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Case Report

A case of fibrodysplasia ossificans progressiva with sternocleidomastoid muscle calcification

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ABSTRACT

Fibrodysplasia ossificans progressiva (FOP) is a rare and disabling genetic condition characterized by congenital skeletal malformations and progressive heterotopic ossification in humans with no ethnic, racial, gender, or geographic predilection. Diagnosis of this condition can be made clinically in the presence of radiographic evidence of heterotopic ossification along with symmetrical malformations of the great toes. The course of the disease is unpredictable and often progresses in the early childhood and patients become immobile and confined to a wheelchair by their twenties. Survival beyond the third decade is uncommon. We hereby report a case of FOP in a 7½-year-old girl.

Key words: *Fibrodysplasia ossificans progressiva, Heterotophic calcification, Myositis ossificans progressive*

Fibrodysplasia ossificans progressiva (FOP) is also known as myositis ossificans (MO) progressiva (Stone man disease, Munchmeyer's disease) is an exceptionally rare autosomal dominant disorder of connective tissue characterized by congenital malformations of the great toes and progressive heterotopic ossification in the characteristic extraskeletal sites [1]. The likely incidence is about 1 in 2 million with no ethnic, racial, gender, or geographic predilection. The diagnosis is based on the clinical findings and radiological demonstration of the skeletal malformations [2]. We hereby report a case of FOP in a 7½-year-old girl from Chhattisgarh.

CASE REPORT

A 7½-year-old female child admitted in our hospital with complaint of inability to raise both shoulders. She had painless swelling over lower back on both the sides 6 months back which gradually regressed in size and disappeared. There was no history of trauma, systemic illness, or prior hospitalization. Perinatal history and family history were insignificant. She was well and active. Her growth and developmental milestones were appropriate for the age.

On examination, there was the drooping of the left shoulder. There was a single bony hard subcutaneous swelling overlying the right scapula. The swelling was non-tender, and no signs of inflammation were present. There was a significant restriction of abduction and internal rotation of both the shoulders. Partial restriction of all movements of the spine and restriction of flexion of the neck were also present. Physical examination showed stiffness in the entire spine, affecting from cervical to lumbar region. Affected muscle was stony hard in consistency, immobile, and painless to palpation and showed no inflammatory signs. Clinodactyly of both the little fingers and slightly shortening of both great toes were present. Her hearing assessment reveals a normal study.

Radiographs revealed hypoplasia of proximal phalanges of great toes bilaterally, ectopic ossifications in bilateral axillary region, both sides of the neck, and in the paravertebral muscles. There is calcification of entire right sternocleidomastoid muscle (Figs. 1 and 2). Laboratory tests such as blood cell count, serum calcium, alkaline phosphatase, parathormone, vitamin D3, C-reactive protein, rheumatoid factor, and complete urine examination showed normal values. Considering these results, and analyzing the signs and symptoms presented by the patient, the clinical diagnosis of FOP was made. At present, there is no effective prevention and curative treatment for this debilitating disease. Parents were counseled about the condition, and prognosis was explained.

DISCUSSION

MO is an extraosseous non-neoplastic growth of new bone. The two main recognized MO subtypes are (1) traumatic MO which occurs following trauma (uncommon in children), and (2) FOP. FOP is a rare, fatal, inherited disorder causing fibrosis and ossification of muscles, tendons, and ligaments [1,2]. Pathogenesis of traumatic MO still remains unclear. Most of the researchers believe that repetitive small traumas (not recognized by the patient), infection, inflammation, and ischemia may be the underlying factors that contribute to non-traumatic MO [3].

