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Fibrodysplasia ossificans progressiva in a Nigerian: a case report

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Summary
A case of fibrodysplasia ossificans progressiva in a 3-year and 10-month-old Igala boy is presented. It was characterized by progressive ossification of connective tissue and muscles of the neck and chest. Characteristic digital anomalies particularly microdactyly of the hallux and pollex, hallux valgus, and clinodactyly were also noted. The 2nd to 5th metacarpals had epiphyses at both the proximal and distal ends instead of only at the proximal ends. Although the patient presented in the first year of life, the diagnosis was not made until in his fourth year of life. Early diagnosis of the disease is important in order to avoid all forms of trauma that may worsen the prognosis.

Keywords: *Microdactyly, hallux valgus, clinodactyly, synostoses, ossifications.*

Résumé
Un cas d'ossification fibrodysplasique progressive chez un enfant de 3 ans 10 mois Est présente, caractérise par une ossification progressive des tissues connective et des muscles du cou et de la poitrine. Les caractéristiques des anomalies digitales particulièrement microdactyle du hallus et pollex, hallus valgus et clinodactyle étaient aussi enregistrés. Les 2ime au 5 ieme métacarpes avaient d'épiphyses à la fin distale et proximale au lieu des fins proximal seulement. Cependant le patient de la première an de vie n, avait eu aucun diagnostie jusqu a la 4iéme année. Le diagnostie précoce de la maladie est important dans l'ordre d'éviter toutes les formes de traumatisme qui pourraient aggraver le prognosti

Introduction

Fibrodysplasia ossificans progressiva is a rare congenital disorder characterized by progressive ossification of connective tissue and muscle as well as digital anomalies particularly short hallux, hallux valgus and short thumb [1].

The incidence of the disease is not known for certain. Most recorded cases have been in North America and Europe but it has also been recorded in India [2]. There is paucity of data on Africans. The first two documented cases in Nigeria were by Nwobi [3] and Obidike *et al* [4]. This case is being reported to highlight the need for early diagnosis of the disease in order to avoid all forms of trauma that may worsen the prognosis.

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Case report

HA is a 3-year and 10-month-old Igala boy who was well until at the age of 9 months when he was noted to have recurrent painful swellings on the head, and later on the shoulders and the upper part of the trunk on both sides. The swellings were of various sizes and were associated with fever. Each swelling lasted for about 2-4 weeks before resolving.

They resolved with progressive stiffness of the neck, the shoulders and the elbow joints. The pregnancy and the neonatal periods were uneventful. The developmental milestones were slightly delayed.

On examination there were cord-like contractures of the trapezius, pectoralis major and biceps brachii muscles giving rise to limitations of movement of the head and neck, fixed abduction deformity of both shoulders, and fixed flexion deformity of the elbows. There were also short halluxes and hallux valgus deformity of the feet. He also had hypospadias.

The laboratory investigations included PCV of 33%, WBC of 5,200/mm^3 (neutrophils 49%, lymphocytes 50%, and eosinophils 1%) and ESR of 14 mm/1st hr. The alkaline phosphatase was 114 iu/L while the serum calcium was 2.5mmol/L. They were within normal limits. The inorganic phosphorus was high (2.2 mmol/L in comparison with a normal range of 1.0-1.8 mmol/L). The quadriceps muscle biopsy showed a normal histology while the biceps brachii biopsy done two months later showed increased fibrous tissue.

In the cervical radiograph, there was fusion of the posterior elements of C4 , C5, C6, C7 and a thorn-like calcified plaque overlying the spine of T1 (Fig. 1). The chest radiograph showed soft tissue swellings in the chest wall, thorn-like ossifications in the left and right anterior axillary walls, the right cubital fossa and the right chest wall (Fig. 2). In both hands, the first metacarpals were short and there were clinodactyly of the little fingers with short middle phalanges. There were synostoses of the bases of the left first and second metacarpals. The 2nd to the 5th metacarpals had epiphyses at both the proximal and distal ends instead of only at the proximal ends (Fig. 3). Each foot showed short hallux, hallux valgus and synostoses of the first metatarsal and proximal phalanx. Bilateral syndactyly of the 2nd to 4th toes was also noted. The proximal phalanges appeared normal while the others were small and ill defined (Fig. 4).

The treatment the patient received included prednisolone and analgesics. He also had incisional

biopsy, tenotomy of biceps brachii, and physiotherapy to increase mobility at the joints. The patient's condition did not improve. Diphosphonate was later added to the regimen but soon afterwards he was lost to follow up.

