Case report

Abdominal wall desmoid tumors: Three case reports

Abstract

Desmoid tumors, also known as aggressive fibromatosis, deep fibromatosis, or musculoaponeurotic fibromatosis, are rare, locally aggressive tumors arising from connective tissue within musculoaponeurotic structures. These tumors, though benign and non-metastatic, are notorious for their local invasiveness, leading to significant morbidity and occasional mortality. They have an incidence rate of approximately 2-4 cases per million individuals and represent about 0.03% of all neoplasms.

Desmoid tumors most commonly present in individuals aged 30 to 40 years and are more prevalent in females than in males. They can occur in various locations, including the abdominal wall, intra-abdominal regions, and extra-abdominal areas. Approximately 5% to 10% of cases are associated with familial adenomatous polyposis (FAP).

Imaging modalities such as CT scans and MRI are essential for diagnosing desmoid tumors, as they help determine the extent of the tumor, its relation to adjacent structures, and inform surgical planning. Optimal treatment typically involves surgical excision with the goal of achieving negative margins to reduce the risk of recurrence and enhance oncological outcomes.

In our study, we report on three female patients over the age of 40 with desmoid tumors and diagnosis was confirmed through CT imaging. Surgical excision was performed with clear margins in all cases, and no recurrences were observed.

Key words

Desmoid tumor, Desmoid fibromatosis, Familial adenomatous polyposis, Gardner syndrome

Introduction

Desmoid tumors, or aggressive fibromatosis, are rare connective tissue tumors that do not metastasize but can be locally aggressive. They are categorized into three subtypes: extra-abdominal, intra-abdominal, and abdominal, the latter often confused with gastrointestinal stromal tumors (GISTs). [1,2,4]

Extra-abdominal desmoid tumors can appear in various locations but are most frequently found in the shoulder girdle, chest wall, trunk, and lower extremities. Intra-abdominal desmoids typically develop in the mesentery or retroperitoneum and are often associated with Gardner syndrome. Abdominal desmoid tumors generally arise from the musculoaponeurotic structures of the abdominal wall, particularly from the rectus abdominis and internal oblique muscles. Less frequently, they originate from the external oblique muscle and transversalis fascia. [1,3,5]

These tumors are most commonly seen in young women, especially during or after pregnancy. Estrogen is known to stimulate fibroblast proliferation in these tumors, which may regress

post-menopause. There is a strong association between abdominal desmoid tumors and prior abdominal or pelvic surgery, trauma, estrogen therapy, familial adenomatous polyposis (FAP), and Gardner syndrome. [1,2,3]

Clinical Presentation: Abdominal wall desmoid tumors typically present as a mass or swelling, ranging from 5 to 15 cm in diameter. On gross examination, these masses have a firm, gritty texture, a glistening white appearance, and resemble scar tissue. They appear as well-circumscribed swellings without a distinct capsule. [4,5]

Histology: Desmoid tumors consist of elongated fibroblasts and myofibroblasts. Ultrasonographically, they appear as well-defined lesions with irregular borders. On CT or MRI, they can be homogeneous or heterogeneous and may present as hypodense, isodense, or hyperdense relative to muscle tissue. Histopathological analysis is essential for definitive diagnosis. [5,6]

Imaging: Ultrasonography, CT, and MRI are critical in diagnosing, staging, and monitoring desmoid tumors. Ultrasonography is particularly useful for defining tumors in the abdominal wall, chest, and extremities, where desmoids appear as well-defined, heterogeneous solid masses with variable echogenicity based on collagenous, fibrotic, and cellular content. CT is the primary modality for intra-abdominal lesions, which typically present as well-defined soft tissue masses with varying densities. MRI provides excellent soft tissue resolution and is especially useful for evaluating extra-abdominal lesions in the extremities, chest wall, and abdominal wall. Recurrence Rates: Abdominal wall desmoid tumors have a relatively low recurrence rate of 20% to 30%. [2,4,5]

Case Reports

Case I - A 20-year-old female patient presented to our center on July 9, 2019, with a painless mass located on the left side of the umbilicus, extending into the left lumbar and iliac fossa regions. The mass had been gradually enlarging over the previous 12 months. The patient had no history of abdominal surgery or trauma.

Clinical Examination: On physical examination, a single large mass was observed in the left lumbar, umbilical, and left iliac regions. The mass, which was fixed to the anterior abdominal wall, measured 8x6x6 cm. It was non-tender, rounded, with a smooth surface, and had a firm consistency.

