

## AGGRESSIVE FIBROMATOSIS WITH BONE INVOLVEMENT- A CASE REPORT



### Orthopaedics

**KEYWORDS:** aggressive fibromatosis, aggressive fibromatosis with bone involvement.

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### ABSTRACT

Aggressive fibromatosis is a condition marked by the presence of fibroblasts, which are benign, slow-growing musculo-aponeurotic tumours with no evidence of any metastatic potential<sup>6</sup>. It is known to involve muscle, subcutaneous tissue and neurovascular structures. However, bone involvement is very rare<sup>1</sup>. We have presented the case report of a 13 year old boy with aggressive fibromatosis in the left gluteal region involving acetabulum, ilium and ischium. Clinical examination revealed a firm swelling about 9x7x3 centimetres in the lower outer quadrant of left gluteal region. Routine haematological and radiological investigations were performed. Complete blood count and ESR were normal. X-ray pelvis with both hips revealed an osteolytic lesion in the left ilium and ischium. Computed Tomography showed geographic, lytic lesions involving the supra acetabular region of left ilium and a part of ischium. CT scan guided FNAC showed degenerated osteocytes with occasional osteoclast giant cells with few inflammatory mononuclear cells. MRI showed a soft tissue mass adjacent to the left iliac wing. The differential diagnoses were chronic osteomyelitis, eosinophilic granuloma, cystic type of fibrous dysplasia. We did a wide local excision of the tumour with curettage of the surrounding bone. Histopathological examination of the specimen was suggestive of aggressive fibromatosis. In our two years of follow up the patient did not show any signs of recurrence. This case is being reported for its rarity since fibromatosis involving the bone is an unusual event.

### Introduction:

Aggressive fibromatosis is a locally infiltrative tumour that originates from fascial planes of soft tissues. Though it is characterised by an uncontrolled proliferation of the fibroblasts, it does not have any metastatic potential<sup>6</sup>. It usually involves muscle, subcutaneous tissue and neurovascular structures. Fibromatosis involving the bone is very unusual<sup>1</sup>. We have presented the case report of a 13 year old boy with aggressive fibromatosis in the left gluteal region involving ilium and ischium. The differential diagnoses were chronic osteomyelitis, eosinophilic granuloma, cystic type of fibrous dysplasia. We did a wide local excision of the tumour with curettage of the surrounding bone. Histopathological examination of the specimen was suggestive of aggressive fibromatosis. In our two years of follow up the patient did not show any sign of recurrence.

### Case Report:

#### History:

A 13 year old boy presented to us with the complaint of a painless swelling in the left buttock for a period of six months. Initially the swelling was smaller and it gradually increased in size. The patient gave a history of fall while playing, six months back. Following the injury he was walking with pain in the left buttock for a period of one month. After a short period of treatment with NSAIDs the pain disappeared, but the patient developed a swelling in the left buttock which was gradually increasing in size.

#### Clinical examination:

Examination revealed a globular swelling of about 9x7x3 centimetres in the lower outer quadrant of the left gluteal region. The swelling was smooth; firm in consistency; not pulsatile; not warm; not tender; not mobile; and the edges were clearly defined. There was no other swelling elsewhere. Skin over the swelling was of normal colour; not tense; pinchable and did not have any dilated veins. Clinical examination of spine and hip joints was normal. The neurological status was within normal limits.

#### Investigations:

Complete blood count and ESR were normal. X-ray pelvis with both hips showed an osteolytic lesion in the supra acetabular portion of left ilium.



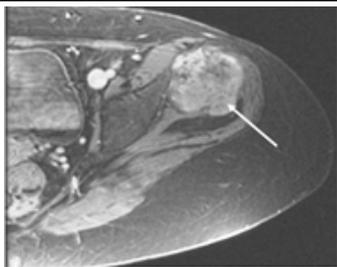
**Fig 1 : X-ray pelvis with both hips showing osteolytic lesion in the left ilium.**

Computed Tomogram of pelvis showed geographic, multifocal moth eaten lytic lesions involving supra acetabular portion of left iliac bone, quadrilateral plate and ischium. There was localised expansion of supra acetabular region with areas of cortical thickening and breakdown.



