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# Rare Abdominal Desmoid Tumor: Case Report and Review of Literature

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**Abstract:**
A desmoid tumor is a clonal fibroblast proliferation, occurring in deep soft tissues with a significant recurrence tendency. Aggressive fibromatosis is a rare disease. The prevalence in the population ranges from 2 to 4 cases per million. Mentioned neoplasms are highly unpredictable in terms of development. Typically, in the course of desmoid tumors, metastases do not occur, but they often infiltrate the surrounding tissues, which may be associated with numerous complications. There are currently no randomized standards of therapy designated, which is undoubtedly a challenge when referring to treatment.

To a 37 year old patient, due to a pathological mass in the abdomen, the MR examination was performed. It showed the presence of an irregular, heterogeneous structure on the right side of the abdominal cavity. A laparotomy was performed. During the procedure adhesions were released and the giant abdominal tumor was removed in one block with part of the duodenum, caecum, ascending colon and part of ileum. End-to-end duodenal anastomosis and stapled, side-to-side ileo-transverse anastomosis were used. The surgical field was being drained. The postoperative course was uneventful. Tissue material was collected for a routine, histopathological examination in which the intra-abdominal form of fibromatosis was

diagnosed. The macroscopic image describes a tumor with dimensions of 25.0 cm x 25.0 cm x 20.0 cm.

**Key words: desmoid tumor, abdominal tumor, fibromatosis**

**Introduction:**

According to the World Health Organisation, a desmoid tumor is a clonal proliferation of fibroblasts occurring in deep soft tissues, characterized by a significant tendency to recur. (Fletcher, 2013) These tumors are lesions that are not specific in terms of occurrence in a particular location; they can localize to almost any part of the body. They are tumors that do not give distant metastases, however, ingrowth into adjacent tissues is relatively common. (Wang, 2018) The incidence of aggressive fibromatosis in the population is 2 to 4 cases per million. (Nieuwenhuis et al., 2011) Desmoid tumors affect women more often than men, with a peak incidence between 30 and 40 years of age. (de Camargo et al., 2010) These tumors are highly unpredictable; they can develop into sizable lesions that lead to the destruction of neighboring structures, while cases of spontaneous regression have also been described. (Garcia-Ortega et al., 2020) The etiology of aggressive fibromatosis is not known. Approximately 5-10% of cases are associated with familial adenomatous polyposis (FAP), with the remaining 90% showing an idiopathic occurrence. (Braun et al., 2021) Currently, there are no designated randomized standards of treatment, but a multidisciplinary approach is undoubtedly required. Depending on the size, growth and complaints, different treatments are used. (Martínez-Martínez et al., 2021) The treatment for large, growing desmoid tumors is surgical resection, while the risk of recurrence is high. (Wang et al., 2022) Other therapeutic modalities are also used, such as radiotherapy or hormone therapy especially used in

inoperable DT cases or as an adjunct to surgical methods. (Ishikawa et al., 2021) In cases in which fibromatosis does not produce symptoms and does not spread, only observation or prophylactic treatment with NSAIDs is indicated. (Church et al., 2005)

**Case report:**

A 37 year old patient came to the Oncological Surgery Outpatient Clinic due to a gradually enlarging mass in the abdomen. During a visit to the Oncological Surgery Outpatient Clinic a decision was made to refer the patient to the Department of Oncological Surgery for a tumor of the abdominal lining, with the aim of surgical removal in its entirety or taking sections.

The patient underwent an abdominal MR examination with contrast in T1, T1-FS, T2, T2-FS, DWI, RESTORE, VIBE, in-phase and counter-phase sequences. It was described, visible on the right side of the abdomen, an irregular, heterogeneous nodular structure measuring 210x118x177 mm. Hypointense at T1 and T2, its structure was differentiated by small areas of diffusion and fragments of increased IS at T2 and T2-FS, which were not enhancing and could be due to necrosis or cystic degeneration. The lesion was not extinguished in counterphase, modeled the adjacent intestinal loops, compressed the OGD, and had obliterated outlines in contact with the hook process of the pancreas and the abdominal wall. On examination, the following conclusions were obtained: an abdominal tumor mass with an unclear point of origin, possibly corresponding to, inter alia, a fibrous tumor with the presence of necrotic foci, unlikely pathological lymph nodes, a focal lesion of the liver in the nature of a hemangioma, splenomegaly.

