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Familial aggressive fibromatosis of the lower extremities

A report of two cases

Aggressive fibromatosis occurred in a father and his son. Both are alive without signs of recurrence 3 and 10 years after below-knee amputation and exarticulation of the hip joint, respectively. When possible, this tumour should be excised radically.

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Aggressive fibromatosis is a rare disease which presents diagnostic as well as therapeutic problems. Among these is the striking feature of disproportion between the apparently innocent histology, and the clinical manifestation of rapid invasive growth. A perusal of the literature has failed to demonstrate familial occurrence of the disease located in the extremities.

Case report

Case 1 (father). From age 10, he suffered from a posttraumatic equinovarus deformity of the right foot with recurrent ulcerations. After 10 years, in 1941, a subtalar arthrodesis and an elongation of the Achilles tendon were performed. The tendency to ulcer formation, however, remained unchanged. Because of osteitis of the fifth metatarsal bone, an excision was performed, followed by uneventful healing. Two years later, recurrent ulcerations appeared opposite the heads of the first and fourth metatarsal bones. When admitted to our department in 1981, at the age of 60 years, he had observed a rapidly growing tumour on the back of the foot for 5 months.

The patient presented a flattened polypous tumour with a cauliflower-like surface, 10 × 15 cm in size, extending from the proximal part of the back of the foot to the toes excluding only the nail of the big toe and the pulps of the other toes (Figure 1). The lower leg showed brownish pigmentation, the muscles were fibrotic, and sensibility was reduced over the distal third. No arterial pulsation could be felt at the ankle, probably because of local oedema. The skin temperature was normal. Radiography revealed periosteal ossifications throughout the whole length of the lower leg, possibly secondary to vascular disturbances. The bones of the foot were practically unrecognizable. Below-knee amputation was carried out and the patient was provided with a PTB prosthesis. No signs of recurrence were found 3 years postoperatively. Histologic examination of the tumour tissue was consistent with aggressive fibromatosis.

Case 2 (son). This patient was hospitalized for the first time in 1971 at 11 years of age, because of a 4 × 5 cm tumour of the right popliteal region, first noticed 6 months previously. He had slight local pain and limited knee extension. At operation the tumour was found to extend from the medial head of the gastrocnemic muscle down to the interosseous membrane and the insertion of the soleus muscle. Macroscopically the tumour was radically removed. Histologic examination of tumour tissue showed aggressive fibromatosis. In the course of the following year a progressive talipes equinus developed, together with a 20 degree extension defect of the knee. An elongation of the Achilles tendon was performed.

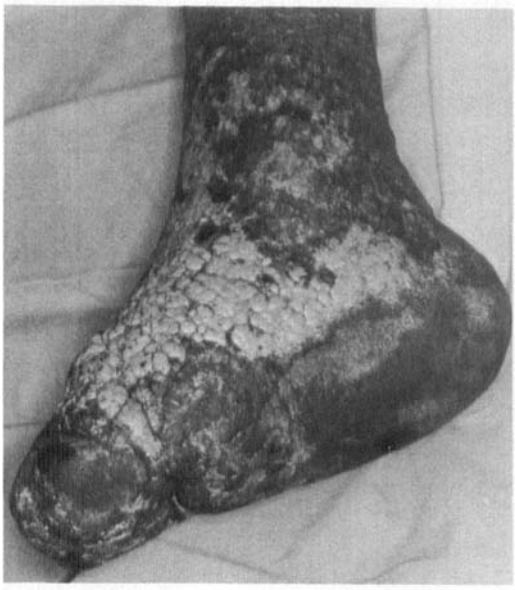
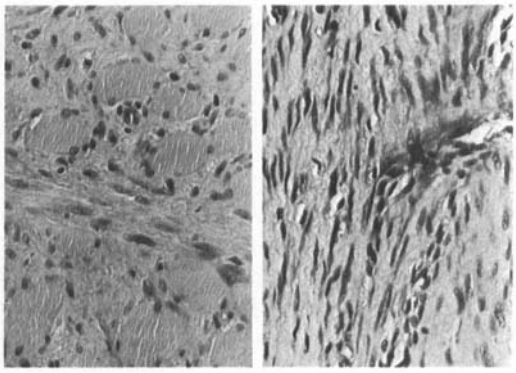


Figure 1. Aggressive fibromatosis of the right foot (Case 1)



A B

Figure 2. A. Aggressive fibromatosis of the right thigh infiltrating muscle cells (Case 2), (H&E, ×1000). B. A more cellular part presenting bundles of well-differentiated fibroblastic tumour cells with enhanced vascularity, (H&E, ×1000).

When the patient was 14, an exploratory operation was performed because of a local recurrence. The sciatic nerve was totally enveloped by tumour tissue reaching the proximal half of the femur. The patient was then transferred to our department. Arteriography revealed total occlusion of the femoral artery in the adductor canal. Radical excision was attempted, but, as pathologic changes were found in the adductor muscle and its origin from the tuber ischii, an exarticulation of the hip joint was done with radical removal of all pathologic tissue. The patient was provided with a prosthesis and had no signs of recurrence 10 years after surgery. Histologic examination of the tumour tissue revealed no signs of malignancy, the diagnosis being consistent with aggressive fibromatosis (Figure 2). Tumour invasion of the femoral artery and the perineurium of the sciatic nerve could be demonstrated.

Discussion

Enzinger & Weiss (1983) have presented the most comprehensive analysis of aggressive fibromatosis which is frequently located in the shoulder girdle and thorax, the supraclavicular region being the area most commonly involved (Masson & Soule 1966). The tumour is rarely seen in children; the peak incidence is around the age of 30 years, with a slight preponderance in the male sex.

The macroscopic appearance of the tumour is that of an ill-defined, grayish-white, firm mass with a tendency to infiltrate surrounding muscles, connective tissue and adipose tissue. It may encase vascular and neural structures without apparent invasion, although this may occur. McDougall & McGarrity (1979) have reported the only two cases of bone destruction, affecting the upper arm and the foot, respectively. Histologically, the tumour is composed of proliferative fibroblasts within a collagenous component. Mitoses are rare and never atypical. The infiltrating surface of the tumour induces atrophy of neighbouring striated muscle fibres.

The pathogenesis remains unexplained. Trauma (surgical trauma?) and endocrine factors might be of etiologic significance (Das Gupta et al. 1969). Enzinger & Weiss (1983) have reported several cases of desmoid tumours in scars and previously irradiated areas. A survey of the literature has revealed only one case of familial occurrence of desmoid tumours (a mother and two of her seven children) not related to Gardner's syndrome; none of the tumours presented in the extremities (Zayid & Dihmis 1969). We found no symptoms of intestinal polyposis or other manifestations of Gardner's syndrome, in either our two patients or in their family.

Because of the 25–65 percent tendency of the tumour to recur *in loco,* radical excision is important. In advanced cases or after recurrence, ablative surgery may be necessary. In Case 1, an amputation was performed as there was neither any function of the foot nor any possibility of weightbearing. In Case 2, the extent of the tumour made amputation necessary.

References

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