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Case Report Open Access

A Rare Case of Desmoid Tumour in Upper Abdomen: Case Report

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Abstract

Recently we dealt with a case of a female patient. The patient who suffering from abdomen pain from the past 60 days, she was dull aching in nature aggravated on taking meals associated with early satiety and complaints of nausea and fever since 2 months intermittent relieved on taking medication associated with myalgia and weight loss 4 kg's in 2 months. No complaints of vomiting, diarrhea, constipation, blood in vomit, blood in stool, melena. No history of DM, hypertension, tuberculosis, blood transfusion, jaundice or any surgery. Sleep pattern, bowel and bladder habits are regular and undisturbed. Earlier in the patient family there is no significant history.

Keywords: Desmoids; Monitoring; Awareness; Surgery; Ventilation; Circulation

Introduction

Abdominal desmoids are rare fibroblastic tumors. Though these tumors do not display metastatic potential, their locally aggressive nature can cause severe outcomes [1].

Desmoids tumors are rare soft tissue tumors arising from connective tissue that provides strength to muscle, ligaments and bones. Desmoids tumors are rare accounting for about 0.03% of all tumors. The incidence of desmoids tumor is 2-4 per million per year. They are common in 10-40 years of age group and common in females after childbirth with female to male ratio of 2:1.They can be solitary or multiple. Desmoids tumor can be abdominal desmoids tumor (arising from abdominal wall), intra-abdominal desmoids tumor (arising from structures connecting abdominal organs) and extra-abdominal desmoids tumor(occurring in shoulder, upper arms and upper legs). Desmoids tumors are fibrous much like scar tissue. They are not considered malignant but they have tendency to invade the surrounding tissue aggressively hence they are difficult to remove surgically.

Desmoid tumors can be classified as extra-abdominal and abdominal. Abdominal desmoid tumors are either superficial or intra-abdominal [2]. Desmoid tumors are rare, benign, fibromatous lesions that are the result of abnormal proliferation of myofibroblasts [3].

Etiology

Desmoids tumors frequently occur in people with an inherited form of colon cancer called familial adenomatous polyposis. Desmoids tumors that are not part of an inherited form are called sporadic desmoids tumors. Mutation in *CTNNB1* gene or APC gene causes desmoids tumors. Mutation of *CTNNB1* gene accounts for about 85% of all sporadic desmoids tumor. Both these genes are involved in important cell signaling pathways that controls proliferation and differentiation of cells. The *CTNNB1* gene codes for beta-catenin protein that interacts with other protein to regulate expression of genes involved in proliferation and differentiation of cells. Mutation in *CTNNB1* gene leads to abnormally stable beta-catenin that accumulates in cells and acts in an uncontrolled way. The protein produced from APC gene binds with beta-catenin and signals for it to be broken down. Mutation in APC gene causes abnormally short protein that is unable to interact with beta-catenin which is not broken down and results in accumulation and uncontrolled cellular proliferation and differentiation leading to formation of desmoids tumors (Figure 1).

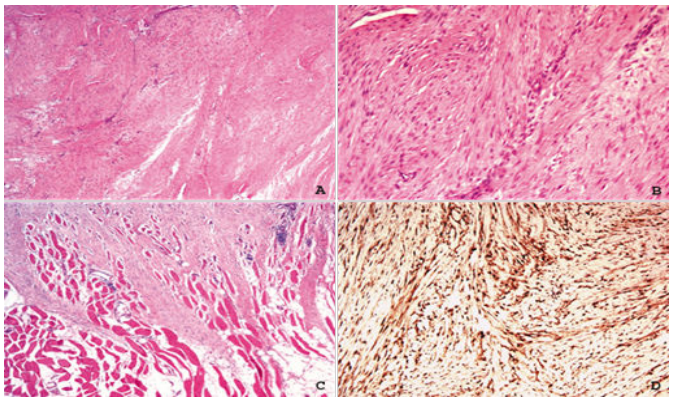

Figure 1: H and E staining of desmoids tumors.

Pathology

Retro peritoneum benign intra-abdominal and retroperitoneal tumors are uncommon and have diverse etiologies, including but not limited to duplication and mesenteric cysts, lymphoceles, lymphangiomas, lipomas, and peripheral nerve sheath tumors, with mesenteric fibromatosis and sclerosing mesenteritis comprising the prevailing diagnoses [4].

