Acta Orthop. Scand. *58*, 289–291, 1987

Fracture in progressive ossifying fibrodysplasia

A case report

Progressive ossifying fibrodysplasia is a rare genetic disorder of connective tissue. A 6-year-old boy sustained a fracture of the humerus and afterwards of the femur. The former fracture was treated closed and the latter with internal fixation. Both fractures healed with severe restriction of joint motion.

Jacobo Nerubay
Henri Horoszowski
Richard M. Goodman[1]

Sackler School of Medicine, Tel-Aviv University Departments of Orthopedics and [1]Genetics, the Chaim Sheba Medical Center, Tel-Hashomer 52621, Tel-Aviv, Israel

Progressive ossifying fibrodysplasia is a rare autosomal dominant connective tissue disorder with irregular penetrance characterized by progressive ossification of fasciae, aponeuroses, tendons, ligaments, and interstitial tissue of skeletal muscles. In addition to having multiple palpable calcified masses over the body, such patients typically have small thumbs and halluces (Rosenstirn 1918, Lutwak 1964, Cramer et al. 1981, Connor & Evans 1982). Guy Patin (1692) first reported a woman who gradually became "dure comme du bois" (hard as wood). Most of the reports deal with clinicopathologic features, genetics, and therapy. Fractures do not figure among the more than 550 cases reviewed by Connor & Evans (1982).

We present the management of a child with progressive ossifying fibrodysplasia of the humerus and femur on two separate occasions.

Case report

A 6-year-old boy was seen in the emergency room after a fall. The radiographs showed an oblique fracture of the midshaft of the left humerus. He was known to suffer from progressive ossifying fibrodysplasia (Figure 1) and had been treated during the past year and a half with steroids and high doses of vitamin A. The arm was immobilized in a hanging cast for 4 weeks. After removing the cast, physiotherapy was started. The fracture healed, but the mobility of the joints slowly decreased; presently he has only 30° abduction of the upper arm, whereas the flexion of the elbow is only from 90° to 105° with full pronosupination (Figure 2).

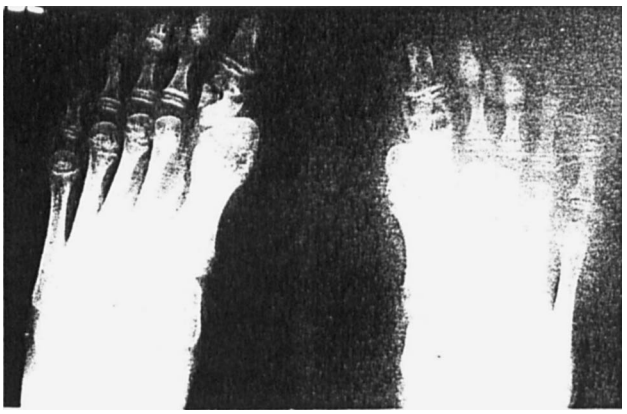
One year later, he fell again and was admitted with a midshaft fracture of his right femur. He was initially treated by skeletal traction. One week later, an open reduction with plate fixation was performed with mobilization after 10 days. The mobility decreased in spite of physiotherapy. Now, 1 year later, the range of motion of his hip joint is 0° in extension, 30° in flexion, and 30° each in abduction, adduction, and external and internal rotation, whereas the knee lacks 15° of full extension and has further flexion of only 25° (Figure 3).

Discussion

Because this is a disorder primarily affecting connective tissue with secondary changes in the muscle, Bauer & Bode (1940) proposed the term fibrodysplasia ossificans progressiva. There have been many theories regarding the basic defect in this disease, and also controversy as to whether it is a systemic or multifocal condition (Cramer et al. 1981). Virchow, in 1894, postulated that the lesions are caused by a disturbance in embryonic development with ectopic periosteal rest cells. Others have tried to explain the lesions as a reaction to dystrophic tissue calcification.

It has recently been shown that the interaction of the fibroblast with collagen and other connective tissue molecules is mediated by cell surface protein that modulate fibroblast behavior (Zetter et al. 1978). It may be relevant that bone, fascia, ligament, and tendon all share Type I collagen as their main connective tissue protein, but so far no biochemical defect has been noted here in this disease.

A



B

Figure 1. A 6-year-old boy with progressive ossifying fibrodysplasia.
A Rigid posture and masses over the back.
B Characteristic big toe malformation.

Figure 2.

Figure 3.