Fig. 1: Lateral cervical radiograph showing fusion of the posterior elements of C4, C5, C6 and C7 Noted also is a thorn-like calcified plaque overlying the spine of T1

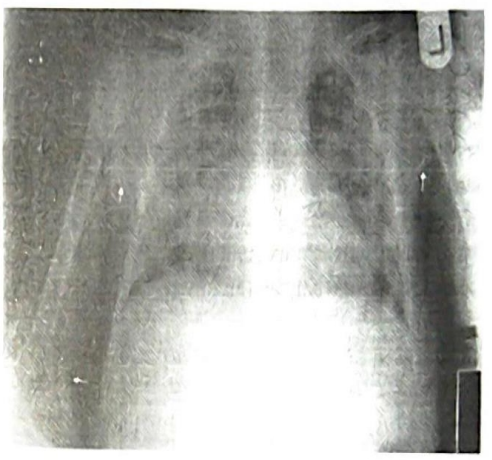

Fig. 2: Anteroposterior chest radiograph showing thorn-like calcified plaques in the left abnd right anterior axillary walls, the right cubital fossa and the right chest wall.

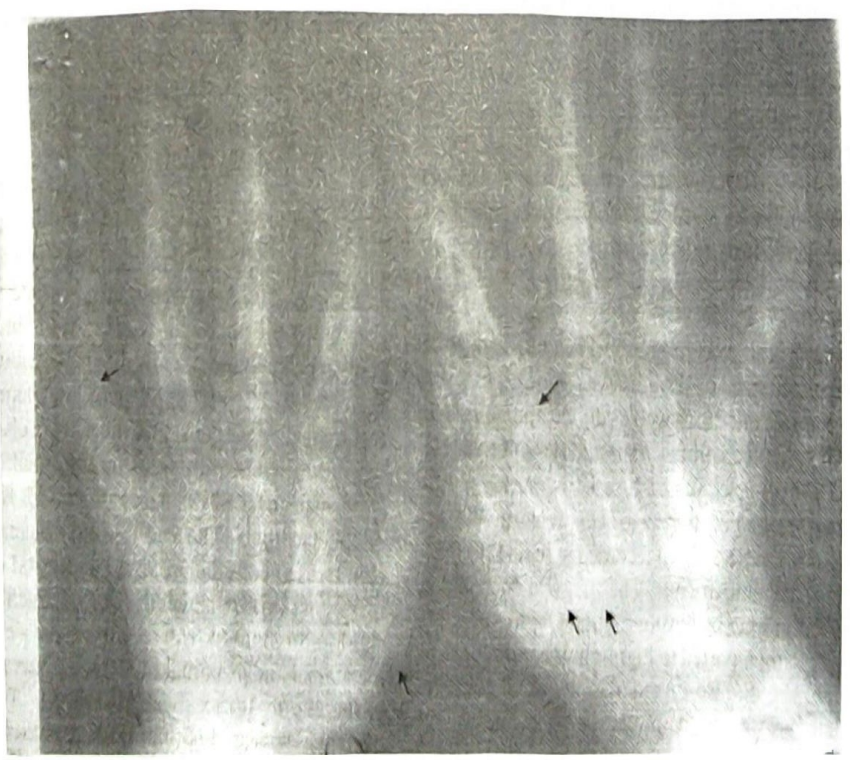

Fig. 3: Anteroposterior radiograph of the hands showing short first metacarpals, clinodactyly and short middle phalanges of the little fingers. Noted also are synstoses of the bases of the left first and second metacarpals The 2nd to the 5th metacarpals have epiphyses at both the proximal and distal ends instead of only at the distal ends

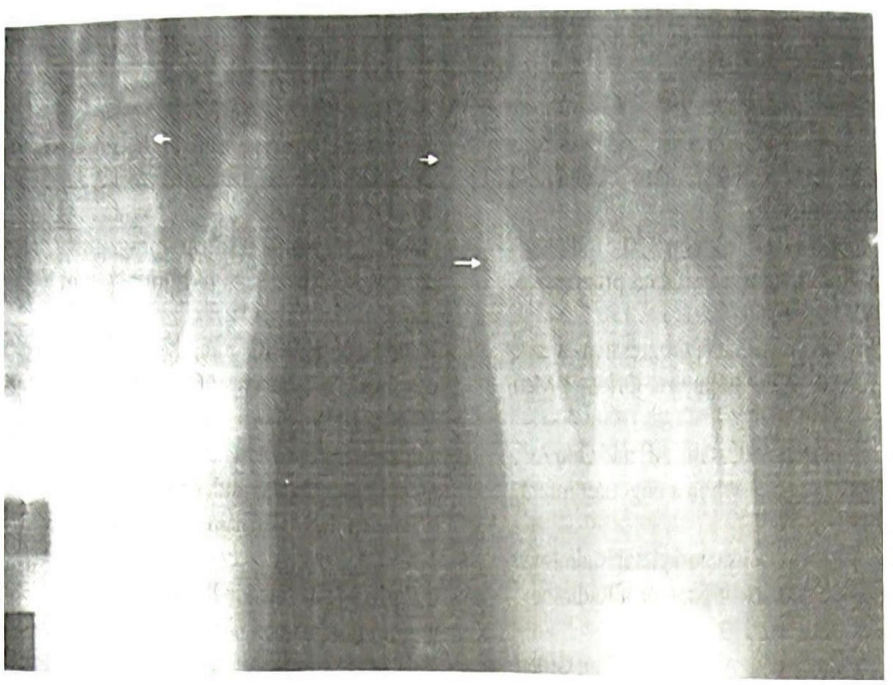

Fig. 4: Radiograph of the feet showing bilateral short halluxes, hallous valga and synostoes of the first metatarsals and proximal phalanges. There is also bilateral syndactyly of the bases of 2nd to 4th toes.