Grossly, desmoids tumors are firm and display white and whorled cut surface which may be poorly circumscribed. Microscopically, the lesion is proliferation of bland appearing spindle-shaped fibroblast in a collagenous stromal with infiltrative border. Mitosis is rare and no atypia is seen. Keloid like collagen or extensive hyalinization may be

present. Desmoids tumors stain positive for vimentin and variably positive for smooth muscle actin or other muscle specific markers by immunohistochemistry.

Nuclear staining for beta-catenin is positive in approx. 80% of sporadic desmoids tumors. Therefore, beta-catenin may be extremely helpful in distinguishing desmoids tumors from other spindle cell tumor.

CTNNB1 mutations and loss in chr6 are the causative mutations of desmoid tumors. The expression of IFI6 is the most significant marker to stratify the prognosis. High-risk patients are potentially good candidates for wait-and-see management [5].

Case Report

A 29-year-old Hindu female presented to the OPD with complaints of pain in upper abdomen since 2 months which was dull aching in nature aggravated on taking meals associated with early satiety and complaints of nausea and fever since 2 months intermittent relieved on taking medication associated with myalgia and weight loss 4 kgs in 2 months.

No complaints of vomiting, diarrhea, constipation, blood in vomit, blood in stool, melena. No history of DM, hypertension, tuberculosis, blood transfusion, jaundice or any surgery. Sleep pattern, bowel and bladder habits are regular and undisturbed. No significant family history.

Examination

General examination, Pulse: 78 beats/minute, Blood pressure: 130/80 mmHg, Respiratory rate: 18/minute, Temperature: normal.

Systemic examination: per abdominal examination; Soft and non-tender with a palpable lump of approx. 20 × 15cm size with well-defined margins in the umbilical, epigastria and hypogastria region with firm consistency and mild tenderness upon palpating the lump.

Investigations

Routine lab investigations including CBC, coagulation profile and liver function tests and renal function tests and blood grouping and typing. Chest and abdominal x-rays.

Ultrasonography s/o 15 × 20 cm sized mass in abdomen possibility of GIST more likely.

CECT abdomen with pelvis was done which was suggestive of a 19 × 15 × 14cm sized well defined heterogeneously enhancing soft tissue density lesion with a possibility of large neoplastic lesion arising from proximal small bowel probably GIST (Figure 2).