Figure 2. Healed humeral fracture and ectopic bone formation in the axilla and thoracic wall.

Figure 3. Healed femoral fracture with exuberant ossification.

Multiple forms of treatment using steroids, agents binding minerals or blocking calcifications (EDTA and EHDP), and high doses of vitamin A have all failed (Holmsen et al. 1979, Illingworth 1971, Russell et al. 1972). Surgical excision of the bony bridges to relieve the ankylosis and mobilization of joints has been disappointing: trauma rather seems to aggravate the condition (Rogers & Geho 1979).

A few authors (Connor & Evans 1982, Rogers et al. 1977, Rogers & Geho 1979) mention that these patients may be prone to long-bone fractures, but no guidelines for the treatment of such fractures is discussed. In our patient, the humeral

fracture was treated conservatively with great impairment of joint motion. After the patient sustained a fracture of the femur, we decided that early mobilization and physiotherapy could be hastened by internal fixation. Whether our relatively poor result could have been improved by immediate surgical intervention or would have been better without the operative intervention remains unanswered. Hopefully, others will report on fracture treatment in progressive ossifying fibrodysplasia.

References

Bauer, K. H. & Bode, W. (1940) Erbpathologie der Stutzgewebe beim Menschen. (1972) In: *Erbpathologie Korperliche Zustande und Funktionen.* (Ed. Bauer, K. H. et al.), Vol. 1., p. 105. Springer Verlag, Berlin, quoted by McKusick (1972).

Connor, J. M. & Evans, D. A. (1982) Extra-articular ankylosis in fibrodysplasia ossificans progressiva. *Br. J. Oral Surg.* **20**, 117–121.

Connor, J. M. & Evans, D. A. (1982) Fibrodysplasia ossificans progressiva. The clinical features and natural history of 34 patients. *J. Bone Joint Surg.* **64-B**, 76–83.

Cramer, S. F., Ruehl, A. & Mandel, M. A. (1981) Fibrodysplasia ossificans progressiva: a distinctive bone-forming lesion of the soft tissue. *Cancer* **48**, 1016–1021.

Holmsen, H., Ljungdall, S. & Hierton, T. (1979) Myositis ossificans progressiva. Clinical and metabolical observations in a case treated with a diphosphonate (EHDP) and surgical removal of ectopic bone. *Acta Orthop. Scand.* **50**, 33–38.

Illingworth, R. S. (1971) Myositis ossificans progressiva (Munchmeyer's disease). Brief review with report of two cases treated with corticosteroids and observed for 16 years. *Arch. Dis. Child.* **46** , 264–268.

Lutwak, L. (1964) Myositis ossificans progressiva. Mineral, metabolic and radioactive calcium studies of the effects of hormones. *Am. J. Med.* **37**, 269–293.

McKusick, V. A. (1972) *Hereditable disorders of connective tissue.* 4th ed., pp. 687–732. Mosby, St Louis.

Patin, G. (1692) *Lettres Choisies de Feu de M. Guy Patin.* Vol. 5., p. 28. Cologne (cited by Lutwak 1964).

Rogers, J. G., Dorst, J. P. & Geho, W. B. (1977) Use and complications of high-dose disodium etidronate therapy in fibrodysplasia ossificans progressiva. *J. Pediatr.* **91**, 1011–1014.

Rogers, J. G. & Geho, W. B. (1979) Fibrodysplasia ossificans progressiva. A survey of forty-two cases. *J. Bone Joint Surg.* **61-A**, 909–914.

Rosenstirn, J. (1918) A contribution to the study of myositis ossificans progressiva. *Ann. Surg.* **68**, 485–520.

Russell, R. G., Smith, R. Bishop, M. C. & Price, D. A. (1972) Treatment of myositis ossificans progressiva with a diphosphonate. *Lancet* **1**, 10–12.

Smith, R., Russell, R. G. & Woods, C. G. (1976) Muyositis ossificans progressiva. Clinical features of eight patients and their response to treatment. *J. Bone Joint Surg.* **58-B**, 48–57.

Virchow, R. (1894) Über myositis ossificans progressiva. *Berlin Klin. Wochenschr.* 727.

Zetter, B. R., Martin, G. R., Birdwell, C. R. & Gospodarowicz, D. (1978) Role of the high-molecular-weight glycoprotein in cellular morphology, adhesion, and differentiation. *Ann. N.Y. Acad. Sci.* **312**, 299–316.