**Fig 2: CT scan showing osteolytic lesions in the supra acetabular region of left iliac bone.**

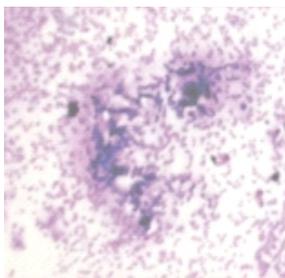
MRI showed a soft tissue mass of about 12x8x5 centimetres with heterogenous low signal intensity in the supra acetabular portion of left iliac bone and part of ischium. The MRI suggested benign soft tissue tumour and advised for tissue biopsy.



**Fig 3: Axial T1 weighted MRI showed a soft tissue mass adjacent to left ilium.**

With these findings our differential diagnoses were chronic osteomyelitis, eosinophilic granuloma and cystic type of fibrous dysplasia.

CT scan guided Fine Needle Aspiration Cytology was done. The smear showed degenerated osteocytes with occasional osteoclastic giant cells with few inflammatory mononuclear cells in a haemorrhagic background. No malignant cells were seen.



**Fig 4: FNAC showing osteocytes with occasional osteoclastic giant cell and inflammatory mononuclear cells.**

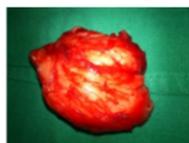
The cytological findings were inconclusive and we planned for an open biopsy.

**Management:**

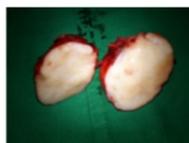
With the patient in right lateral position, under spinal anaesthesia, we did a wide local excision of the tumour through posterior approach. We found a smooth globular mass arising from the left gluteal muscles. It had a glistening surface and was firm in consistency. The tumour was removed in toto with a 3 centimetre margin of surrounding normal tissue. The tumour was found to infiltrate into the supra acetabular portion of left iliac bone and a part of ischium. The affected portions of bone were decorticated and curetted up to the level of fresh bleeding. The resected mass was measured to be about 13x9x5 centimetres. The tumour was bisected on table and was found to be a solid mass with its inner aspect greyish white in colour. It was later sent for histopathological examination.



**Fig 5**



**Fig 6**



**Fig 7**

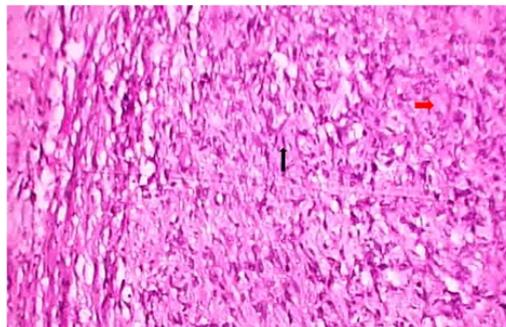
**Fig 5: Surgical exposure**  
**Fig 6: Gross appearance of the mass**  
**Fig 7: Cut section of the mass**



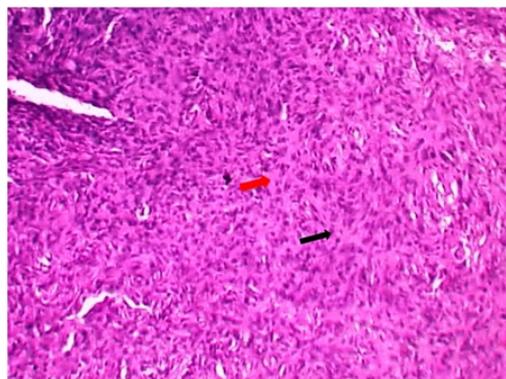
**Fig 8: Post operative X Ray**

**Histopathological examination:**

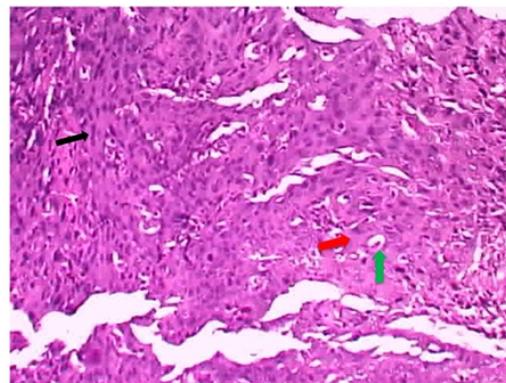
Biopsy showed fascicles of spindle shaped cells having scanty cytoplasm with elongated bipolar pointed basophilic nucleus. Periphery of the cytoplasm appeared to merge with the surrounding eosinophilic collagen. These findings were suggestive of Fibromatosis / Desmoid tumour.