Imaging Studies: Ultrasonography revealed a large, solid, heterogeneous hypoechoic mass located in the left lumbar and left iliac fossa. A subsequent CT scan confirmed the presence of a large mass originating from the left rectus abdominis muscle, extending from the left lumbar to the left iliac fossa, without crossing the midline. The mass measured 8x6x6 cm and was superficial to the left rectus abdominis muscle, with no signs of enlarged lymph nodes or ascites. These findings were indicative of a benign desmoid tumor arising from the anterior abdominal wall.

Preoperative Workup: Laboratory investigations were normal. The patient was scheduled for surgery following a comprehensive preoperative assessment. Surgical Intervention: Under general anesthesia, a complete excision of the tumor was performed, ensuring a 2 cm margin. The mass was located anterior to the rectus abdominis muscle and did not involve the peritoneum. The left rectus muscle remained intact and did not require mesh repair. The abdomen was not opened during the procedure. A vacuum suction drain was placed beneath the subcutaneous tissue, and the skin was closed. The excised mass weighed 350 grams and exhibited a gristly texture with a glistening white appearance on cut section.

Histopathology: Histopathological examination confirmed the diagnosis of a desmoid tumor with negative surgical margins.

Postoperative Course: The patient's recovery was uneventful, and she was discharged on the 10th postoperative day. After a follow-up period of five years, there was no evidence of recurrence or incisional hernia. **[Fig 1-6]**

