

A Memorable Case Report

Fibrodysplasia ossificans progressiva

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Abstract

Fibrodysplasia ossificans progressiva (FOP) is a rare genetic disease. It is characterized by widespread soft tissue ossification and congenital anomalies of the extremities. We report a 6 year old boy who was diagnosed as FOP on the basis of heterotopic calcification and hallux valgus.

Introduction

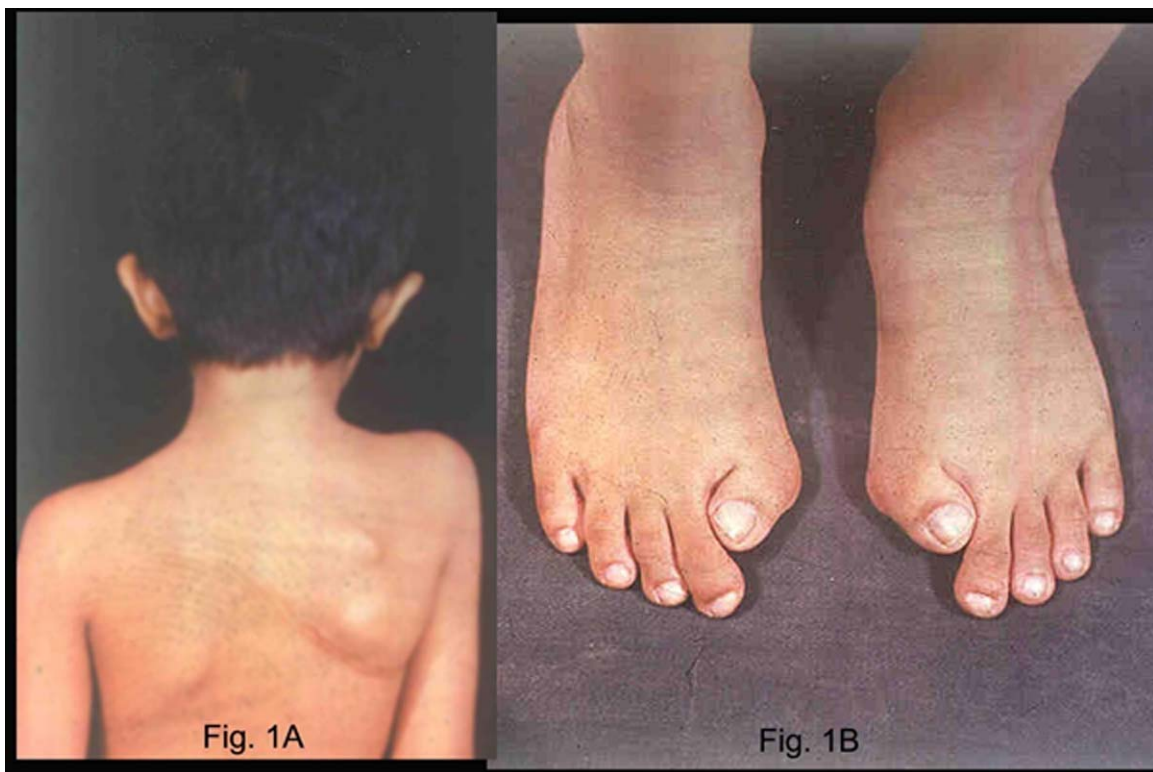
Fibrodysplasia ossificans progressiva (FOP) is a rare autosomal dominant disorder of connective tissue characterized by congenital malformation of the great toes and by progressive post-natal heterotopic ossification of soft tissue. We report a 6 year old boy who was diagnosed as FOP on basis of heterotopic calcification and deformities of great toes.

Case Report

A 6-year-old male child presented with a bony swelling over scapula and back of the neck with restrictions of movement of neck towards right side for the past 6 months. At 2 years of age he had empyema on right side for which thoracotomy and drainage was done. He was the first child born to non-consanguineous parents by cesarean section. His development was normal. On examination, his weight was 20 kgs, height was 119 cms and head circumference was 53 cms. He had torticollis towards right side. He had bony swelling over the right scapula over the thoracotomy scar and also on back side of neck (Fig.1a). He had restriction of movements of right shoulder. His thumb was stiff and hallux valgus deformity was seen in both the great toes (Fig 1b). His systemic examination did not reveal any other abnormality. Laboratory investigations revealed serum calcium of 9.5 mg%, phosphorus 5.3 mg% and alkaline phosphate 513 IU/L.

Fig 1a. 6 year old male child having a bony swelling over the scapula and back side of neck. Note the torticollis towards right side.

Fig 1b. Hallux valgus deformity of great toes



His skeletal radiographs revealed ectopic calcification over the right scapula. The calcification extended into the posterior facial planes of the neck (Fig.2).

Fig 2. Skeletal radiograph showing ectopic calcification over the right scapula. The calcification extending into the posterior facial planes of neck



He was diagnosed to have FOP on the basis of typical radiological features and hallux valgus. He was advised to take steroids during acute flare-ups followed by non-steroidal anti-inflammatory drugs. Intravenous pamidronate was recommended every 3 months to prevent further progression.

Discussion:

Gay Patin in 1962 described the first case of FOP as a woman who “turned into a log of wood”. [1] FOP is a disease of the mesodermal tissue, characterized by initial inflammation followed by subsequent proliferation of the fibrous tissue and formation of ectopic bone tissue. Its incidence is one case per two million. It is a disorder which generally occurs in the first three decades of life, and a majority of patients have onset of symptoms by the age of four years, but there can be a delay of many months before the diagnosis is made. [2] The disease predominantly affects boys. It is transmitted as a dominant trait.

The ectopic bone tissue formed is located in soft parts, mainly in the connective tissue of the striated musculature. The lesions begin in the paravertebral musculature and progress into the scapula, proximal portion of the arms, pelvis, jaw and skull. [3] The ossification process can

intensify in the presence of trauma, as occurred in our case. Exacerbation of FOP may occur spontaneously or be precipitated by trauma, such as intramuscular injections including vaccines [4], local anesthesia, muscle biopsy and careless venipuncture. Biopsy of calcified nodules is to be avoided if the diagnosis of FOP is clear on clinical and radiological grounds. Biopsy may result in recurrent ossification of the site, sometimes worse than the original lesion. Symmetrical congenital malformations of the hands and feet occur in up to 90% of the cases and are considered essential for its diagnosis. [5] The most frequent congenital abnormalities are hypoplasia of the thumbs, big toes and hallux valgus. These are due to shortening of the first phalanges, as observed in our child.

There are no specific laboratory alterations in FOP. Skeletal radiography is the preferential examination for its diagnosis and follow-up, by depicting the appearance of new ossification. Bone scintigraphy with ^{99m}Tc-MDP may demonstrate early the heterotopic ossification and aid in the assessment of the extent and progression of the disease. [6] The phenotype and natural history of FOP are by now so well defined that differential diagnosis is limited. Other disorders of ectopic ossification may be considered, such as Albright hereditary osteodystrophy, pseudomalignant heterotopic ossification, progressive osseous heteroplasia and even osteosarcoma. [7]

To date, there is no effective therapy to impede progression of the disease. However, several drugs have been suggested for the treatment of FOP such as retinoic acid [8], warfarin, and diphosphonates. [9] Corticosteroids can be useful for the acute process, but they do not prevent ectopic calcification. Some authors have demonstrated that the use of oral or intravenous diphosphonate appears to be a therapeutic alternative to reduce ectopic calcifications. [10]

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