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DESMOID TUMOR OF ANTERIOR ABDOMINAL WALL: A CASE REPORT AND REVIEW OF RECENT MANAGEMENT STRATEGIES

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Abstract: Desmoid tumor is a slow growing neoplasm with aggressive infiltration of adjacent tissue with very rare metastatic potential. We report a 35 years old female patient with desmoid tumor of the anterior abdominal wall who underwent primary resection followed by mesh pasty. Patient had a past history of abdominal surgery in form of caesarean section, and after 5 years of surgery, she presented with a lump over right hypochondrium since last one year. Patient was assessed before surgery by clinical evaluation, abdominal ultrasound, computerized axial tomography scanning (CT scan), magnetic resonance imaging (MRI), and histopathology and immunohistochemistry report.Patient was operated with wide local excision of rectus abdominis muscle mass with meshplasty under general anaesthesia. After all, histology in this case revealed a desmoid tumor with negative β-Catenin report of operated tissue. In view of the negative surgical margins, patient has been put on regular follow up.

Introduction

Desmoid tumors are benign myofibroblastic neoplasms usually originating from the muscle aponeurosis and classified as aggressive fibromatoses[1]. They constitute 3% of all soft tissue tumors and 0.03% of all neoplasms[2]. Despite their aggressive local infiltration, desmoid tumors usually lack a metastatic potential[3]. However, because of this local infiltration and compression of surrounding structures, a high recurrence rate exists and in anatomic locations with restricted access to surgical resection, desmoid tumors can lead to significant morbidity and mortality. In patients with familial adenomatous polyposis (FAP), undergoing prophylactic colectomy, desmoid tumors are the leading cause of morbidity than colon cancer[4].

Molecular studies demonstrated desmoid tumors in FAP as clonal neoplasms arising from germline mutation or changes in the adenomatous polyposis coli (APC) alleles[5, 6,7]. Cytogenic data has verified clonal chromosome aberrations in deep-seated sporadic extra-abdominal fibromatoses and lesions of the abdominal wall and therefore provide additional evidence for the neoplastic nature of these lesions[8].

The locations of desmoid tumor may vary from abdominal wall of young pregnant females to intra-abdominal mesenteric masses to large extremity masses in males and females. In sporadic cases, they occur in localizations of trauma, and scars or irradiation. The therapeutic management of these tumors is still controversial[9].

Case Report

A 35-year-old female patient reported with a history of abdominal surgery in the form of caesarean section in 2010, and after 4 years, in the year 2014, she presented with a history of lump over right hypochondrium for 1 year. On clinical examination, a painless mass was found without any fixity to surrounding bony structures.

On investigation, blood parameters were within normal range and tumour markers were negative, while ultrasonography of abdomen suggested an approximately 33 x 19 x 23 mm size of hyperechoic lesion with no internal vascularity noted in right rectus muscle suggestive of benign lesions.

On further evaluation, MRI was done to localize the tumor and to exclude metastasis **(Figure 1).** MRI of chest, abdomen and pelvis showed a bulky lesion in epigastric region on right rectus abdominis muscle and reveals ill defined hypo dense lesion measuring 25 x 16 x 20 mm. Moreover, no evidence on intra abdominal extension or obvious bony pathology was detected, which was suggestive of muscle pathology.

The MRI finding revealed tumor's hypo-intensity on T1 and demonstrates variable signal intensity on T2 weighted imaging, depending on the accumulation of mucoid structures **(Figure 2).**

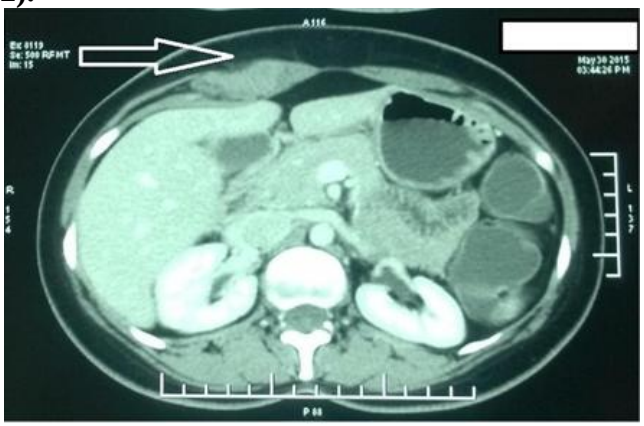

Figure 1 - MRI demonstrates the desmoid tumor originating from the rectus abdominis muscle and internal oblique muscle fascia with inhomogeneous formation.

