoid tumors is mainly by the histopathological examination. On histopathological examination, there would be proliferating stellate to spindle cells. Positive for beta-catenin is seen in almost all cases in both the cytoplasm and nucleus. Desmoid tumor cells are also strongly stained with vimentine. But this is not specific for desmoid tumor.[5,8,9] Most GISTs diffusely and strongly express KIT (CD-117). Tumors other than GISTs developing in the GI tract are usually negative for KIT, but some tumors such as PEComas **(**Perivascular epithelioid cell tumor) and melanomas might be positive for KIT.[12]

Staging

Stage I-Asymptomatic/not growing

Management of such type of tumors would be simple observation or non-toxic (‘‘prophylactic’’) therapy (i.e., NSAID). Resection of the tumor without resecting significant portion of bowel is done, if the desmoid tumors found incidentally.[13]

Stage II-Symptomatic and<10 cm in maximum diameter/not growing

Resection of the tumor is the method of choice. If unresectable, combination therapy (i.e., tamoxifen or raloxifene +NSAID).

Stage III-Symptomatic and 10-20 cm in size, or asymptomatic and slowly growing

Symptomatic tumors include those cause intestinal or ureteric obstruction.

Slowly increasing tumors are those which grow less than 50% in maximum diameter in 6 months. Active treatment is required. It includes NSAIDS, tamoxifen, raloxifene, and vinblastine/methotrexate. Antisacroma therapy like Adriamycin or dacarbazine can be given if the non-toxic therapy is not working.

Stage IV-Symptomatic/>20 cm/rapid growth/complicated tumors

Urgent interventions which include surgery or antisarcoma chemotherapy and radiation therapy.

Management strategies

Simple observation

For asymptomatic patients, patients with minimal symptoms and small desmoid tumors not invading any structures.[13,14]

Pharmacological therapies

Non-steroidal anti-inflammatory drugs (NSAIDS), antiestrogen (tamoxifen or teremifene), interferon-alpha and cytotoxic chemotherapy: Preferred in following cases-If non-toxic management is failed and inoperable cases.

Surgery

Radical resections are the gold standard management of desmoid tumors arising from the abdominal wall. In these patients, abdominal wall reconstruction may be needed. Intra-abdominal Desmoid tumors with their origin from the mesentery unless symptomatic/ growing in size/more than 10 cm should be managed conservatively. Resection of the tumors in such cases involves resection of large portions of bowel. Hence such cases should be approached with utmost caution.

Radiation therapy

Limited role for the management of abdominal wall or intra-abdominal desmoid tumors, due to the high risk of complications.

Newer therapeutic approaches

Their therapeutic value is currently under investigation-Radiofrequency ablation, cryoablation, high intensity focused ultrasound, percutaneous chemical ablation and gene transfer therapy.

CONCLUSION

Desmoid tumors are rare, soft tissue tumors, that can occur anywhere in the body. Because of the rarity of cases, one needs high level of suspicion during assessment and investigation. Unless symptomatic, most of the desmoids are treated conservatively. Surgery is mainstay of management in symptomatic cases. All the

patients with desmoid tumors should get evaluated for FAP/Gardener syndrome, as they are at high risk of developing desmoid tumors.

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