

Case Report

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Massive Abdominal Wall Desmoid Tumor Following Deep Dorsal Vein Arterialization: A Case Report

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Abstract

Desmoid Tumor (DT), also called aggressive fibromatosis, is a non-malignant tumor of the mesenchymal stem cells. Although DTs are not malignant and do not metastasize, they are locally aggressive tumors and are hence referred to as a “benign malignancy.” Due to the variable natural history of DTs, there is no standard treatment modalities. However, surgical resection with microscopic negative margins (R0) is considered the mainstay therapy for a DT.

There is scarce literature discussing this rare tumor. Here, we report a case of a 30-year-old male patient who presented with an extremely large abdominal wall DT, with the size of 15x16x20 cm. The tumor had developed beneath the incision site of previous surgery. The tumor was removed with microscopic negative surgical margins. In a one-year follow-up, patient had no complaints and there were no signs of tumor recurrence. His MRI did not show regrowth of the tumor.

Background

Desmoid Tumors (DTs), also known as aggressive fibromatoses, are a benign proliferation of soft-tissue myofibroblasts [1]. While they do not metastasize, they are locally aggressive and are referred to as a “benign malignancy” [1-4]. DTs can develop in any part of the body although they most commonly arise in the abdomen [5]. Also, while they can occur at any age or in either sex, young females in their child-bearing age are particularly susceptible [2]. DTs account for 0.3% of all neoplasms and 3% of the neoplasms of soft tissue, affecting approximately one per 3-4 million individuals in the general population [6,7]. The exact etiology of DTs is not fully understood. However, it has been shown that genetics, especially mutations in the β -catenin gene, CTNNB1, or the Adenomatous Polyposis Coli Gene (APC), can increase in the risk of developing desmoid tumors [8-12]. DTs develop at surgical sites through a mechanism of hyper-proliferation of the mesenchymal stem cells responsible for wound healing [9,13,14]. Due to their unpredictable natural history and variable clinical course, there is no standard method of treatment for DTs [15-17]. Treatment plans range from watchful waiting to negative surgical

resection followed by adjuvant therapy including radiotherapy, chemotherapy and targeted therapy in selected cases. However, the mainstay of therapy is surgical resection of the tumor, followed by adjuvant radiotherapy, especially in cases where there is a gross-positive surgical margin [15-18]. Negative surgical margins, as well as adjuvant radiotherapy, have been proven to significantly decrease the recurrence rate in patients with DT [18].

Case Report

A 30-years-old male patient was referred to our center with the chief complaint of a progressively growing left lower quadrant abdominal mass and skin discoloration. He had no other complaints other than difficulty sleeping in prone position and cosmetic concerns. He had a past surgical history of deep dorsal vein arterialization due to erectile dysfunction performed 4 years ago. His surgical operation was successful, and he did not experience any immediate post-op complications. However, he noticed a small mass developing in the region of the surgical incision site one year following surgery. He reported to his surgeon, who prescribed NSAIDs for him. Nevertheless, the mass kept progressively growing and 2 years later, he presented again to his surgeon, where

a biopsy was taken from the mass. Results of pathology showed abdominal wall fibromatosis. His surgeon decided to observe the patient rather than surgically remove the tumor at the time since the patient did not have any major complaints due to the mass. However, the mass had kept progressively growing one year later, to the point where the patient had difficulty sleeping in the prone position in addition to cosmetic concerns.

Patient denied any history of using tobacco, alcohol or illicit drugs. He had no significant past medical history except for ED

which was initially treated with Testosterone Enanthate injections and Phosphodiesterase inhibitors prior to surgery. He also denied any family history of cancer or soft tissue diseases. Physical exam showed a non-tender firm mass in the left lower quadrant abdomen lateral to the surgical incision site. Mass was approximately 15x20 cm in size, and the overlying skin showed a brown pigmentation. Abdominopelvic CT scan with and without contrast showed a 20x15.5x12 cm left lower quadrant abdominal wall soft tissue mass with mild enhancement (Figure 1).

