

Case Report

DESMOID-TYPE FIBROMATOSIS OF THE MESENTERY: A CASE REPORT

Tatyana M. Betova,
Savelina L. Popovska,
Radoslav G. Trifonov¹,
Konstantina S. Karakadieva¹,
Genoveva B. Valcheva²,
Lachezar S. Tanchev³

*Department of Pathoanatomy,
Medical University – Pleven,
Bulgaria*

*¹Student of Medical University –
Pleven, Bulgaria*

*²Division of Oncological Surgery,
Oncology Center,
Medical University – Pleven,
Bulgaria*

*³Division of Gynecologic Oncology,
Oncology Center,
Medical University – Pleven,
Bulgaria*

Corresponding Author:

Tatyana M. Betova
Department of Pathoanatomy,
Medical University – Pleven
1, Sv. Kl. Ochridski Str.
Pleven, 5800
Bulgaria
e-mail: betova@abv.bg

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Summary

Desmoid-type fibromatosis is a rare mesenchymal neoplasm with locally aggressive, infiltrating and destructive growth that is not characterized by a metastatic potential. According to their anatomical position, desmoid-type fibromatoses can be divided into three groups: extra-abdominal, intra-abdominal, and fibromatoses of the abdominal wall. Mesenteric fibromatoses account for 8% of the intra-abdominal ones. The latter are characterized by myofibroblastic proliferation and infiltration of both the pelvic and abdominal organs. We report a 26-year-old woman who complained of abdominal enlargement, feeling of heaviness, discomfort and reflux, which symptoms dated back 1-2 months prior to hospitalization. The patient underwent laparotomy due to subocclusive symptoms. Intraoperatively, a tumor sized 30 cm in diameter was found. The tumor originated from the mesentery and infiltrated in the jejunum, the entire ileum, and part of the caecum with perforation towards the abdominal cavity. The histological and immunohistochemical examinations are important for clarification of the diagnosis. The treatment requires a multidisciplinary approach, in which the surgical method has the key role.

Key words: desmoid tumors, mesenteric fibromatosis, pathology, surgery

Introduction

The desmoid-type fibromatoses (DTF) are rare benign lesions of the soft-tissues that belong in the group of tumors of (myo) fibroblastic origin, characterized by local aggressive growth and relapses but without metastatic potential.

According to the anatomical location, DTF are divided into: extra-abdominal (60%), abdominal (25%) and intra-abdominal (8-15%) [1, 2]. The latter are subcategorized as mesenteric fibromatoses (MF), mesenteric fibromatoses associated with Gardner's syndrome, and pelvic fibromatoses [3-5].

Eighty percent of the intra-abdominal fibromatoses originate from the mesentery of the small intestine, and rarely – from the ileocolic mesentery [2, 6].

DTF demonstrate benign morphology. However, their biological behavior, such as infiltrative growth and local recurrence, has provided reasons for some

authors to refer to them as intermediate malign neoplasms [2, 3, 5].

We report a case of a 26-year-old female presented by abdominal enlargement and abdominal mass, who was initially observed for tumor of the genital or gastrointestinal systems.

Case Report

A 26-year-old female was admitted to the oncogynecology department of University Hospital – Pleven with the following complaints: enlargement of the abdomen, scanty genital bleeding and gastro-esophageal reflux, dating 2-3 months back. Her surgical history revealed an appendectomy, performed 15 years before, and childbirth – 6 years before admission. There was no family history of desmoid tumors.

The physical examination revealed an abdominal enlargement (as that of a 5-month pregnancy) and a tumor mass, which on was movable and nonpainful on palpation.

The laboratory findings were normal. The CT-scan revealed a tumor sized 170/120 mm in axial perspective, situated in the lower hypochondrium and the pelvis, reaching the fundus uteri.

The patient underwent an explorative laparotomy with biopsy.

Intraoperatively, a solid tumor mass originating from the mesentery with infiltration of intestinal loops was found. The uterus and adnexa were normal, without enlargement of the abdominal lymph nodes.

A tissue sample was collected for morphological examination. The diagnosis of fibromatosis was favored and included in differential diagnoses: leiomyoma, leiomyosarcoma, gastrointestinal stromal tumor (GIST), solitary fibrous tumor and neurofibroma.

On the basis of the clinical data for ileus and peritonitis, the patient underwent emergency surgery in the oncosurgery department. Intraoperatively, a tumor formation with a diameter of 30 cm, occupying a large part of the jejunum, the whole ileum, and part of the caecum, with a perforating opening towards the abdominal cavity, was excised (Figure 1 and Figure 2). Partial resection of the small intestine and the colon was performed. A term-terminal entero-entero anastomosis and lateral-lateral enterocolic anastomoses were made.

