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Case Report
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Desmoid-Type Fibromatosis Tumor Mimic Endometrioma of the Ventral Abdominal Wall after Cesarean Section. A Case Report

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Abstract

Introduction: Desmoid-type Fibromatoses (DF) are rare benign tumors that do not metastasize but have a high rate of recurrence and local aggressive growth. These tumors often develop at surgical sites and are associated with women of reproductive age.

Case Report: A 31-year-old woman presented with a painful palpable swelling of the right lower abdominal wall that occurred one month after cesarean section. Ultrasound and MRI were performed and showed a solid tumor measuring 37x17x29mm. Since the patient reported pain before and during menstruation, she was treated for suspected endometriosis with a progestin-emphasized oral combined contraceptive. As hormonal therapy did not reduce the pain, the tumor was removed. Histopathology showed desmoid-type fibromatosis, which extended at least to the margins. Further surgery had therefore to be performed.

Conclusion: Painful tumors occurring after cesarean section may be caused by rare tumors such as desmoid fibromatosis. Differential diagnosis is therefore required and can allow different treatment options.

Keywords: Desmoid-type tumor; Fibromatosis; Surgery; Cesarean section; Interdisciplinary approach; Active surveillance; Endometrioma

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Introduction

Desmoid Tumors (DT) or synonymously used terms such as Desmoid-type Fibromatosis (DF) and aggressive fibromatosis [1] are locally aggressive tumors of monoclonal, (myo)fibroblastic proliferation with a high rate of local recurrence, and thus characterized by a very variable clinical course and outcome [2]. DF is rare with an incidence of 2-6 cases per million per year [3-5]. It accounts for approximately 0.03% of all neoplasms [6]. The highest probability of developing DF is between the ages of 30 and 40, and females are more prone to be affected than men. The etiology is not yet fully understood. It is believed to be multifactorial with hormonal and genetic factors [4,7-13]. DF is often seen at surgical sites, especially after cesarean section [14,15]. Pregnancy [14,16] and the use of oral contraceptives [17] have been shown to be associated with DF, although the hormonal influence is not yet clear [17]. The tumor develops in the connective tissue of musculoaponeurotic structures [2]. Rapid growth is often seen in the early phase of the disease, and in response to pregnancy or hormonal treatment of any kind [16]. Most of the tumors seem to be sporadic, but in approximately 5-16% of cases there is a correlation between the appearance of a desmoid tumor and Familiar Adenomatosis Polyposis (FAP) [10,18-20]. The probability of developing DF is 1000 times higher in patients with FAP than in the general population [20]. Extra-abdominal locations are more often seen with sporadic tumors than with FAP-associated ones, although FAP-associated tumors appear to be larger, multifocal, intra-abdominal, and more likely in younger patients [4,19,20]. One of the molecular explanations of the development of DF is alterations in either the APC gene or in a CTNNB1 mutation [21,22]. A somatic mutation in the beta-catenin gene CTNNB1 (3q21) can be found in 85% of sporadic cases. In many FAP cases, there is a mutation in the APC gene (5q21-q22). Histologic features of DF include low to moderate cellularity, long fascicles of uniform cells, and dense collagenous stroma. There is an absence of malignant features such as hyperchromatasia and atypical cells [23]. The cells found appear similar to those found in the wound-healing process [24-

27]. The immunohistochemistry is characterized by positivity for nuclear β-catenin, smooth muscle actin, vimentin, Cyclooxygenase-2 (COX-2), and frequently also β-estrogen receptors, and by negativity for desmin, S100, CD34, and KIT [25,28,29]. A CTNNB1 mutation confirms the diagnosis [22,30,31]. In the past, DF was typically treated with surgical resection where possible. Alternatives such as radiation therapy, chemotherapy, hormonal treatment, and treatment with NSAIDs/Cox-inhibitors were only used in cases in which complete resection did not seem possible or would have resulted in loss of function [32-35]. Recent advances in cytogenetics and in the understanding of tumor biology have led to a paradigm shift in the management of DF: There has been a move away from aggressive surgery and towards a strategy of active surveillance [6,36], not least in light of the high risk of recurrence [37].