**Fig 9: Irregular fascicles of spindle cells in a collagenised stroma (HE, X40)**



**Fig 10: Cellular area showing crowded spindle cells with scanty stroma (HE, X40)**



**Fig 11: Spindle cells infiltrating in to bony matrix (HE, X40)**

Black arrow : spindle cells  
Red arrow : collagen stroma  
Green arrow : osteocytes in the bone matrix

**Discussion:**

Fibromatosis / Desmoid tumour is a benign proliferative condition of fibroblasts. It is a slow-growing musculoaponeurotic tumour with no evidence of metastatic potential<sup>6</sup>. The term desmoid, coined by Muller in 1838, is derived from the Greek word *desmos*, which means tendon like. Desmoid tumors most commonly arise from the rectus abdominis muscle in postpartum women and in scars due to abdominal surgery. However they may arise from any skeletal muscle. They are believed to originate from mesenchymal stem cells. Although fixation to musculoaponeurotic structures is apparent, the

overlying skin is normal. The myofibroblast is the cell considered to be responsible for the development of desmoid tumors. Superficial desmoid tumors usually manifest themselves as a painless or slightly painful lump<sup>12,7</sup>. Most cases are sporadic, but some are associated with familial adenomatous polyposis (FAP). Trauma has been theorized to increase the risk of desmoid tumour occurrence. Antecedent trauma, often surgical, has been reported at the site of the desmoid tumour in approximately 25% of cases. There are several lines of evidence to support a role for estrogen in modulating the behaviour of desmoid tumour. An increased frequency rate is demonstrated during pregnancy and in females taking oral contraceptives.

Aggressive fibromatosis is a rare condition marked by the presence of desmoid tumors, which are locally aggressive and damage nearby structures. It is a relatively rare lesion, representing less than 3% of all soft tissue tumours with a reported annual incidence of 0.2-0.5/100000 population<sup>7</sup>. Histologically they resemble low-grade fibrosarcomas, but they are locally aggressive and tend to recur even after complete resection. There is an increased tendency to recurrence in the setting of prior surgery. Pathologists sometimes have difficulty in distinguishing these from fibrosarcomas<sup>6,7</sup>. These often appear as infiltrative, usually well-differentiated, firm overgrowths of fibrous tissue. The prefix "aggressive" describes the marked cellularity, aggressive local infiltrative behaviour and greater tendency to recurrence. The treatment of these relatively rare fibrous tumours is often challenging. Despite their benign nature, they can damage nearby structures causing organ dysfunction.

**Genetics and mechanism of tumor development:** Mutations in the *CTNNB1* gene or the *APC* gene cause fibromatosis. Both genes are involved in an important cell signalling pathway that controls the growth and division of cells and the process by which cells mature to carry out specific functions<sup>4,5</sup>. Mutations in the *APC* gene that cause fibromatosis lead to a short APC protein that is unable to interact with beta-catenin. As a result, beta-catenin is not broken down and, instead, accumulates in cells. Excess beta-catenin, whether caused by *CTNNB1* or *APC* gene mutations, promotes uncontrolled growth and division of cells, allowing the formation of fibromatosis.

**Investigations:** A biopsy is necessary to diagnose fibromatosis<sup>4,5</sup>. Ultrasound is often the first method of examination of a soft tissue tumour. If the mass is solid or firm, a CT and /or MRI scan is used to determine whether it adheres to nearby structures and whether it can be safely removed. A variety of options are available to obtain biopsy from a suspicious swelling:

Core needle biopsy	- takes a small piece of tissue, usually 1 mm wide
Incision biopsy	- takes a portion of the tumour
Excision biopsy	- removal of the visible tumour in toto.

**Surgical management:** The goal of surgery is to remove the tumour as a whole and minimize the risk of recurrence<sup>8,2,1</sup>. However Fibromatosis has a high rate of recurrence with surgery alone. About 25 to 40 percent of patients who undergo surgery alone, have a local recurrence, that is, return of the fibromatosis at or near the original site<sup>8,9</sup>.