The patient was admitted to the Clinical Department of Oncological Surgery due to a tumor of the abdominal lining, for surgical removal in its entirety or taking sections. Physical examination revealed abdominal pathology. A hiatal hernia and an extensive, mobile tumor measuring 12x8 cm on the right side in the intra-abdominal and partially epigastric region were found. The patient's preparation for elective surgical treatment was initiated.

A laparotomy was performed. During the procedure, adhesions were released and the giant abdominal tumor was removed in one block with part of the duodenum, cecum, ascending colon and part of the ileum. Tissue material was collected for routine histopathological examination. An end-to-end duodenal anastomosis and a side-to-side ileocecal stapler

anastomosis were used. The surgical site was drained. The postoperative course was uncomplicated. A follow-up chest X-ray and head CT scan were performed with no abnormalities. Rehabilitation visits were carried out with the aim of upprighting the patient as soon as possible.

The histopathological examination diagnosed intra-abdominal fibromatosis. The macroscopic image described a tumor measuring 25.0 cm x 25.0 cm x 20.0 cm with an adjacent cecum and ascending colon approximately 15.0 cm long, appendix 8.0 cm long and 0.4 cm in diameter. Tumor with a smooth serous surface on cross-section was lymphoid with several fields of mucinous infiltration up to 3.0 cm in diameter, infiltrating the ascending colon at 2.5 cm from the distal margin over a distance of approximately 5.0 cm and covering 3⁄4 of the circumference, without reaching the lumen of the bowel. Immunohistochemical studies showed the presence of calretinin and SMA-focally. The sampled lymph nodes had no tumor metastases.

The patient was discharged home  in good general condition and further recommendations. Daily abdominal wound care including dressing changes, instructions for low molecular weight heparin injections and dietary recommendations in the form of an easily digestible diet were recommended.

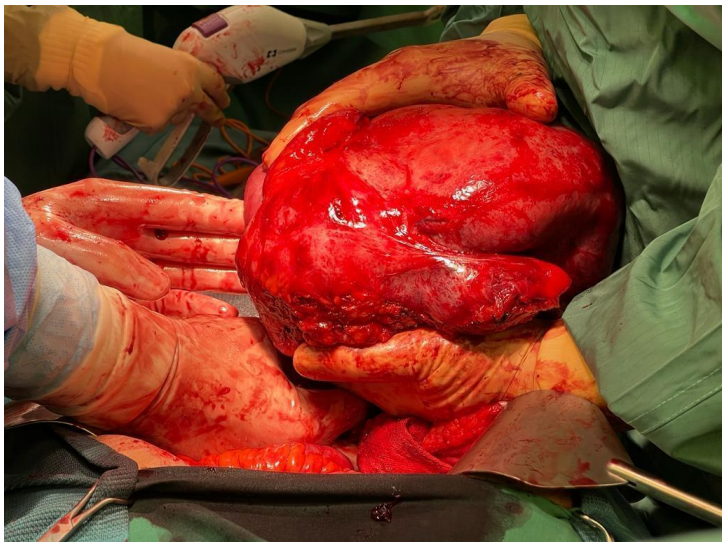

Figure 1. Author's photo of the resected tumor.

**Discussion:**

Desmoid tumor (DT), or deep fibromatosis or aggressive fibromatosis, is a disease entity classified in the ICD as D48.1. It is a rare disease, (Penel et al., 2017), (Desmoid Tumor Working Group, 2020) affecting 2-4/ 1 million people per year. (Nieuwenhuis et al., 2011) Desmoid tumors are characterized by an unpredictable and varied course, a lack of metastatic tendency, and a tendency towards multifocal proliferation. (Penel et al., 2017), (Desmoid Tumor Working Group, 2020) In their course, one encounters both a sudden, rapid increase in tumor mass and unexpected, long-lasting remissions, progressing without a cause that has not been grasped so far, so one must be vigilant in the diagnostic and treatment process. (Master SR et al., 2022), (Penel et al., 2017)

