Case Report

Histopathological and radiological findings of desmoid tumor of abdominal cavity

Muthu Saravanan A[1], I. Gurubharath[2*]

[1]Second-year Postgraduate Student, [2]Professor and Head
Department of Radio-Diagnosis, Shri Sathya Sai Medical College and Research Institute, Kancheepuram, Tamil Nadu, India
[*]Corresponding author email: **kavinilangovan@yahoo.co.in**

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Abstract

Desmoid tumors are rare slow growing, monoclonal, fibroblastic proliferation characterized by variable and often unpredictable clinical outcome, sometimes with aggressive infiltration of adjacent tissue but with no metastatic potential. Desmoids are histologically benign. On a molecular study level, they have been characterized by mutations in the β-catenin gene, APC (Adenomatous Polyposis Coli*)* gene or the CTNNB1. Gene studies are useful when the pathological diagnosis is difficult. Application of various multidisciplinary assessments along with multimodality treatment forms the basis of management for these patients. In selected asymptomatic patients, watchful waiting might be the most appropriate management. But for patients with desmoid located in other sites like the mesentery or at the head and neck region which might present with serious complications, thus requiring a more aggressive approach of treatment. In this article, we reported a 28-year-old female with a desmoid tumor of the anterior abdominal wall who underwent surgical resection. Preoperative evaluation was done using abdominal ultrasound, computed tomography, and magnetic resonance imaging. The histopathological examination of the resected specimen revealed a desmoid tumor. Because of the heterogeneity of the desmoid tumors, treatment needs to be individualized. However, complete surgical resection with negative margins seems to be the treatment of choice for this tumor entity and postoperative radiotherapy can help reduces the local recurrence rate.

Key words

Desmoids tumors, Ultrasound, Magnetic resonance imaging (MRI), Computed tomography (CT).

Introduction

Desmoid tumors are cytologically bland fibrous neoplasms originating from the musculoaponeurotic structures throughout the body. Desmoid tumors are benign neoplasm originating from the muscle aponeurosis and classified as deep fibromatoses [1]. They constitute 3% of all soft tissue tumors and 0.03% of all soft tissue neoplasm [2]. They are locally aggressive but lack any malignant potential [3]. This local infiltration causes compression of surrounding structures. High recurrence rate exists. Desmoid tumors can lead to death in cases with restricted surgical access [4]. Desmoid tumors are considered to be the most common cause of morbidity and mortality in post-colectomy patients with FAP (Familial Adenomatous Polyposis) [5]. Molecular studies have demonstrated desmoids in FAP as clonal neoplasm arising from mutations in or changes in APC alleles [6-8]. Cytogenic data provides additional evidence for the malignant potential of these lesions [9]. Desmoid tumors are commonly noted in the anterior abdominal wall, the proximal extremities, and the mesentery of the intestine (FAP) Grey-Turner's sign refers to bruising of the area between the last rib and top of the hip and is a sign of retroperitoneal hemorrhage. Cullen's sign is superficial edema and bruising in the subcutaneous fatty tissue around the umbilicus. Causes include acute pancreatitis, ruptured ectopic, aortic rupture, and coagulopathies [10]. In sporadic cases, they occur in sites of post-trauma scars or irradiation.

Case report

A 28-year-old female presented with complaints of painless swelling/lump in the anterior abdominal wall. There was no history of any previous abdominal surgeries done. The blood investigations were normal. Further evaluation was done using abdominal ultrasound, computed tomography, and magnetic resonance imaging. The histopathology of the resected lesion revealed a desmoid tumor. Written consent was obtained from parents or guardians before analysis Study period was between 2017-2018 in the department of radiology and imaging sciences, Shri Sathya Sai Medical College and Research Institute.

USG, CT and MRI findings were as per **Photo – 1 to 3**. On microscopic examination of the resected specimen, section studied shows a tumor mass composed of long, fascicles of spindle cells with ovoid or tapering nuclei, vesicular chromatin, small nucleoli, and indistinct cytoplasm. The tumor cells were admixed with abundant collagen and hyalinization. Few foci show tumor cells infiltrating the adjacent striated muscle and entrapping atrophic muscle fibers. Numerous dilated blood vessels were seen (**Photo – 4**). All these features are consistent with desmoids a tumor.

Photo – 1: USG B-mode image showing a well-defined homogeneously hypoechoic masses in the left rectus abdominis muscle.