Discussion

The pathophysiology of fibrodysplasia ossificans progressiva is not well known. The ossification that occurs in tendons and fasciae of muscles may eventually result in total immobilization of the patient [5]. The cases reported in Nigerians by Nwobi [3] and Obidike *et al* [4], and our case are all male patients. Rogers and Geho [1] in a review of forty cases showed that the incidence of the disease was approximately the same for both males and females. However earlier studies on the disease showed a higher incidence in males.

The disease may appear in utero, in childhood or in adulthood. However, most cases present before the age of 5 [1]. Although the disease may be diagnosed in early childhood, it may be followed by years of relative quiescence up to late adolescence or may progress throughout childhood. In this case report the disease presented in the first year of life but the diagnosis was not made until the patient was almost 4 years old.

The disease usually presents first with localized soft tissue swellings that may be warm and painful and may be associated with constitutional symptoms like fever [6] as seen in this case report. According to Steibert *et al* [7], the swellings most commonly arise in the head and neck region and progress in a cranio-caudal direction. They may resolve entirely or may be followed by hardening, contraction and ossification over a period of few months. This mode of progression of the disease was also noted in this case report. Usually there is remission and exacerbation of nodular swellings and progressive ossification of the muscle and connective tissue. Torticollis in early childhood is common and is mainly due to ossification in the paraspinal muscles [8]. This case report, in addition to having remission and exacerbation, also had a mild degree of torticollis.

The trunk musculature was also involved in this case report. This involvement is seen in practically all cases of the disease. Affectation of the paravertebral region and the chest limits lung expansion and increases the patient's susceptibility to pneumonia. Conduction deafness and involvement of the muscles of mastication have also been reported.

Radiology plays a very important role in the early diagnosis of fibrodysplasia ossificans progressiva. The use of CT and MRI helps in early diagnosis of the disorder especially when the radiographic features are subtle [9]. Bone scintigraphy is also helpful because it will show increased uptake of tracer elements in centres of abnormal ossification [10]. In this case report the diagnosis was made on plain radiographs because the radiographic features were already gross at the time the patient presented. Fibrodysplasia ossificans progressiva may be confused with traumatic myositis ossificans that give rise to ectopic ossification. The presence of digital anomalies particularly short hallux, hallux valgus, short thumb and clinodactyly as was seen in this case report is pathognomonic of fibrosdysplasia ossificans progressiva.

There is no known effective treatment for this disease. Corticotrophin, prednisolone, calcitonin, indomethacin, ethylenediamine tetraacetic acid and disodium etridronate have been tried with little effect. Life expectancy of patients with fibrodysplasia ossificans progressiva is below normal [11]. Trauma including surgical intervention is implicated in the development of ectopic ossification and progression of the disease [6]. Early diagnosis is important in order to avoid subjecting the patient to surgery since the site of operation may rapidly progress to focal ossification [1].

References

1. Rogers JG and Geho WB. Fibrodysplasia ossificans progressiva: a survey of forty-two cases. J Bone Joint Surg 1979; 61:909-914.
2. Cremin B, Connor JM and Beighton P. The radiological spectrum of fibrodysplasia ossificans progressiva. Clin Radiol 1982; 33:499-508.
3. Nwobi DG. Myositis ossificans progressiva- a case report and a review of the literature. Orient J Med 1990; 2:76-79.
4. Obidike EO, Nwokocha ARC, Gini AE and Onu AC. Myositis ossificans progressiva in a Nigerian infant. Orient J Med 2000; 12:43-45
5. Poznanski AK. Pediatric Musculoskeletal Radiology. In: Pettersson H., ed. A Global Textbook of Radiology. Oslo: The NICER Institute, 1995; 523
6. Thickman D, Bonakdar-pour A, Clancy M, Van Orden J and Steel H. Fibrodysplasia ossificans progressiva. Am J Roentgenol 1982; 139:935-941.
7. Seibert JJ, Morrissy RT, Fuller J and Young LW. Fibrodysplasia ossificans progressiva. Am J Dis Child 1983; 137: 78-79.
8. Smith R. Myositis ossificans progressiva: a review of current problems. Semin Arthritis Rheum 1975; 4:369-380.
9. Reinig JW, Hill SC, Fang M. *et al*. Fibrodysplasia ossificans progressiva- CT appearance. Radiology 1986; 159:153-157.
10. Carter SR, Davies AM, Evans N and Grimer RJ. Value of bone scanning and computed tomography in fibrodysplasia ossificans progressiva. Br J Radiol 1989; 62: 269-272.
11. O Reilly M and Renton P. Metaphysial abnormalities in fibrodysplasia ossificans progressiva. Br J Radiol 1993; 66: 112-116.

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