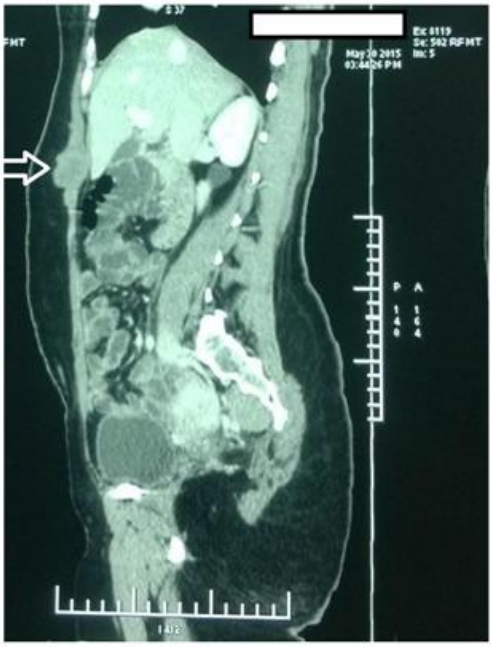

Figure 2 MRI shows the tumor's location embedded in the adjacent musculature. Arrow shows a hypo intense part of the tumour indicating the high mucoid proportion.

Patient underwent wide local excision of rectus abdominis muscle mass with meshplasty under general anesthesia. The postoperative course was uneventful and the patient was discharged on the eighth postoperative day.

Histopathological examination of specimen of excision biopsy

Histopathological report of operated soft tissue mass had shown following findings:

On macroscopic examination, surgical specimen was creamish brown soft tissue mass like structure, with about 80 x 60 x 20 mm size with external surface being irregular white, homogenous about 35 x 17 x 10 mm with infiltrating margin. No necrosis or haemorrhagic area was noted on gross examination.

On microscopic examination of the resected specimen, a **Fibromatosis** of abdominal wall, in other word it was a **Desmoid Tumour**.

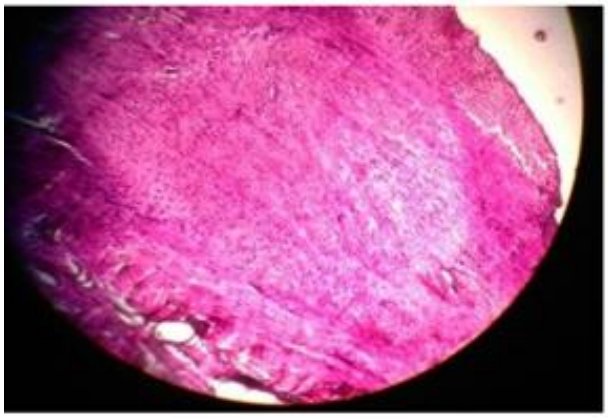

Figure 3. Microscopic examination (10x) showing layers of tumor cells with pale eosinophilic cytoplasm and nuclei and sporadic mitoses characterize the histological picture.

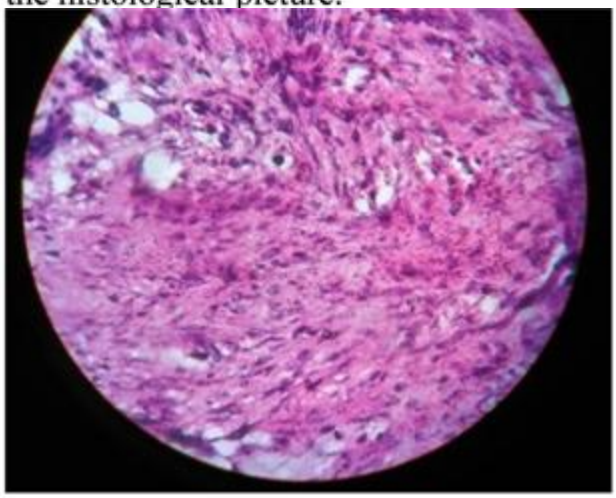

Figure 4. Microscopic examination (40x) showing layers of tumor cells with pale eosinophilic cytoplasm and nuclei and sporadic mitoses characterize the histological picture.