The postoperative period was complicated with *anastomotic insufficiency and peritonitis*, leading to lethal outcome.

Macroscopic examination of the specimen showed an unencapsulated formation and 30/30 cm in size. The cut surface of the tumor was homogeneous with infiltration of intestinal loops but without necroses and hemorrhages (Figure 3).

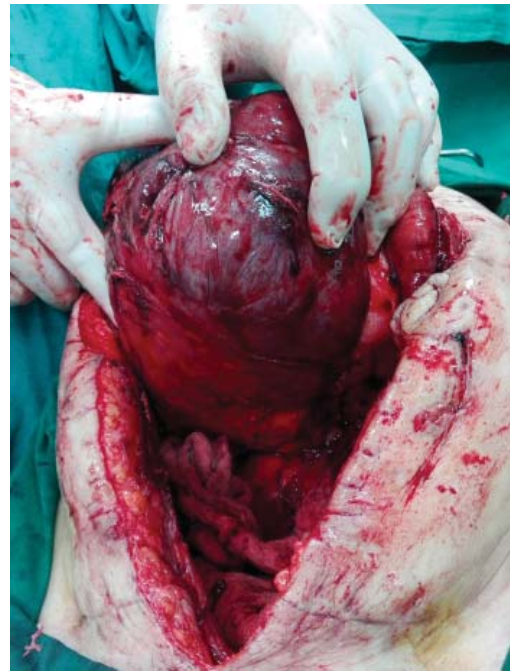


Figure 1. Intra-abdominal desmoid-type fibromatosis (macroscopic view-intraoperatively)

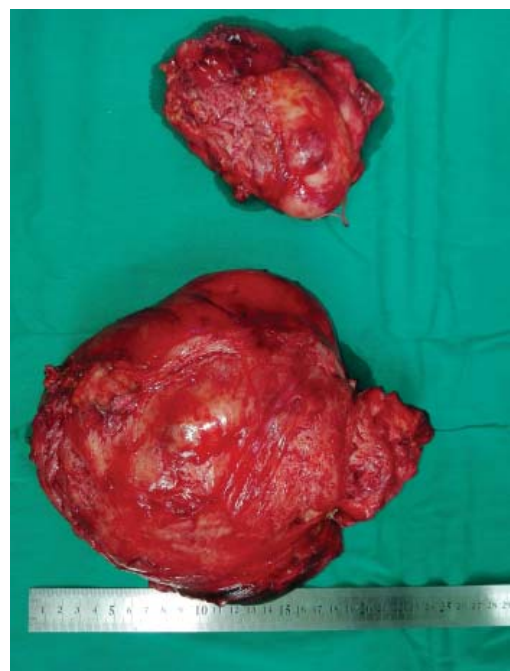


Figure 2. Specimen from the mesenteric desmoid-type fibromatosis (macroscopic view)



Figure 3. Macroscopic view of mesenteric desmoid-type fibromatosis with infiltration of small intestine



Figure 4. Microscopic view of desmoid-type fibromatosis, Haematoxylin and eosin stain, magnification 400x

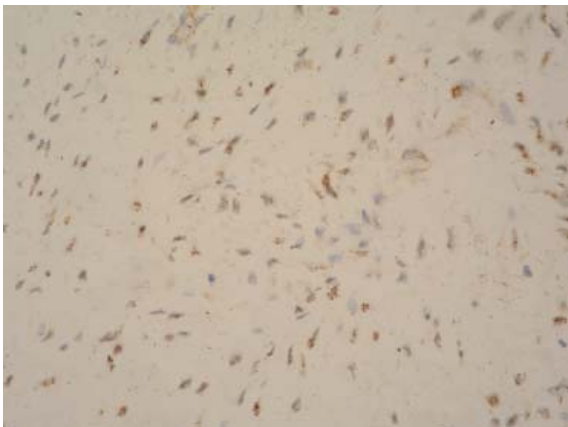


Figure 5. Moderate diffuse expression of beta-catenin in tumour cells, immunohistochemistry (IHC), magnification 400x

Histologically, the lesion consisted of elongated spindle-shaped fibroblasts, scant cytoplasm, fascicle-like structure, and collagenous stroma with prominent blood vessels without polymorphism, atypical mitoses, necroses, and hemorrhages (Figure 4). The microscopic examination revealed infiltration of the desmoid-type fibromatosis in the wall of the small intestine.