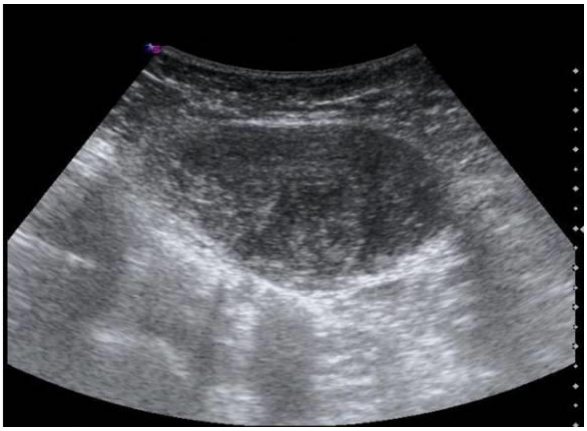


Photo – 2: CT abdomen axial image showing a well-circumscribed homogenous mass of soft tissue density, seen arising from the left rectus abdominis muscle.

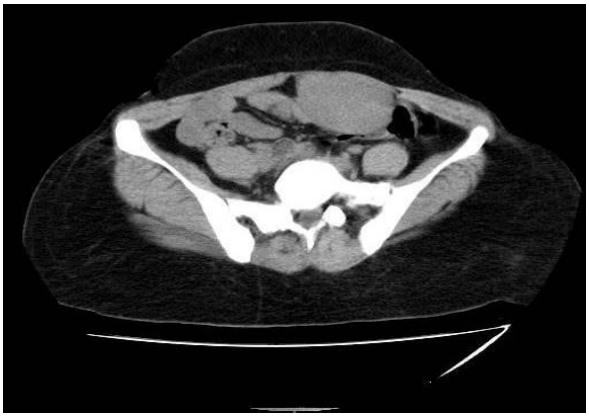
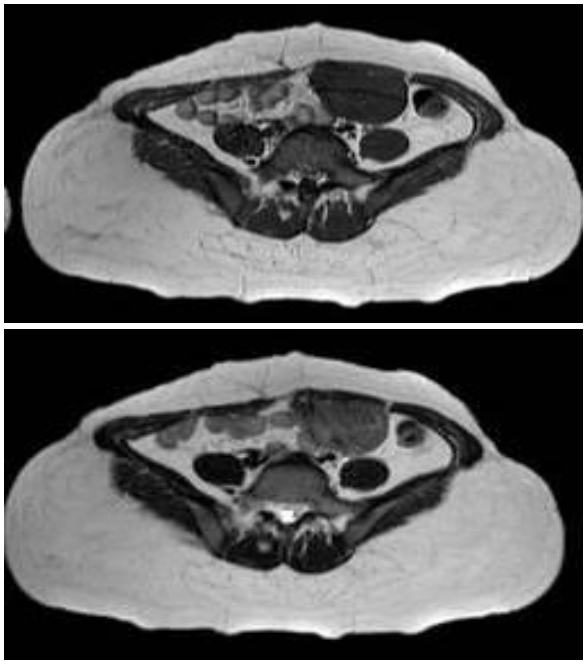


Photo – 3: MRI shows a well-defined lobulated soft tissue lesion measuring 11.4X5.4X3cm noted in the left anterior abdominal wall involving the left rectus abdominis muscle. Superiorly, the lesion extends to the level of supraumblical region and inferiorly up to the level of bladder. The lesion appears hypointense on T1WI and heterogeneously intense on T2WI.