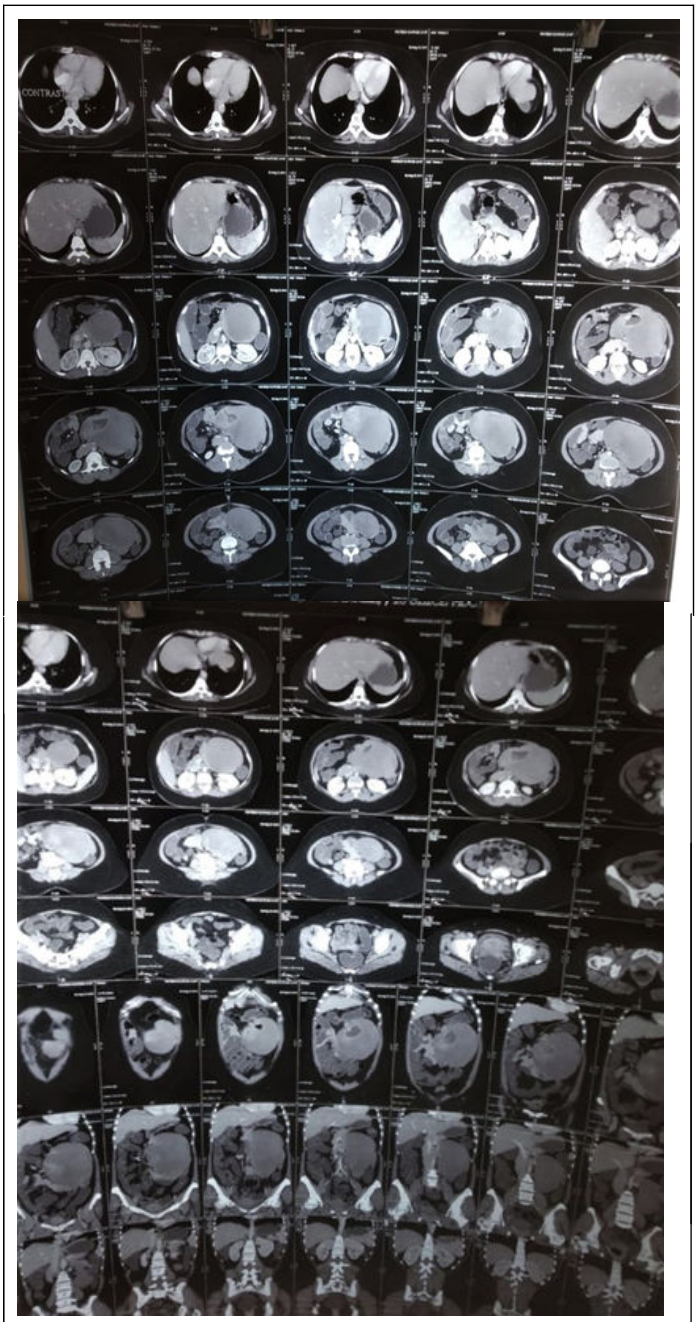

Figure 2: Contrast enhanced computerized tomography images.

Management

The main aim of management in this patient was the excision of the tumor with negative surgical margins. The procedure opted was, exploratory laparotomy with excision of the tumor. Bowel preparation was done prior to the operation and patient was on liquid diet for two days before the date of surgery. Pre-operative anesthetic assessment was done and patient was declared fit for the procedure.

Operative note

Intraoperative, approx. 20 × 15 × 15 cm sized tumor was found arising from mesentery of proximal small bowel at duodenojejunal junction with approx. 60 cm length of jejunum adherent to tumor. Approx. 10 cm distal to jejunely loop(which was adherent to tumor)bowel was transected and mesentery sequentially ligated to mobilize the tumor.

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Tumor pedicle identified and clamps were applied. Tumor resected along with jejunum at duodenojejunal junction and pedicle of the tumor transfixed using vicryl no.1 and doubly ligated using vicryl no.1. Duodenum mobilized by kocherisation and distal end of duodenum was closed by silk 2-0 in two layers. Similarly jejunum was closed by silk 2-0 in two layers. Closed end of jejunum was brought through a window in transverse mesocolon and side to side duodenojejunal anastomoses was done by silk 2-0 in double layer.

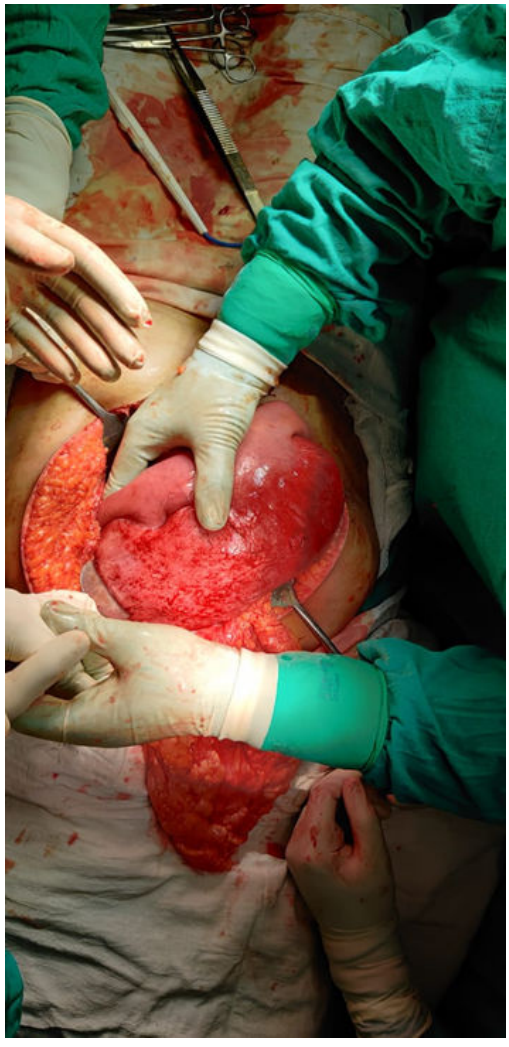

Figure 3: Tumor in Situ.