Immunohistochemically, 90% of the cells demonstrated a positive nuclear expression for β -catenin, low proliferative index for Ki-67 (2.7%), focal positivity for smooth muscle actin (SMA), and negative immune reaction for CD117, DOG1, CD34, CD99, Cytokeratin AE1/AE3 (CKAE1/AE3), Desmin, Epithelial Membrane Antigen (EMA), S100, Estrogen (ER) and Progesterone (PR) antibodies (Figure 5 and Figure 6). The diagnosis – mesenteric fibromatosis – desmoid type was made on the bases of the aforementioned histological and immunohistochemical features.

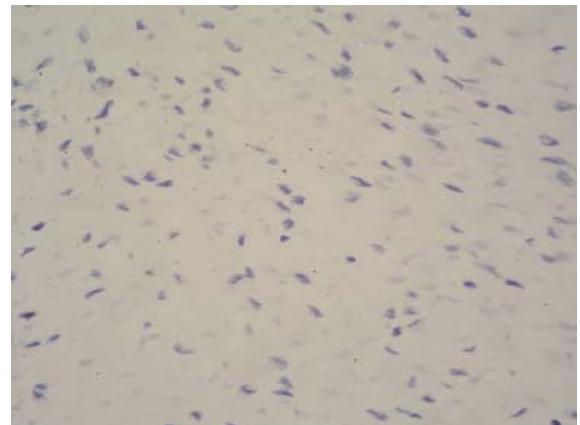


Figure 6. Negative immunostaining for DOG1 antibody, IHC, magnification 400x

Discussion

The desmoid-type fibromatosis is an intra-abdominal fibromatosis, also known as “aggressive fibromatosis” or “deep fibromatosis” [1].

The term “desmoid” (Greek “desmos”, meaning “tendon-like”) was introduced by Muller in 1838 [6]. Of DTF, 12-18% are intra-abdominal, originating from the mesentery of the small intestine and, rarely, from the omentum, retroperitoneum, pancreas, gastroesophageal junction, diaphragm and appendix [2, 4-7].

The frequency varies between two and five per million people annually [1, 6]. They are found primarily in young age: 25-40 years of age, with predilection in females (F:M ratio=3:1). Only one research on 130 patients established 55% male prevalence [2, 5, 7].

The reasons behind the clonal proliferation are still unclear but it has been found that there is a connection with trauma, previous abdominal surgeries, hyper-estrogenic state and hormonal stimulation. Most DTF cases are sporadic, and in 80-90% of them a somatic mutation of the Adenomatous Polyposis Coli gene (APC) is determined, which activates a mutation in Catenin Beta 1 gene (CTNNB1), which results in accumulation of β -catenin and turns out to be the triggering mechanism of the fibroblastic proliferation [2, 3, 5].

Clinically, DTF are presented by a slow abdominal enlargement and various symptoms such as compression, intestinal obstruction or circulatory disorders, associated with ischemia and hemorrhages due to intestinal perforation [2-4]. In this case, the initial abdominal enlargement was asymptomatic but later on subileal manifestations, perforation and peritonitis were found. Most mesenteric DTF are sized over 10 cm, as confirmed in the case we report [1, 4, 6].

Images of DTF could imitate GIST, lymphoma, sarcoma or carcinoma [2, 4, 6, 8]. In this case, the CT-scan of the abdomen visualized an intra-abdominal tumor with the characteristics of sarcoma. The final diagnosis for DTF was based on the histological and immunohistochemical examinations, which demonstrated a benign soft-tissue lesion with fibroblastic and myofibroblastic differentiation and infiltrative growth in the wall of the hollow organs [1, 2, 7, 9]. The diagnosis we made was supported by the immunohistochemical nuclear expression for β -catenin and the patchy positivity for SMA. The differential diagnosis of DTF includes a wide spectrum of intra-abdominal lesions such as GISTs, leiomyomas, leiomyosarcomas, solitary fibrous tumor, neurofibroma, and these were also suspected in this case. The differentiation from GIST is done with CD117, DOG1 and CD34, which do not have an expression for DTF. Leiomyomas and leiomyosarcomas express markers for myogenic differentiation – SMA, Desmin, H-caldesmon

but they are negative for β -catenin. The solitary fibrous tumor has a similar histological structure and positive expression for CD34 and a negative one for β -catenin [2, 8, 9].

Although some mesenteric DTF may undergo spontaneous regression, the principal method for treatment remains the surgical one – with wide resection and negative margins without infiltration. Alternative approaches like radiotherapy, adjuvant chemotherapy, and regular hormonal therapy are recommended for large tumorous masses, partial resection and positive edges of resection [2].

Conclusions

DTF of the mesentery are rare fibroproliferative lesions that simulate intra-abdominal malignant tumors. The histological and immunohistochemical examinations are important for clearing up the diagnosis. The treatment requires a multidisciplinary approach, in which the surgical method has the key role.

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