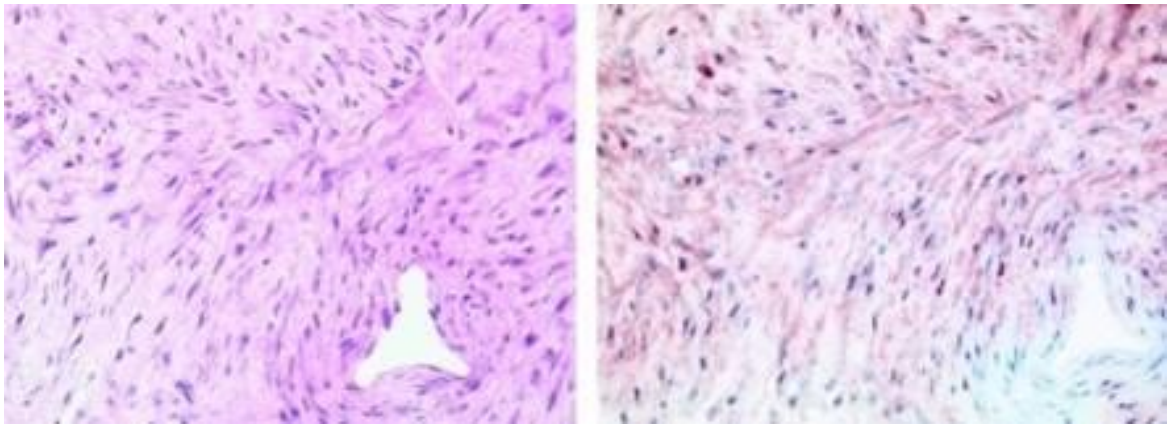


Results

Desmoid tumors are benign fibromatoses, originating from muscle aponeurosis and fascia with an infiltrating growth [11]. They are located primarily either abdominally or intraabdominal [1], meanwhile sporadic cases describe a localization within the chest wall [12] or retroperitoneum [13]. The desmoid tumor is often associated with Familial Adenomatous Polyposis (FAP) [14], female sex and sometimes with surgical trauma [15]. It has a high prevalence in pregnant women [16]. Recurrences occur in up to 45% of cases depending upon the size of tumors, mode of treatment and on the negative resection margins [17]. CT scan isolates the tumor and excludes any metastasis. MRI shows tumor hypointensity on T1WI and variable signal intensity on T2WI, depending on the collection of mucoid structures. Therefore, a differentiation from the rest of the solid tumors is not possible using morphological criteria [18]. Histology is the method which shows spindle cells arranged in long fascicles of variable cell-density with a few mitoses. Also, there is the absence of atypical nucleus-separations. Diffuse cell infiltrations of adjacent structures are noted. Immunohistochemical muscle cell markers like actin can help in differentiating desmoid tumors from fibrosarcoma [19, 20]. Therapeutic options are [21], hormone-therapy [22], anti-inflammatory treatment and chemotherapy [23] are ineffective and are done for patients for whom surgery cannot be done. The indication and effectiveness of adjuvant radiation are not yet proven. In an analytical study, significantly better local recurrence control was achieved with radiation and surgical resection in contrast to resection only. However, higher complication is associated with radiation therapy [24]. Other studies show a progress in the tumor even after radiation therapy [25] and a high local recurrence rate [26]. Due to the chromosomal aberrations and germline mutations of the APC alleles Bright-Thomas et al. did a pre-clinical study for desmoid treatment in FAP with gene transfer [27]. After the success of transgene expression, more work is needed to effects clinically on gene therapy and work on animal models. The most effective treatment of smaller desmoid tumors is the resection with negative margins, although it may not prevent local recurrence. However, in FAP patients, massive mesenteric lesions occur in post-colectomy patients.Nonsurgical treatment in these patients has a varied and unpredictable outcome [10, 21, 28, 29]. Middleton, et al. gave a report on successful short-term surgery in four cases of intra-abdominal tumors where treatment was inefficient without the surgery [28]. In conclusion, we need many therapeutic choices to treat abdominal tumors. Surgery will always aim at radical tumor resection with free margins, which, depends on the localization of surgery and may leave major soft tissue defect behind [28, 30, 31]. In spite of the fact, abdominal wall full-thickness surgery can be re-establish with direct sutures [4, 32]. Reconstruction with synthetic materials can be used in major abdominal wall defects [33]. Albeit, et al., conducted a study on the reconstruction of the

abdominal wall with a Bard Composix-Mesh in patients with post small tumor resection with tumor-free margins. Recent literature data recommends, muscle flaps for abdominal wall defect coverage [30, 32], moreover, the prosthetic material is likely susceptible to the bacterial infection and other complications [34, 35].

Photo – 4: Microscopic findings.



Conclusion

Thus, the treatment of desmoid tumors is difficult to understand. Non-surgical treatment resulted in various unpredictable outcomes. It is a treatment option reserved for patients with unresectable lesions or as an adjuvant therapy. Radical resection with clear margins is the mainstay treatment with a good prognosis but with minimal risk of local recurrence. The response can take months and continue for years.

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