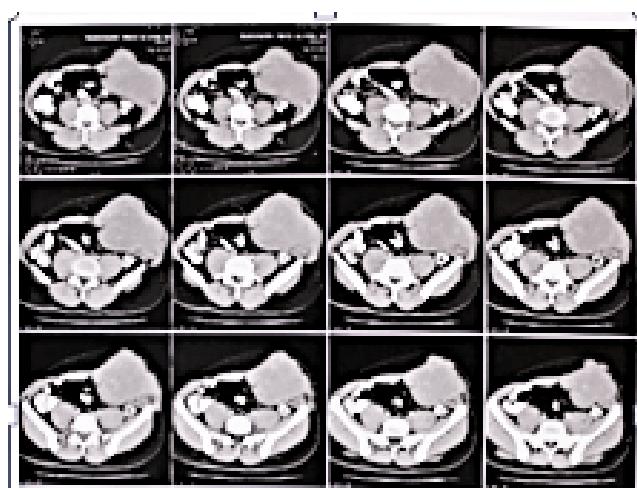


Figure 1: CT-scan of the patient's abdominal DT. CT scan was performed prior to surgery with and without contrast. The CT scan shows the presence of a large abdominal tumor. There is a 154x118 mm tumor soft tissue mass lesion on the left side of the abdominal wall with subtle enhancement. The differential diagnosis based on the CT scan was a solitary fibroma tumor, abdominal fibromatosis or soft tissue sarcoma.

Due to the problems caused by the mass and its progressive growth, the medical team decided to surgically remove the tumor. Patient underwent surgical resection of the tumor. The abdominal mass was approached through a left side pararectus incision under the inguinal canal. There was a large 20x15x14 cm hard mass extending from external oblique fascia deep to the peritoneum with severe adhesion to the abdominal wall muscles. It was surgically removed with gross negative surgical margins. The abdominal mass prior to the surgery and 2 weeks following surgical resection are shown in Figure 2.



Figure 2: The patient's abdominal mass immediately prior to surgical resection and two weeks after surgery. The DT, due to its large size, was causing cosmetic concerns and difficulty sleeping for the patient.

A specimen sized 15x12x8 cm was obtained and sent to pathology (Figure 3). The macroscopic exam of the specimen showed well-defined, creamy white rubbery mass with a lobulated external surface. The sections showed a creamy-white whirling pattern. The microscopic exam (Figure 4) showed proliferation of benign-looking spindle cells with a vesicular nuclei and eosinophilic cytoplasm. Foci of collagen bundle deposition were also observed without any evidence of mitotic figures or necrosis. The final histopathological diagnosis was confirmed to be abdominal fibromatosis without metastasis.

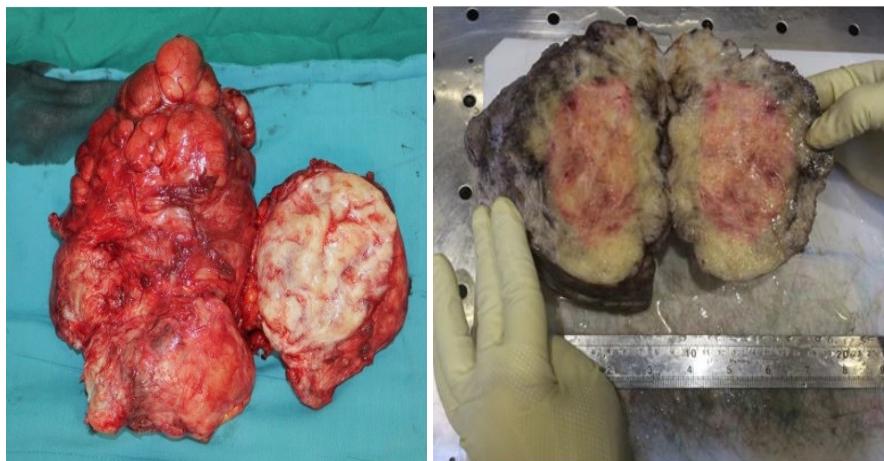


Figure 3: The raw DT that was recovered from the patient immediately following surgery. The DT was extremely large, with the size of 15x112x8 cm, one of the largest cases of abdominal fibromatosis reported in the literature.