Despite the lack of detailed knowledge on the etiology of DT, it is thought that the influence of genetic and hormonal factors may be crucial in the multifactorial pathogenesis of the described entity. (Master SR et al., 2022), (Penel et al., 2017), (Desmoid Tumor Working Group, 2020) When considering the etiology of DT, mention should be made of so-called sporadic desmoid tumors, which account for the majority of diagnoses, and desmoid tumors associated with a germline mutation in the APC gene. (Master SR et al., 2022), (Penel et al., 2017), (Desmoid Tumor Working Group, 2020) In 85-90% of individuals with developed sporadic desmoid tumors, mutations in the CTNNB1 gene, (Master SR et al., 2022), (Penel et al., 2017) concerning the beta-catenin pathway, which is a protooncogene, are recognised. This mutation leads to the accumulation of beta-catenin, its over-stabilisation and a series of transformations that lead, among other things, to the expression of proliferative factors. (Master SR et al., 2022) Desmoid tumors associated with a germline mutation in the APC gene account for 10-15% of all cases and, in addition to their strong association with familial adenomatous polyposis, they have been observed to coexist with Gardner syndrome and Turcot syndrome (Penel et al., 2017) In the diagnostic process, it can be assumed that the detection of one mutation can virtually rule out the presence of the other. (Master SR et al., 2022)

There are three forms of desmoid tumor, depending on their location: abdominal, intra-abdominal and extra-abdominal forms. (Master SR et al., 2022), (Desmoid Tumor Working

Group, 2020) DTs can localize in any area of our body, but statistically it most commonly occupies the extremities, abdominal wall, bony rims and mesenteric region. (Penel et al., 2017)

The peak incidence is in the third and fourth decades of life, (Master SR et al., 2022), (Penel et al., 2017), (Desmoid Tumor Working Group, 2020) but it is important to remember that it can be encountered in a patient of any age. (Master SR et al., 2022) Women are at higher risk, especially those who are pregnant, in the perinatal period (Master SR et al., 2022), (Penel et al., 2017) or taking contraceptive hormones. This is most likely related to the high levels of estrogen found in the body in the situations mentioned (Master SR et al., 2022). This risk visibly and significantly decreases during the menopausal period. Another important risk group is those with familial adenomatous polyposis (FAP). According to the data, 5-10% of all desmoid tumors affect those burdened with FAP. (Master SR et al., 2022), (Desmoid Tumor Working Group, 2020) In 25% of patients, a history of mechanical trauma or previous surgery in the area of tumor formation is noted, which may suggest an association of tissue damage with a predisposition to desmoid tumor development. (Master SR et al., 2022)

Desmoids are often tumors with no specific symptoms. (Master SR et al., 2022) With time of tumor growth, increasing pain and/or pressure-related impairment of nearby organs may occur, which may be life-threatening. The above symptoms may suggest the presence of a neoplastic process at the beginning of the diagnostic procedure. (Master SR et al., 2022), (Penel et al., 2017)

The diagnosis is based on histopathological examination of a biopsy specimen. (Master SR et al., 2022), (Penel et al., 2017) Desmoid tumors are histologically characterized by proliferation of uniform spindle cells against a background of abundant collagenous lining and vascular network. The cells show similarity to myofibroblasts. (Master SR et al., 2022) Immunohistochemical examination primarily uses the appearance of coloured complexes, when testing for nuclear catenin beta. A positive result greatly supports the diagnosis of DT, but its absence does not exclude the condition. (Master SR et al., 2022) It is important to bear in mind the limited usefulness of the above test, depending on the immunohistochemical method used, (Penel et al., 2017) and that immunohistochemistry cannot replace next-generation sequencing, which is suggested in most indications. (Master SR et al., 2022) In addition to histopathology and immunohistochemistry, it is important to bear in mind imaging studies to assess the extent of the tumor and the extent of possible infiltration, such as CT and

