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CASE REPORT

A rare case report: desmoid-type fibromatosis

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ABSTRACT

Desmoid tumors, also known as desmoid-type fibromatosis (DF), develop from musculoaponeurotic tissues. They are fibrous tumors of benign character that do not metastasize. However, they can take an aggressive course by showing local growth in the area where they are located. Although they can originate fromany skeletal muscle, they are most commonly seen on the anterior abdominal wall. Various treatment options are available depending on the clinical condition of the patient. We aimed to discuss the diagnosis and treatment of DF with a 55-year-old male patient who has DF and applied to our clinic.

Keywords: mesenchymal tumor, fibrous type, local growth

INTRODUCTION

Desmoid-type fibromatosis (DF) is a rare, locally infiltrative, mesenchymal neoplasm associated with local recurrence but without the potential for metastasis. DF accounts for 0.03% of all neoplasms and 3% of soft tissue tumors. It occurs most commonly between the ages of 10 and 40 and is the most common cause of anterior abdominal wall mass in young women of childbearing age (1). DF tends to develop in surgical scarring sites, especially after cesarean sections and intra-abdominal resections (2). Trauma, pregnancy, and the use of oral contraceptives play a role in etiopathogenesis (3). However, it can affect almost any part of the body, including the extremities, head and neck, trunk, and abdominal cavity. Although the vast majority of these tumors are sporadic, DF can also be hereditary (1). Familial DF develops predominantly in patients with familial adenomatosis polyposis (FAP). The risk of developing DF in patients with FAP is 1,000 times greater (2). However, in about 90 percent of cases, no cause can be found. In sporadic cases, it is characterized by a mutation in the CTNNB1 (cadherin-associated protein beta 1) gene. In hereditary cases, 5–15% are associated with the APC adenomatous polyposis coli gene mutation. There may be an abnormal accumulation of β-catenin within the cell in both hereditary and sporadic cases (4). Granular nuclear expression of catechins is found in approximately 80% of neoplasms (1).

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The diagnosis is made by histopathological evaluation. Depending on the location of the tumor, ultrasound, computed tomography, and magnetic resonance are the main imaging methods (4).

Histopathologically, DF is usually infiltrative and consists of sweeping fascicles of uniform myofibroblasts within a dense collagenous stroma. The histological differential diagnosis ranges from scar formation to abdominal gastrointestinal stromal tumors and "low-grade liposarcoma" to a wide range of spindle cell lesions (1).

Clinical presentation ranges from asymptomatic to pain, paresthesia, and intestinal obstruction. DF treatment includes combinations of surgery, radiation therapy, and systemic therapy (5).

CASE

A 55 year old male patient, with no known extracomorbidity, underwent surgery for a mass in his small intestine as a result of the examinations conducted in an external center. He had complaints of abdominal pain, nausea, green-colored vomiting, and weight loss of 20 kg in the last two months. No remarkable pathological finding revealed in laboratory tests. When we questioned the family history of the patient, it was learned that his father had died of lung cancer. The patient's upper gastrointestinal endoscopy and colonoscopy were negative for malignancies. A large number of polyps, measuring 11x7 mm in size, were observed in the gallbladder lumen in the entire abdominal USG of the patient. Free fluid was detected in the pelvis in the form of smearing.

A contrast-enhanced CT of the upper abdomen showed a mass in the distal jejunum, which was primarily evaluated for carcinoma, and a pleural effusion in the right abdomen. During the surgery for excision of the mass, a white solid lesion (3.5x2.5x3.5 cm) was observed. There was no direct relationship between the lesion and the intestinal lumen. In the pathological examination of the lesion material, beta-catenin (+), desmin (-), SMA (weak +), CD117 (-), CD34 (-), DOG-1 (-), and Ki67 (proliferation index up to 3%) were found. Because the patient's symptoms persisted, Pet CT was performed. 3 lesions (SUVmax = 5.0), the largest of which was 5.5 cm in diameter were detected. Additionally, increased FDG uptake in the serosal peritoneal surfaces of the were seen in PET CT. The effusion accompanied by minimal FDG uptakes in the pelvis was thought to be benign.

