

A Case Report: A Rare Case of Desmoid Tumor.

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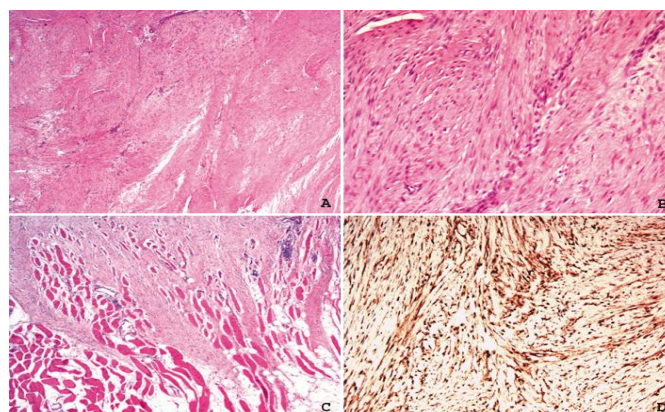
I. Introduction

Desmoid tumors are rare soft tissue tumors arising from connective tissue that provides strength to muscle, ligaments and bones. Desmoid tumors are rare accounting for about 0.03% of all tumors. The incidence of desmoid 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. Desmoid tumor can be abdominal desmoid tumor (arising from abdominal wall), intra-abdominal desmoid tumor (arising from structures connecting abdominal organs) and extra-abdominal desmoid tumor (occurring in shoulder, upper arms and upper legs). Desmoid 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.

II. Etiology

Desmoid tumors frequently occur in people with an inherited form of colon cancer called familial adenomatous polyposis. Desmoid tumors that are not part of an inherited form are called sporadic desmoid tumors. Mutation in CTNNB1 gene or APC gene causes desmoid tumors. Mutation of CTNNB1 gene accounts for about 85% of all sporadic desmoid 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 accumulate 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 desmoid tumors.

H and E staining of desmoid tumors



PATHOLOGY

Grossly, desmoid 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 stroma with infiltrative border. Mitosis are rare and no atypia is seen. Keloid like collagen or extensive hyalinization may be present. Desmoid 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 desmoid tumors. Therefore, beta-catenin may be extremely helpful in distinguishing desmoid tumors from other spindle cell tumor.

III. Case Reort

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, melaena.

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. 20x15cm size with well defined margins in the umbilical, epigastric and hypogastric region with firm consistency and mild tenderness upon palpating the lump.

Investigations

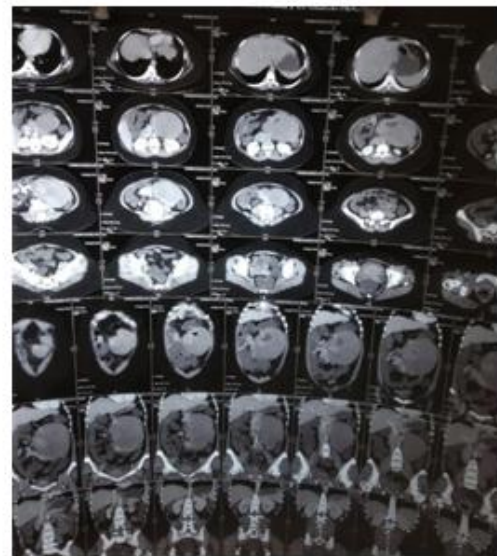
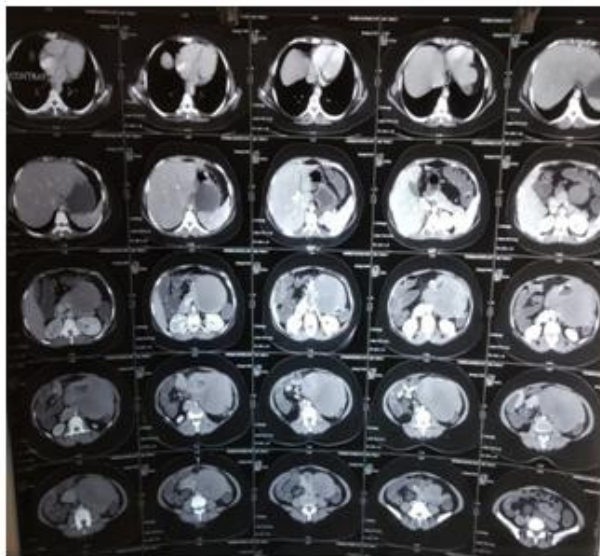
Routine lab investigations including CBC, coagulation profile, liver function tests and renal function tests and blood grouping and typing. Chest and abdominal x-rays.

Ultrasonography s/o 15x20 cm sized mass in abdomen possibility of GIST more likely.

USG guided FNAC was suggestive of "benign spindle cell tumor".

CECT abdomen with pelvis was done which was suggestive of a 19x15x14cm sized well defined heterogeneously enhancing soft tissue density lesion with a possibility of large neoplastic lesion arising from proximal small bowel probably GIST.

CECT 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

Intraoperatively, 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 jejunal loop(which was adherent to tumor)bowel was transected and mesentery sequentially ligated to mobilize the tumor. 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 mobilised 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.

Intraoperative images

figure 1; tumor insitu



figure 2; tumor mobilised

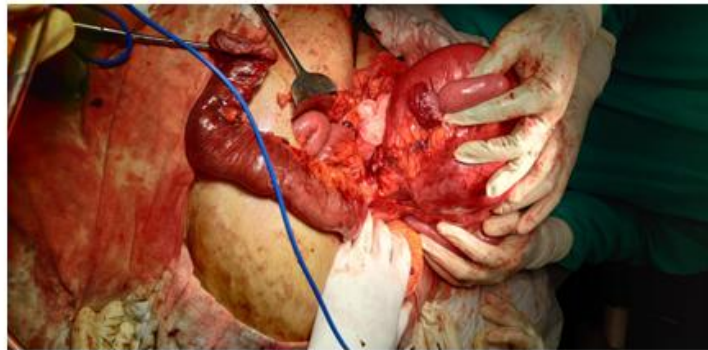


Figure 3; tumor specimen



Biopsy

Biopsy report was suggestive of spindle cell tumor arising from serosa and subserosal fatty tissue which was abutting muscularis propria 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:-(i)GIST (ii)Benign nerve sheath tumor (iii)Inflammatory myofibroblastic tumor (iv)Mesenteric fibromatosis. 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 desmoid tumor.

IV. Discussion

Desmoid tumors are slowly growing benign tumors having tendency to invade surrounding tissue aggressively.The differential diagnosis of desmoid tumor is broad with fibroblastic sarcoma on one extreme and reactive fibroblastic and myofibroblastic processes such as nodular fasciitis and hypertrophic scars and keloid on the other.Differential diagnosis of intra-abdominal desmoid tumor includes gastrointestinal stromal tumor,inflammatorymyofibroblastic tumor,sclerosing mesenteritis and retroperitoneal fibrosis.To differentiate desmoid tumor from these entities requires immunohistochemistry in which nucleus of spindle cells stain positive for beta-catenin in cases of desmoid 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.

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