**Radiation therapy:** Radiation therapy is an effective option for many patients who cannot have surgery, or as an adjunct to surgery or chemotherapy<sup>9</sup>. The duration of radiation therapy usually is 6 to 8 weeks. Radiological evidence of tumour shrinkage may take months to years to become apparent. Local control is often achieved at a considerable cost, due to significant treatment-related morbidity from the high doses of external radiation therapy (>50 Gy) or the disfiguring surgical procedures that are often required. In addition, despite the use of surgery and radiation therapy 20–36% of patients will show local recurrence<sup>9</sup>.

**Medical Treatment:** Although it is argued that chemotherapy is not effective in benign tumors with low mitotic rates, cases of aggressive fibromatosis successfully treated with cytotoxic and non-cytotoxic chemotherapy are being reported. *Sze et al.* report significant tumor shrinkage with low dose methotrexate and vinblastine. Another trial concluded that pegylated liposomal doxorubicin leads to tumor shrinkage and partial remission in some cases; or at least stops proliferation, leading to a stable disease state. Meloxicam, a COX-2 inhibitor, has also been shown to be effective in controlling extra-abdominal aggressive fibromatosis. Sarcoma Alliance for Research through Collaboration (SARC) initiated a multi-center phase II trial on the efficacy of imatinib in aggressive fibromatosis and estimated progression-free survival was 94% and 88%, for two and four months respectively; whereas one year progression-free survival was 66%.

The use of NSAIDs in aggressive fibromatosis is based on the concept that endogenous prostaglandin synthesis plays a role in neoplastic growth and that prostaglandin inhibitors can control the growth of experimental tumors<sup>10</sup>. NSAIDs such as indomethacin and sulindac (a long-acting analog of indomethacin) are commonly used. There is also a suggestion that IFN- can be used as an adjuvant following surgical resection of aggressive fibromatosis to prevent recurrence<sup>10</sup>.

Recently radio frequency ablation has also been advocated by *Ilaslan et al.*, with no recurrence noted after a mean follow-up of 30 months. Early data from the Mayo Clinic, USA shows that percutaneous cryoablation may provide an alternative treatment for small to middle sized tumours. However, both modalities have been recently tried on a limited number of patients and it is too early to comment on their application.

#### Summary:

Our patient, a 13 year old boy developed a painless lump in the left gluteal region following a trauma. Clinico Radiological examinations identified it as a soft tissue mass with involvement of the underlying bone. The tumour was removed in toto with a 3 centimetre margin of surrounding normal tissue. The affected portions of bone were decorticated and curetted upto the level of fresh bleeding. Histopathology confirmed it as a fibroblastic proliferation with infiltration in to bony matrix. Considering the hypercellularity and locally aggressive behaviour of the tumour, it was diagnosed as aggressive fibromatosis. The main treatment modality of aggressive fibromatosis is wide surgical excision of the lesion except in areas where preservation of vital structures and their function may impede this objective. In such cases a multi-modality management strategy is usually employed. Surgery is usually combined with radiation therapy for control of residual disease and to prevent recurrence. Pharmacological therapy can be considered for unresectable disease or in cases where surgical and radiation therapy may lead to significant morbidity. Since we achieved complete excision of the tumour with a 3 cm margin of normal tissue and adequate curettage of the involved bone, we decided that there is no need for post operative chemotherapy and radiotherapy. In our two years of follow up the patient did not show any clinical or radiological signs of recurrence.



**Fig 12 A& B: After 2 years post operative follow up X ray**



**Fig 13: After 2 years clinical follow up photo**

#### **Conclusion:**

Since aggressive fibromatosis with bone involvement is extremely rare, no guidelines are currently available on the indications and extent of each modality of treatment. Due to the aggressive behaviour and tendency to invade local structures and recur, a multi-modality management strategy is usually employed. But in our patient we used wide surgical excision with curettage of the involved bone as a single modality treatment and did not find any recurrence in two years of follow up. Therefore we suggest that wherever complete excision of the tumour is possible without compromising the function, surgery alone may be used as a single modality treatment successfully. However where functional preservation and aesthetics need to be taken into account, a multi-modality treatment plan involving chemotherapy and radiation therapy can be considered for its management.

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