Diffuse and linear atelectatic changes in both lungs were reported as pleural thickening, mostly in the field, on a thorax CT performed as the patient's complaints continued. The patient was operated because of a large lesion caused ileus in the left lower quadrant, which seen on CT. As a result of the examination of the pathological sample taken during the operation, a white solid lesion (3.5x2.5x3.5 cm) was observed when the serosa was sectioned. In the subsequent comparative abdominal CT, no mass causing ileus was observed in the left lower quadrant, but stable solid masses, the larger ones in the left upper quadrant, were observed in the mesentery. Tamoxifen (2x20 mg) was prescribed to the patient whose pathological diagnosis was desmoid-type fibromatosis.

DISCUSSION

DF accounts for 0.03% of all neoplasms and 3% of soft tissue tumors (6). The aannual incidence is reported to be 2-4 people per million. It was the first DF case that we have seen in our 13 years of clinical experience in oncology. Although most DF are sporadic, incidences related to familial adenomatous polyposis (FAP) were reported to range between 7.5% and 16% (7). About 85 to 95% of sporadic DFs are characterized by activating mutations in the exon 3 of the CTNNB1 gene (8). According to various studies, DF most commonly occurs between the third and fourth decades of life, where both sexes are almost equally affected (6,8). Some studies indicate that 9-16 year olds are affected (9). Our patient was much older than cases who are reported in the literature. Abdominal type is the most common type, with up to 37% to 50% reported in various publications (10). Our case was an abdominal DF.

One of the effective treatment method for abdominal desmoid tumors is resection and abdominal reconstruction. Some studies show that the average period of local nuclei development for patients with desmoid fibromatosis diagnosis in abdominal wall with

negative surgical borders is 83.4 months and 13.1 months for patients with positive surgical borders. Radiotherapy can provide 75-80% local control in large populations or in cases where the surgical borders cannot be macroscopically clear (11). Studies are available for systemic treatment options in patients where negative surgical borders are not met. Our patient also needed additional systemic treatment due to the residual tumor. Pharmacological treatment of desmoid tumors includes non-steroidal anti-inflammatory drugs (NSAIDs), such as methotrexate, and low-dose chemotherapeutic drugs such as vinblastine and vinorelbine. In addition, any of the tyrosine kinase inhibitors (TKI) can be used, including imatinib, sunitinib, and sorafenib (12).

In the study made by van Maren et al. both pazopanib and sorafenib were found effective. Liposomal doxorubicin is also recommended for the treatment of abdominal desmoid tumors (9). Systemic treatment options include antihormonal treatment, regardless of the presence of ER/PR positivity. Antihormonal agents such as Tamoxifen can be used as first-line treatment alone or in combination with NSAIDs, with limited toxicity, rare side effects, and low costs (13,14). That's why we chose the tamoxifen treatment in our case and we are still monitoring the patient in remission with tamoxifen. Bini et al. (15) reported that the disease is very rare and may occur spontaaneously. van Maren et al. (9) used a combination of vinorelbine, methotrexate, and thalidomide, doxorubicin. Response rates of 19.2-40% have been reported (9). We think we result is important for our patient to guide further treatment steps.

CONCLUSION

As a result, it is difficult to establish a standard treatment regimen for DF. Some studies suggest that various treatment options, such as surgery, radiotherapy, and systemic treatment can be used. Although the disease does not cause metastases, it can affect the quality of life of the individual. In these patients, a treatment decision must be made with a multidisciplinary approach and consideration of the literature.

Ethical approval

Written informed consent was obtained from the participants.

Author contribution

Surgical and Medical Practices: NK; Concept: NK; Design: NK, EB; Data Collection or Processing: EB, YEE, MD; Analysis or Interpretation: NK, EB; Literature Search: EB; Writing: EB, YEE, MD. All authors reviewed the results and approved the final version of the article.

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Conflict of interest

The authors declare that there is no conflict of interest.

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