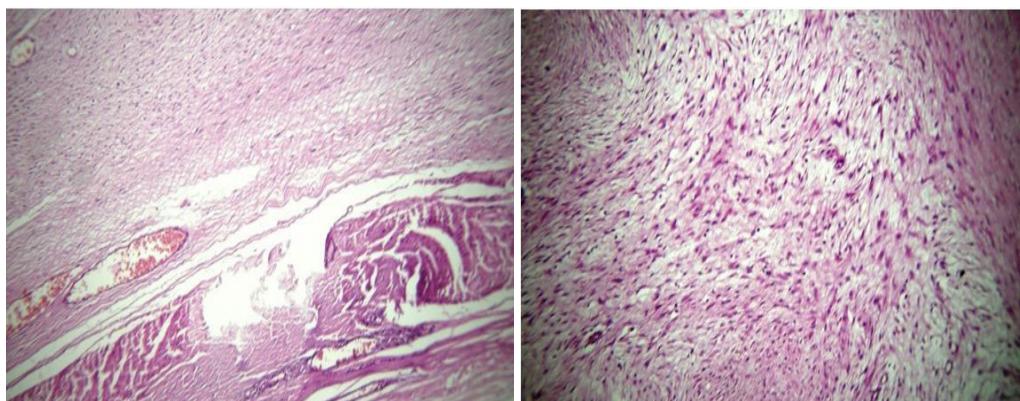


Figure 4: The histologic sample biopsy. The microscopic examination of the biopsy shows “neoplastic component with a proliferation of benign-looking spindle fibers with a fascicular architecture”. The cells show a vesicular nucleus with inconspicuous nucleoli and eosinophilic cytoplasm separated by bundles of collagen. No mitotic figures or necrosis were present in the microscopic exam of the biopsy slides. According to the presentation and the microscopic features of the biopsy, the final diagnosis was made to be a DT.

Discussion

Approximately half of all cases of sporadic DTs have shown mutations in the CTNNB1 gene [8,10,12]. The CTNNB1 gene is responsible for the expression of the β -Catenin protein. β -Catenin is a cadherin-binding protein that is involved in both cell to cell adhesions as well as gene expression [19]. Mutations in CTNNB1 gene lead to a dysregulated β -Catenin expression and are strongly associated with the development of sporadic DTs [8-12]. Adenomatous Polyposis Coli (APC) also plays an important role

in regulating CTNNB1 gene expression and β -Catenin levels [9,10,12,20]. A dysfunctional APC gene can increase CTNNB1 gene expression with a dysregulated production of β -Catenin protein, increasing the risk of developing DTs [9,10,12,20]. Wu et. al. injected mice with MSCs that had normal and elevated β -Catenin expression. In his experiment, mice that were injected with MSCs with elevated β -Catenin expression developed a DT, whereas those injected with wild-type cells without β -Catenin elevation did not [9].

The dysregulated expression of β -Catenin leads to uninhibited proliferation of MSCs by keeping them in a less differentiated state [9,14]. The abnormal proliferation of the MSCs and fibroblasts can in-turn lead to the development of a DT [9,14]. MSCs are involved in the proliferative phase of wound healing, where they differentiate into fibroblasts that form scar tissue [13,21]. Normally, the recruitment of mesenchymal progenitor cells into the wound-healing site is a tightly regulated and self-limiting process [13]. This tight regulation inhibits the development of tumors as the result of cell proliferation. The increased expression of CTNNB1 and the subsequent stabilization of β -Catenin protein in fibroblasts and MSCs leads to their uninhibited proliferation, leading to excessive scar tissue formation and the development of DT [9,13,14]. DTs have features of chronic wound healing, including increased angiogenesis and proliferation of fibroblastic cells within a collagen matrix [14].

In accordance with the underlying pathologic process, our patient developed a DT at site of previous surgery. Several other reports have shown that DTs develop at the site of previous surgery [22,23]. The anatomic site where our patient developed DT, i.e. within the rectus and the oblique muscles, is one of the most common sites for the development of these tumors, especially following abdominal surgery [22]. DTs have a relatively high rate of misdiagnosis (28). MRI or CT-scans, with or without contrast, are the most appropriate imaging technique for diagnosis and follow-up of DTs [24]. In our case, prior to the operation, we performed an abdominopelvic CT-scan, with and without contrast as shown in Figure 2. On a one-year follow-up, the patient had an MRI which showed that he had no recurrence of the tumor.

Watchful waiting is one of the mainstay methods for managing DT [25]. This method was initially used in our patient. In our patient, a decision to remove the tumor was made after the tumor had grown to a sizeable level and the patient started having symptoms. During the surgery, due to the large size of the tumor (Figure 2) and severe adhesion to surrounding structures, the operation was performed with great difficulty. It required partial resection of the abdominal wall muscles, including the rectus abdominis and oblique muscles to fully excise the tumor. According to existing reports, R0 is associated with 68.8% recurrence free survival (RSF), while R1 and R2 are associated with 34.1% RSF. In addition, it seems that adjuvant therapy does not significantly affect the recurrence rate of DT with R0 surgical margin [26,27]. Due to the negative microscopic surgical margins, we did not recommend adjuvant therapy for our patient. In the one year of follow-up, patient had no complaints and there were no signs of tumor recurrence. His MRI did not show regrowth of the tumor. Despite the fact that watchful waiting is one of the main methods of managing DTs, considering that larger DTs are more difficult to resect, as seen in our patient, it seems better to remove

the tumor before it grows to a point where it starts invading the surrounding structures.

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