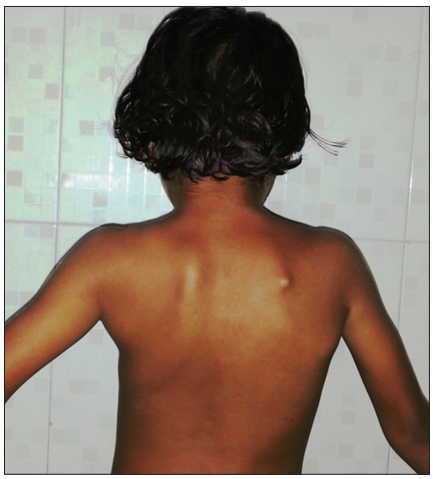

Figure 1: Prominent nodule on back

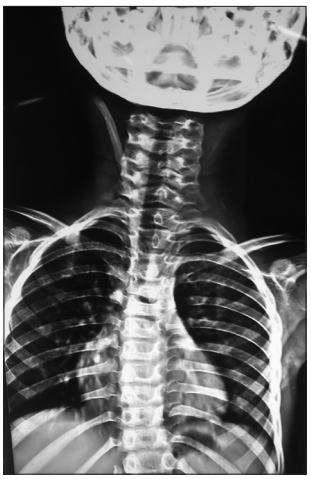

Figure 2: Sternocleidomastoid muscle calcification

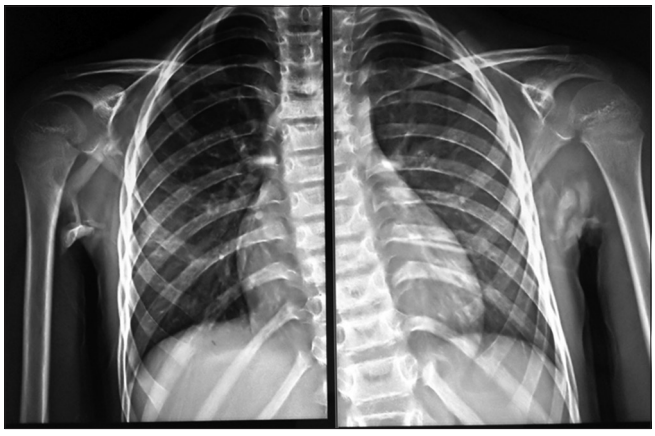

Figure 3: Heterotopic calcification

FOP occurs in approximately 1 in 2 million people [3]. FOP is a genetic disease inherited as autosomal dominant disorder in most of the cases while remaining cases are caused by a spontaneous new mutation in the ACVR1 gene. This mutation causes a deregulation of the bone morphogenetic protein signaling pathway [4,5]. Previous studies have demonstrated that there was no ethnic, racial, gender or geographic predisposition for the development of the disease [6,7].

Recognition of the most characteristic deformity, microdactyly of both halluces along with rapidly changing swellings, helps to make a diagnosis. These begin during the first decade of life and progress until being ossified. Swellings typically affect neck and upper back making them stiff, as in our case. The diagnosis of FOP is therefore based on history, clinical, and radiological findings.

The bilateral great toe anomaly present from birth reported in 79-100% of patients. Deafness has been reported in up to one-fourth of the cases. Malignancy is the most common misdiagnosis with up to 1 in 3 cases being mistaken as a tumor [4]. The soft-tissue trauma can induce rapid ossification of the affected area; therefore, biopsy of calcified nodules is to be avoided if the diagnosis of FOP is clear on the clinical and radiological grounds.

Soft-tissue ossifications are the characteristic radiographic features of FOP (Fig. 3). Bone scintigraphy with 99mTc-MDP may demonstrate the heterotopic ossification early and aid in the assessment of the extent and progression of the disease [8]. Laboratory analysis and biochemical values are usually found to be normal as in our case although we could not carry out bone scans and mutation study. Our case demonstrates the classical presentation and features of FOP. No effective medical therapy is known for FOP; bisphosphonates and corticosteroids are only drugs which have some benefit during acute phase [9]. Gene therapy may have a certain role. The course of the disease is unpredictable, and it often progresses in early childhood, and patients become immobile and confined to a wheelchair by their twenties. Survival beyond the third decade is uncommon as severe restriction of the chest wall results in cardiorespiratory failure.

CONCLUSION

FOP is very uncommon hereditary disorder and must be considered in those cases where extraosseous non-neoplastic new bone formation is seen. Early diagnosis of FOP is important to minimize the trauma, painful flare up, and genetic counseling.

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