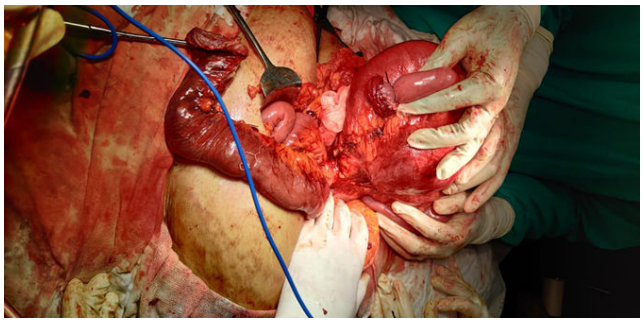

Figure 4: Tumor mobilized.

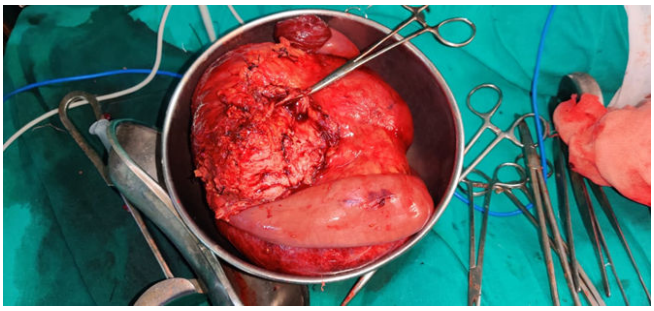

Figure 5: Tumor specimen.

Biopsy report

Biopsy report was suggestive of spindle cell tumor arising from serosa and subserosal fatty tissue which was abutting muscular is propriety of small intestine having moderate cellularity and fascicular growth pattern. No evidence of necrosis or increased mitotic activity was there. Sections from surgical margins were free from tumor. Section from the specimen of jejunum does not show evidence of tumor infiltration. Differentials given in biopsy report were:

- GIST(Gastrointestinal Stromal Tumors)
- Benign nerve sheath tumour
- Inflammatory my fibroblastic tumour
- Mesenteric fibromatosis
- Desmoid tumors are benign locally infiltrative tumors that do not metastasize.
- Pancreatic desmoid tumors are extremely rare, and only 27 cases have been reported.
- Optimal treatment consists of surgical resection with wide margins.
- Laparoscopic pancreatectomy is a surgical option for pancreatic desmoid tumors [6].

Immunohistochemistry was advised for confirmation of the diagnosis. Slide review at GCRI(The Gujarat Cancer Research Institute) was suggestive of spindle cell tumor of small bowel likely gastrointestinal stromal tumor without evidence of necrosis or mitosis. Immunohistochemistry done at GCRI was suggestive of desmoids tumor.

Discussion

Desmoids tumors are slowly growing benign tumors having tendency to invade surrounding tissue aggressively. The differential diagnosis of desmoids tumor is broad with fibroblastic sarcoma on one extreme and reactive fibroblastic and my fibroblastic processes such as nodular fasciitis and hypertrophic scars and keloid on the other. Differential diagnosis of intra-abdominal desmoids tumor includes gastrointestinal stromal tumor, inflammatory my fibroblastic tumor,

sclerosing me enteritis and retroperitoneal fibrosis. To differentiate desmoids tumor from these entities requires immunohistochemistry in which nucleus of spindle cells stain positive for beta-catenin in cases of desmoids tumor. Treatment options include adequate surgical resection with negative surgical margins except when surgery is mutilating and associated with considerable function loss or major morbidity. In cases of positive surgical margins, postoperative radiotherapy alone or in conjunction with systemic therapy in the form of anti-hormonal therapy/NSAIDS can also be considered to prevent the recurrence.

Conclusion

In conclusion, the discoveries made during our study can be easily applied in a clinical setting. Our data emphasize the value of profiling for the prediction of a patient's prognosis after a surgical operation. We believe that our examination during the treatment analyses highlight the importance of precise tumour profiling in the provision of the best possible care to patients with DT. A desmoid tumor is a fibrous soft tissue tumor arising in the fascia and musculoaponeurotic tissues demonstrated that desmoid tumors are characterized by abnormal proliferation of fibroblasts, infiltrative growth pattern, lack of malignant findings, absence of metastases, presence of collagen fibers in intercellular space, and local recurrence. Desmoid tumors can be found in any of the fibrous connective tissues throughout the body

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