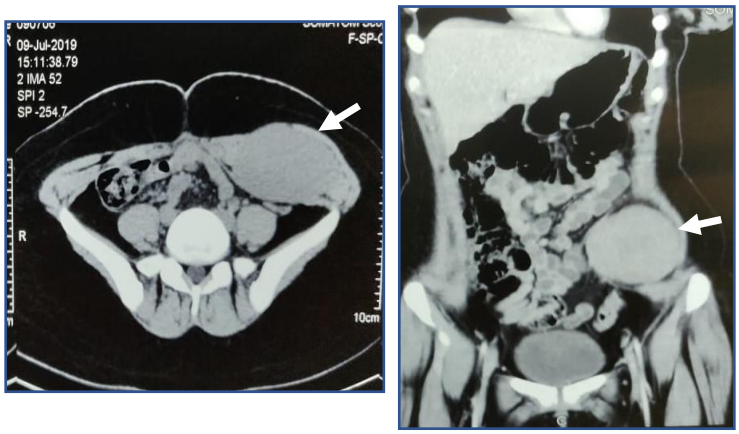

Fig-1 CT Abdomen Photograph showing a mass occupying left rectus sheath

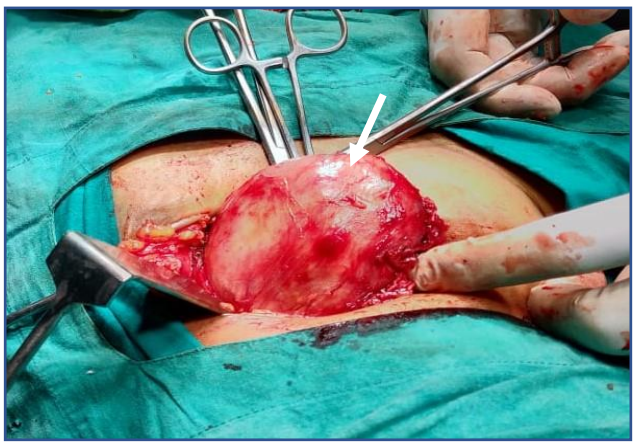

Fig-2 Intraoperative photograph showing a rounded solid mass

Fig-3 Intraoperative photograph showing a rounded solid mass

Fig-4 Intraoperative photograph showing a excision of rounded solid mass

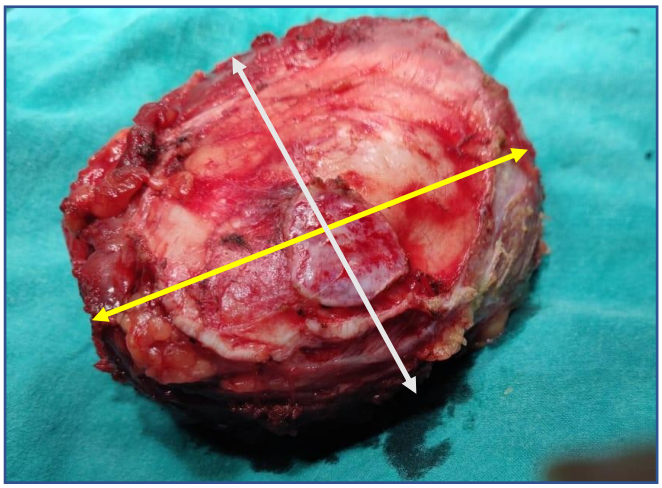

Fig-5 On gross, rounded mass measuring 8x6x6 cm & weighing 350 grams

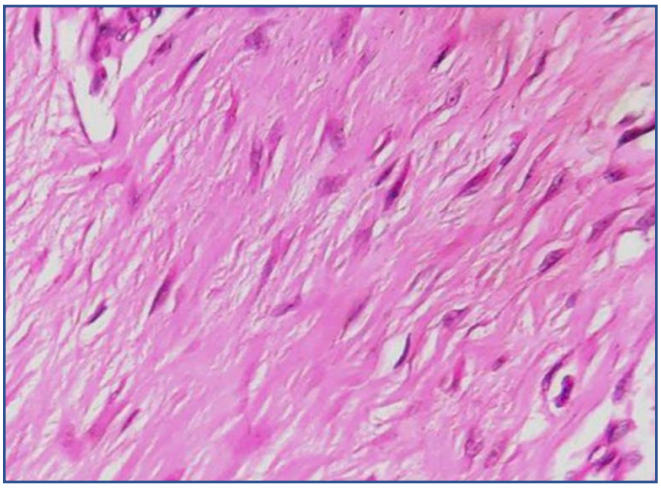

Fig-6 Microphotograph showing the tumor cells are spindle shaped with homogeneous collagenous frame (HE, x200). *HE:* hematoxylin and eosin.

Case II

A 45-year-old female was admitted to our center on October 12, 2015, with a complaint of a painless lump in the anterior abdominal wall, located below the umbilicus. Physical examination revealed a tough, fixed mass crossing the midline and situated below the umbilicus. Routine blood tests were normal.

Imaging Studies: Ultrasonography showed a solid mass with heterogeneous echogenicity involving the left rectus abdominis muscle and extending across the midline. CT imaging confirmed a well-circumscribed, large mass measuring 15x8x6 cm, with an attenuation similar