MRI. These examinations are crucial in the therapeutic process as well as in monitoring the patient's condition. (Master SR et al., 2022), (Penel et al., 2017) It has been suggested that MRI is more diagnostically useful. (Master SR et al., 2022) On T1-dependent images, desmoid tumors are hypointense or isointense, on T2-dependent images they are hyperintense and the lesion enhances moderately after contrast administration. (Master SR et al., 2022) DT should be differentiated from breast cancer, GIST, IMT, SFT, sclerosing mesenteritis, retroperitoneal fibrosis, fibrosarcoma, familial colonic polyposis and the cutaneous form of Gardner syndrome. (Master SR et al., 2022)

Until a few years back, surgery was the preferred treatment for desmoid tumours. (Master SR et al., 2022), (Kasper et al., 2020) Nowadays, many studies question the necessity of surgical intervention and suggest that other active treatment options should be considered and used interchangeably or in combination, bearing in mind the benefits and side effects of each therapy for the patient. (Penel et al., 2017), (Desmoid Tumor Working Group, 2020) Combination treatment appears to lead to better outcomes and reduced mortality, but these data are not statistically significant. (Desmoid Tumor Working Group, 2020) Increasingly, however, active treatment is not offered to the patient as a first-line solution. (Desmoid Tumor Working Group, 2020) Physicians readily adopt the tactic of simply observing the tumor, in the context of its growth, (Penel et al., 2017), (Desmoid Tumor Working Group, 2020) which yields patient comfort and survival outcomes comparable to those obtained by implementing medical interventions. (Desmoid Tumor Working Group, 2020) This belief is based on studies that indicate that only a small percentage of DTs tend to progress. (Penel et al., 2017) Follow-up is performed every three to six months with imaging studies. If there is significant progression or progression within organs that could be life-threatening if affected by DT, the implementation of active treatment should be strongly considered. The type of treatment usually depends on the location of the tumor. Surgery appears to be most beneficial for DT occupying the abdominal wall. Systemic treatment is the treatment of choice when the desmoid tumor is in the abdominal cavity, retroperitoneal cavity or pelvic cavity, and drug treatment when the head, neck or thorax is involved. Radiotherapy is recommended in elderly patients with comorbidities when the lesion is rapidly progressing and poses a threat to vital organs. (Desmoid Tumor Working Group, 2020) Systemic treatment includes chemotherapy, targeted therapy, hormonal therapy and administration of non-steroidal anti-inflammatory drugs. (Master SR et al., 2022) Adequate data are not available to select the best possible

therapy for a particular case, demonstrating the importance of the topic and the need for further research and discussion. (Penel et al., 2017), (Shields et al., 2001)

**Conclusions:**

Desmoid tumors, also known as aggressive fibromatosis, are rare, slow-growing tumors that arise from the connective tissue cells, usually in the abdomen, but they can occur in other parts of the body as well. The symptoms of desmoid tumors depend on their location.

The exact cause of desmoid tumors is unknown, but certain factors may increase the risk of developing them, including a history of trauma or surgery in the affected area, familial adenomatous polyposis (FAP), and pregnancy. Diagnosis of desmoid tumors typically involves imaging tests such as ultrasound, MRI, or CT scan, as well as biopsy to confirm the diagnosis. Treatment options for desmoid tumors vary depending on the size, location, and symptoms. Watchful waiting may be an option for asymptomatic tumors, while active treatment may include surgery, radiation therapy, chemotherapy, or targeted therapy with medications such as tyrosine kinase inhibitors. The prognosis for desmoid tumors is generally favorable, but they can be challenging to manage due to their unpredictable growth patterns and tendency to recur after treatment. Close monitoring and multidisciplinary care are often necessary for optimal management.

**Author's contribution**:

Conceptualization, Gracjan Sitarek; methodology, Marta Żerek, Gracjan Sitarek; software, Paulina Bednarczyk; check, Paulina Bednarczyk; formal analysis, Marta Żerek; investigation, Damian Chruścicki; resources, Gracjan Sitarek; data curation, Marcin Pelc; writing - rough preparation, Wojciech Płonka; writing - review and editing, Wojciech Płonka; visualization, Damian Chruścicki; supervision,Paulina Bednarczyk; project administration, Marcin Pelc;

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