Case Presentation

We present the case of a 31-year-old female who was found to have desmoid-type fibromatosis after surgery for what had been believed to be post-cesarean endometriosis. The patient presented herself after an ultrasound and MRI of her abdomen due to pain and a palpable mass in her right lower ventral abdominal wall. She reported noticing the mass approximately one month after her second cesarean section. It was especially painful before and during menstruation. The patient also described general painful perimenstrual cramping in her lower abdomen, for which she was prescribed a progestin-emphasized oral combined contraceptive pill to treat suspected endometriosis [38]. The patient's second cesarean section had been performed four months prior to her visit to our outpatient clinic. No other operations or diseases were reported. The patient had experienced a significant weight loss of approximately 10 kg in the preceding three months, but without nightly sweating or an increase in infections. Studying the reports from the first cesarean section, we found a description of three small areas on the left and right of the uterus and in the area of the bladder where endometriosis was suspected. However, no sample for histopathology had been taken. There is no family history of malignancies and no allergies were reported. At presentation we saw a healthy female 163 cm tall and weighing 70 kg. An examination of the abdominal wall found a solid and painful tumor measuring 3 cm × 3 cm approximately four centimetres lateral to the umbilicus. The ultrasound performed showed a solid tumor of 37 mm × 17 mm × 29 mm in the exact location of the palpable mass with no abnormal blood flow. Based on the clinical examination, radiology, the patient's history, and the report from the first cesarean section, our working hypothesis was of extragenital endometriosis. We therefore discussed the surgical removal of the palpable mass. The patient was operated on via her existing scar from the cesarean section without any complications. The tumor was easily exposed by distancing the fascia from the musculus rectus abdominus. Clinically complete resection was performed. Instead of endometriosis, the histopathologic findings showed a DF measuring 50 mm in diameter with the previously mentioned characteristics. Molecular genetic testing found a CTNNB1 mutation and this confirmed the diagnosis. Unfortunately, the tumor extended to the circumference of the resected part. Given the R1 resection and the favorable location, we discussed a second surgery to remove the area where the tumor had been in order to reduce the risk of recurrence. This time, no further abnormal cells were found; R0 resection was achieved. Due to the high risk of recurrence, intensive follow-up was discussed with the patient. The patient was also advised to seek genetic counselling due to the 10% risk of FAP after DF.

Figure 1: Ultrasound of the lower abdominal wall with the tumor described.

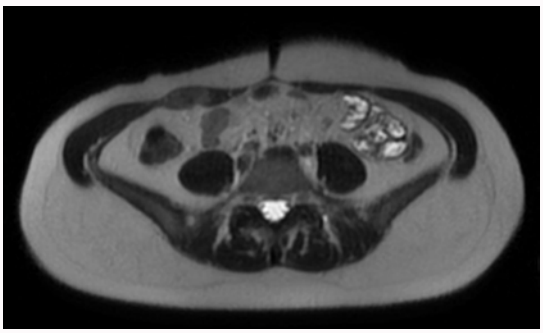

Figure 2: MRI (T2 image) with tumor in M. rectus abdominis of the right side.

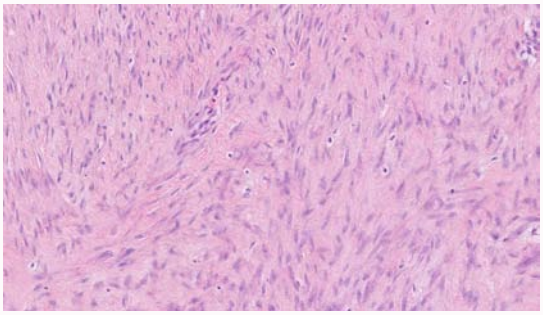

Figure 3: Desmoid Fibromatosis: Examination shows bland fibroblastic and myofibroblastic cells without atypia (hematoxylin eosin (HE), 40x).

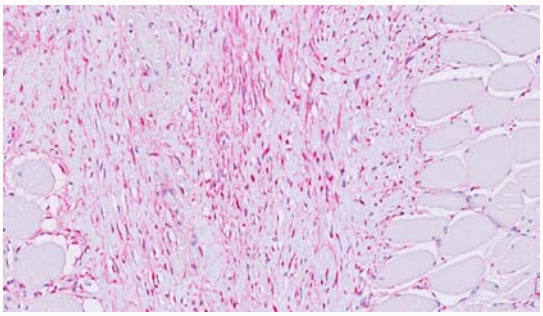

Figure 4: Desmoid Fibromatosis: Infiltration of the adjacent skeletal muscle. Nuclear expression of ß-catenin. (ß-catenin 20x).

Discussion

Retrospectively, we had an interdisciplinary discussion of our case. Taking account of the fact that the patient just had undergone an operation, and correlating the clinical with the MRI findings,

we should have considered the possibility of DF. In MRI, the characteristics of DF depend on the histological components of the tumor [39-41]. The presence of linear, non-enhancing T1 and T2 hypointense bands within the tumor (band sign) is reported to be a characteristic MRI finding in DF, seen in up to 90% of cases [6,39]. However, it should be noted that the band sign is not pathognomonic for DF, as it is also found in other tumors of the musculoskeletal soft tissue. Had DF been suspected, a core needle biopsy could have been offered to the patient to confirm the diagnosis, and other treatment options could then have been discussed with her [1,36]. The patient should have been informed that spontaneous regression occurs in up to 30% of cases [1,18,36] and that event-free survival rates with active surveillance are similar to survival rates after surgery [42], and she could then have made an informed choice between the two options. As described in the literature [43], one third of recently diagnosed patients experience pain (especially those with larger tumors in neck/shoulder locations). Pain has been associated with unfavourable event-free survival rates after adjustment for confounders [43]. Due to the operation, our patient is pain-free up to this date.

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