to that of muscle tissue. The mass originated from the left rectus abdominis muscle and extended across the midline to the right side. There was no evidence of abdominal ascites or lymphadenopathy.

Surgical Intervention: The patient underwent a complete surgical excision of the tumor with a 2 cm margin. The anterior abdominal wall was involved, but the peritoneum remained intact. The left rectus abdominis muscle and the posterior wall of the rectus muscle were not compromised, so no mesh repair was required. The excised mass, weighing 750 grams, displayed a gritty texture and a glistening white appearance on cut section. Histopathology: Histopathological examination confirmed the diagnosis of a desmoid tumor with negative surgical margins.

Postoperative Course: The patient's recovery was uneventful, and she was discharged on the 10th postoperative day. After five years of follow-up, there was no recurrence or development of an incisional hernia**. [Fig 7-12]**

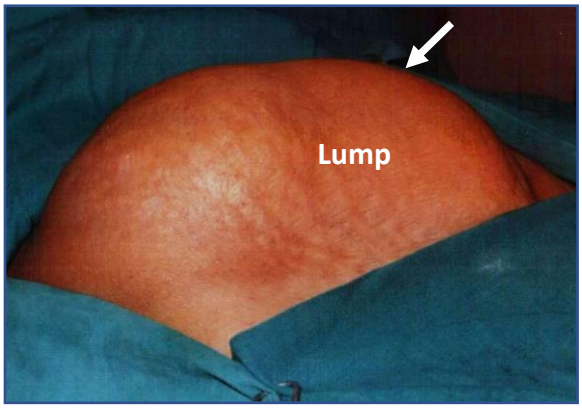

Fig-7 Photograph showing anterior abdominal lump crossing the midline

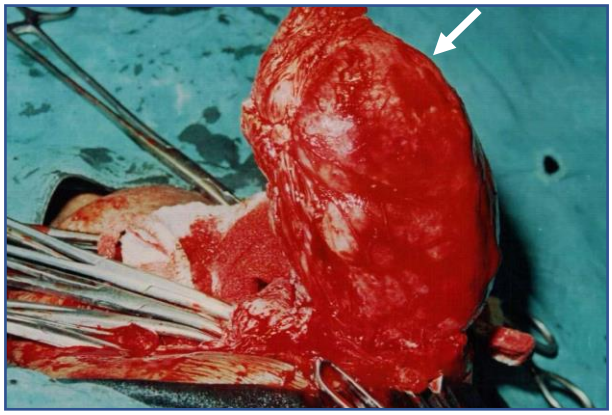

Fig-8 Intraoperative Photograph showing a rounded mass arising from anterior abdominal wall