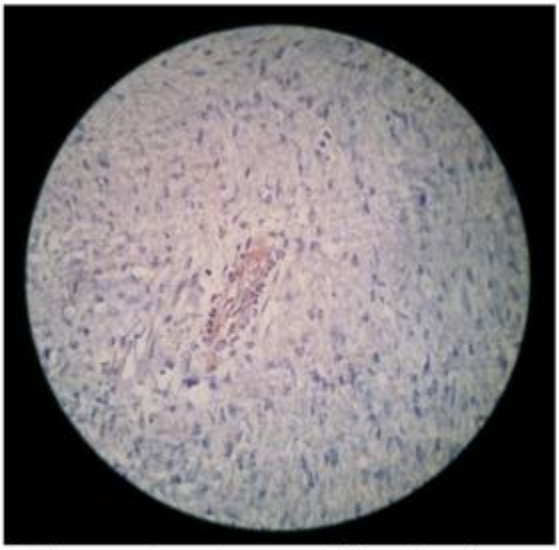

Figure 5. Immunohistochemistry report showing negative β - Catenin staining.

As shown in **figure 5,** in this patient, Immunohistochemistry report of **β-Catenin** was negative, and according to previous studies, nuclear staining of **β-Catenin** is supportive, but not definitive for the diagnosis of desmoid fibromatosis[10]. Moreover, negative results of such an assay would not preclude the diagnosis of desmoid. However, given that most desmoids harbour *CTNNB1* gene mutations, a positive result for such mutation would serve to confirm a diagnosis of desmoid; other benign lesions included in the differential diagnosis are not known to have *CTNNB1* gene mutations nor they show nuclear expression of β-Catenin on Immunohistochemistry[11].

Discussion

Desmoid tumours are benign deep fibromatoses, which originates from fascia and muscle aponeurosis with an infiltrating growth[12]. They are primary located abdominally or intraabdominally, whereas only sporadic cases describe a localisation within the thorax wall[13] or retroperitoneally[14]. Desmoid tumor is generally associated with female gender, FAP[15] and occasionally with surgical trauma[16]. It has shown a higher prevalence in women who experienced caesarean section during pregnancy[17]. Depending on the tumors size, on

the therapy and negative resection margins, recurrence occurs in up to 45%[18].

Histology is the only evidentiary method which confirms diagnosis by showing long fascicles of spindle cells of variable cell-density with few mitoses and absence of atypical nucleus-separations with diffuse cell infiltration of adjacent tissue structures[19].

Surgery is the primary treatment for resectable desmoid tumors. Some studies have reported margin status as independent prognostic factor of recurrence. Other studies have failed to provide any clear association between resection margin status and risk of recurrence[17].

So in case of symptomatic patient with resectable tumor, wide excision should be done. The effectiveness and indication of adjuvant radiation is not proven yet. In a comparative analysis, a significantly better local recurrence control was described with radiation and combined surgical resection in comparison to resection only[17,24]. Other studies have shown that a tumor can progress after radiation therapy and it can show higher local recurrence rate[20,21].

If resection margins are negative (R0), patient should be kept on observation. If resection margins are microscopically positive (R1), observation or re- resection may be done. In case of macroscopically positive margins (R2), various options are; definitive radiotherapy, resection plus radiotherapy, systemic treatment, radical surgery or observation etc[24].

On the other hand, for unresectable desmoid tumor, definitive radiotherapy (54-58 Gy in absence of prior radiotherapy, only in desmoids tumors of extremities, superficial trunk and head & neck regions), systemic treatment and observation are the options. Radiotherapy is usually not given in intra-abdominal and retroperitoneal desmoid tumors.

Recent NCCN guideline (version 1.2015) has included NSAIDS like sulindac or celecoxib, hormonal or biological agents (tamoxifen, toremifine, or low dose interferon), chemotherapy (methotrexate and vinblastine or doxorubicin based regimens), and tyrosine kinase inhibitors (imatinib or sorafenib) are used as systemic treatment for advanced and unresectable desmoid tumors.

Moreover, anti-inflammatory treatment, hormone-therapy and chemotherapy have not been proven yet to be curative. These therapies are limited to patients, in whom resection is not possible because of a widespread tumour infiltration[22,23,24].