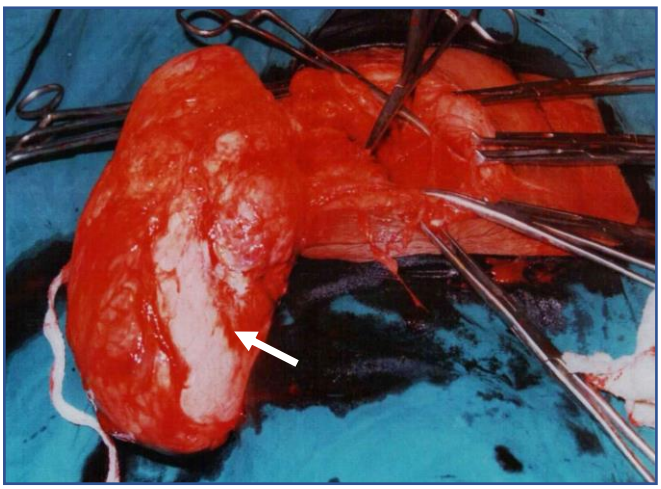

Fig-9 Intraoperative photograph showing rounded mass arising from left rectus abdominis muscles

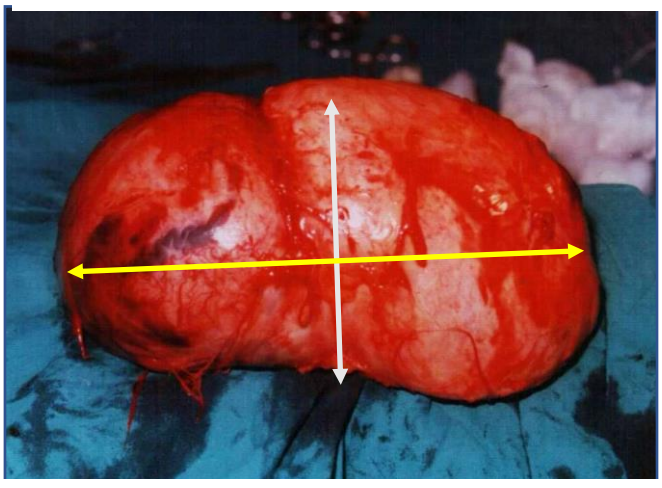

Fig-10 On gross solid mass measuring 15x8x6 cm and weighing 750 grams

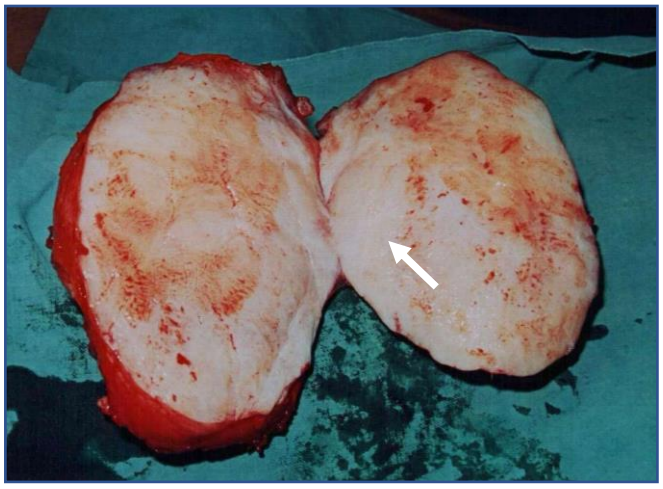

Fig-11 On gross specimen glistening white appearance on cut section

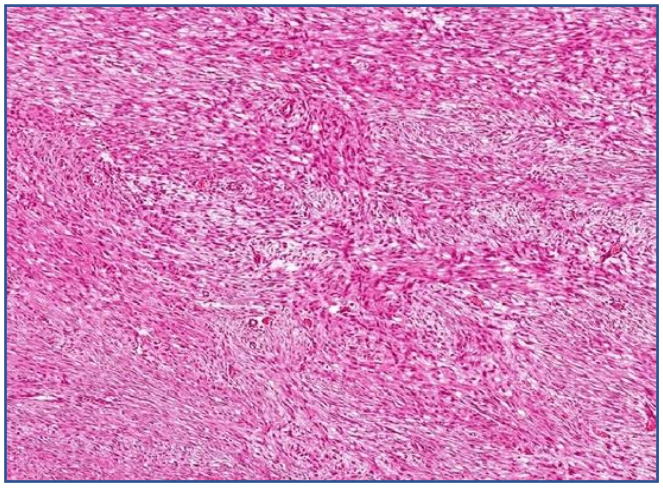

Fig-12 Histopathological examination showing spindle cells, features are consistent with desmoid-type fibromatosis.

Case III

A 50-year-old female was admitted to our center on February 1, 2017, with a complaint of a painless mass on the left side of her lower abdomen, present for one year. Physical examination revealed a firm, non-tender mass fixed to the anterior abdominal wall. The mass had been gradually increasing in size. Blood tests were within normal limits. The patient had a history of abdominal tubectomy performed 20 years earlier.

Imaging Studies: Ultrasonography showed a large, heterogeneous mass with smooth, sharply defined margins located in the left lower anterolateral abdominal wall. CT scan confirmed a well-circumscribed mass measuring 8x6x6 cm, with attenuation similar to muscle tissue. The mass originated from the left rectus abdominis muscle.

Surgical Intervention: Following a preoperative workup, the patient underwent surgery, which involved a complete wide excision of the tumor with a 2 cm margin. The mass was fixed to the anterior wall of the rectus abdominis muscle. Both the anterior and posterior walls of the rectus muscle were preserved, and the peritoneum was not involved. The ovoid mass was restricted to the anterior wall of the rectus abdominis muscle, allowing for primary suturing without the need for mesh or flaps.

Postoperative Course: The patient's recovery was uneventful, and she was discharged on the 8th postoperative day. Histopathological examination confirmed a spindle-cell tumor with negative margins. After five years of follow-up, there was no evidence of recurrence**. [Fig 13-18]**

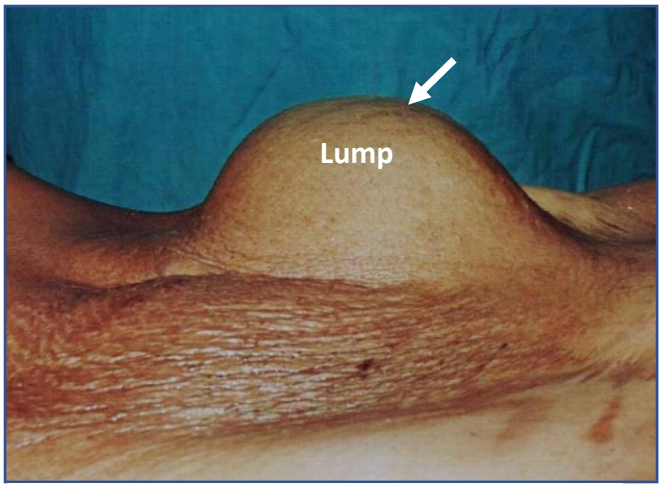

Fig-13 Photograph showing left anterior abdominal lump

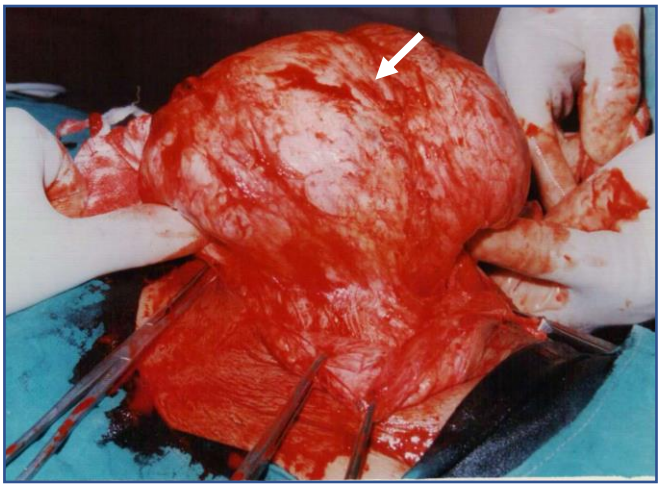

Fig-14 Intraoperative Photograph showing rounded and solid mass

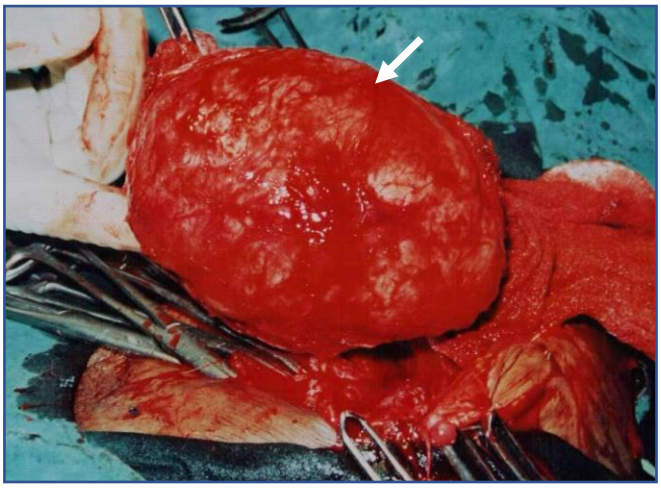

Fig-15 Intraoperative photographs showing excision of a rounded solid mass

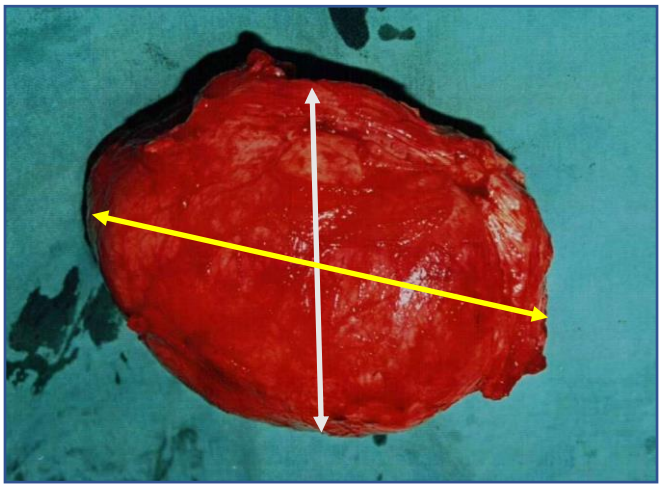

Fig-16 on gross rounded solid mass measuring of size 8x6x6 cm

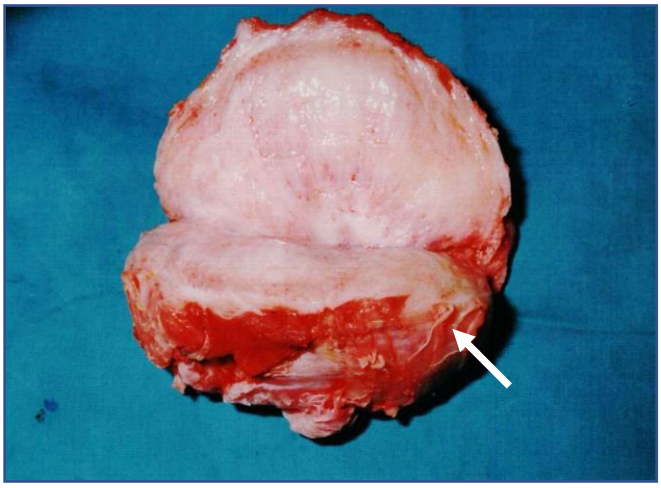

Fig-17 On gross specimen glistening white appearance on cut section

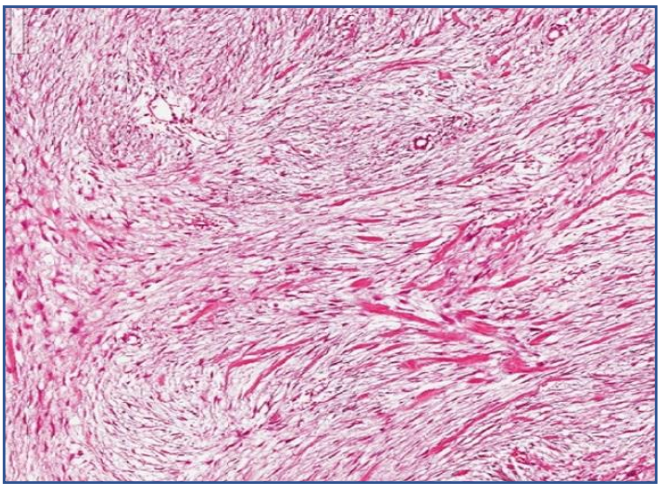

Fig-18 Histopathological examination showing spindle cells, features are consistent with desmoid-type fibromatosis.