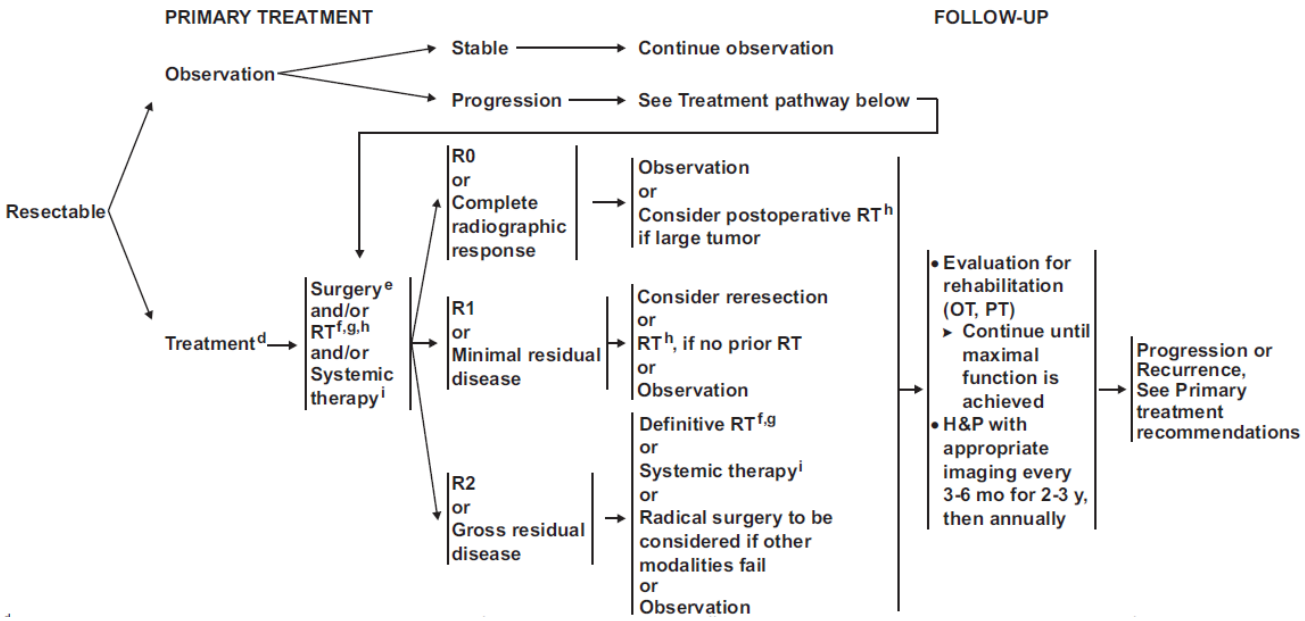

Figure 6 NCCN guideline (version 1.2015) for treatment of desmoid tumour

Moreover, due to the germline mutations and chromosomal aberrations of the APC alleles, Bright-Thomas et al had performed a pre-clinical study of gene transfer

for the treatment of desmoid disease in FAP. Despite the success of transgene expression, further work is needed in animal models of desmoid disease to assess the clinical effects of gene therapy[25].

In short, surgery always aims at radical tumor resection with free margins, which, depending on localisation of surgery, may leave major soft tissue defects behind[26, 27, 28]. Although abdominal wall integrity after full-thickness surgery can be restored with direct sutures[4,26], reconstruction with synthetic materials is a common technique in major abdominal wall defects[27].

Here, a small tumor resection with tumor free margins and reconstruction of the abdominal wall was performed with a Bard Composix-Mesh. According to the recent literatures' recommendation, distant or free muscle flaps is advisable for greater abdominal wall defect coverage which is not accessible to local flaps[30].

Additionally, prosthetic material is more susceptible to bacterial infection and other complications[28], although new developed material showed encouraging experimental results [29, 30].

In this case, operated tumour had shown negative margins histopathlogically and negative β-Catenin mutation, some tumours exhibit a significant tendency for local recurrence even in the setting of negative microscopic margins, others do not recur after R1 resection, and some have been observed to spontaneously regress even without therapeutic intervention[31]. So, after considering less chance of recurrence by obtaining negative histopathological and immunochemistry report for margins of surrounding tissue, patient was discharged with follow up advice as per the NCCN guideline (**Figure 6**).

Conclusion

In conclusion, treatment of desmoid tumor is still controversial so final treatment remains puzzling. A variety of non-surgical treatments like definitive radiotherapy, anti-inflammatory drugs, hormone-therapy and chemotherapy has been advised but they are not proven to be curative. These therapies are limited to patients, in whom resection is not possible because of a widespread tumour infiltration.

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