Discussion and review

Desmoid tumors, also known as aggressive fibromatosis, are rare, benign tumors that originate from musculoaponeurotic connective tissue. First described by Macfarlane in 1832, the term "desmoid" was introduced by Müller in 1938, derived from the Greek word "Desmos," meaning "band" or "tendon-like." Desmoid tumors account for approximately 0.03% of all neoplasms and 3% of soft tissue tumors, with an annual incidence of 2-4 cases per million people. Although their exact etiology is unknown, factors such as pregnancy, estrogen use, trauma, abdominal and pelvic surgery, familial adenomatous polyposis syndrome, and Gardner syndrome are considered significant risk factors. [1,2,3]

Desmoid tumors can arise from any muscle tissue in the body, but they most commonly develop in the anterior abdominal wall and shoulder. They are particularly prevalent in the rectus abdominis muscle, with rates reported between 37% and 50% in various studies. These tumors are composed of spindle cells or myofibroblasts, with a female-to-male ratio of 2:1 and can also occur in childhood.

Differential Diagnosis: The differential diagnosis for desmoid tumors includes acute hematoma, lymphosarcoma, leiomyosarcoma, fibrosarcoma, rhabdomyosarcoma, and neurofibromas. Desmoid tumors of the anterior abdominal wall have a significantly lower recurrence rate of 20% to 30%, with no mortality observed in our study and a 5-year survival rate of 95%. [4,6,7]

Management and Treatment:

1. **Surgery**: The primary treatment for desmoid tumors is wide surgical resection aiming to achieve histologically negative margins. This can be challenging, especially for tumors in the rectus abdominis muscle, and may require abdominal wall reconstruction using sutures, myocutaneous flaps, or synthetic mesh. Polypropylene mesh is commonly used for reconstruction. Desmoid tumors in the abdominal wall have a recurrence rate of 20% to 30%.

2. **Active Surveillance (Watchful Waiting)**: For small, slow-growing, painless tumors, particularly in menopausal women where spontaneous regression may occur, a "wait and see" approach can be appropriate. Asymptomatic desmoid tumors might be managed conservatively. [2,3]

3. **Targeted Therapy**: This includes the use of tyrosine kinase inhibitors (TKIs) that target specific genetic pathways involved in tumor growth.

4. **Radiotherapy**: Useful for unresectable or recurrent tumors, radiotherapy may also be used as an adjuvant treatment post-surgery. A recent meta-analysis suggests that combining surgery with radiotherapy can reduce local recurrence.

5. **Cryoablation**: For patients where surgery is not an option, cryoablation involves using extreme cold to destroy tumor cells. Radiofrequency ablation can also be used, applying extreme heat to achieve the same effect, particularly for smaller tumors. [1,2]

6. **Chemotherapy**: Both oral and intravenous chemotherapy drugs can be used. Methotrexate and vinblastine regimens are generally well-tolerated and effective. Selective tyrosine kinase inhibitors like imatinib, sunitinib, and pazopanib have shown promise in stabilizing disease.

7. **Hormone Therapy**: Tamoxifen, an anti-estrogenic drug, is often used alongside nonsteroidal anti-inflammatory drugs (NSAIDs) such as indomethacin, sulindac, and celecoxib.

8. **Systemic Therapy**: Includes cytotoxic chemotherapy agents like doxorubicin and tyrosine kinase inhibitors such as imatinib. Doxorubicin-based regimens have shown the highest success rates and durable responses in many patients. [1,2,3,5]

Conclusion

Desmoid tumors are benign tumor arising from the connective tissue. Desmoid tumors are locally aggressive and wide surgical resection with negative margins is the goal of surgery. Far abdominal wall desmoid tumors, side local extension followed by reconstruction of the defect with polypropylene mesh to achieve excellent functional results.

Reference

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