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Case Report

Orthopaedics Section

Fibrodysplasia Ossificans Progressive: A Case Report on Rare Musculoskeletal Disorder

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ABSTRACT

Fibrodysplasia Ossificans Progressive (FOP) (also known as Myositis ossificans progressiva/Stone man disease/Munchmeyer's disease) is one of the unfamiliar congenital disorders affecting the musculoskeletal system. It is characterised by extraosseous progressive heterotrophic osteogenesis in muscle, tendon, and ligament and associated deformities in toes. It starts around the age of 3-5 years and aggressively involves the musculoskeletal system; the affected child becomes immobile in the early twenties. The early phase of the disease is often misdiagnosed by medical experts due to its rarity and unfamiliarity. This was a case of FOP in a 5-year-old female child from western Uttar Pradesh. The presenting complaints were swelling in the lower back region, which was gradually increasing in size, and bilateral foot deformity. The diagnosis of FOP was based on elaborated history, clinical examination, and radiological investigation of the skeletal malformations. The child was provided with symptomatic treatment and her parents were counselled regarding the disease course.

Keywords: Congenital disorder, Hallux valgus, Heterotrophic ossification, Skeletal malformation

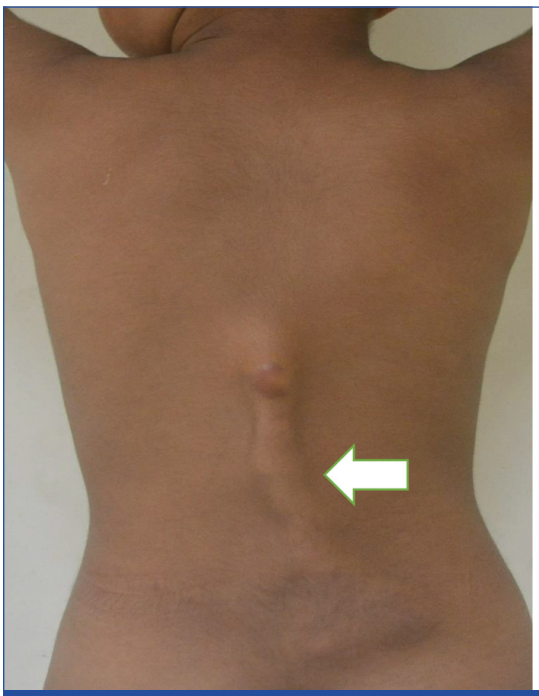
CASE REPORT

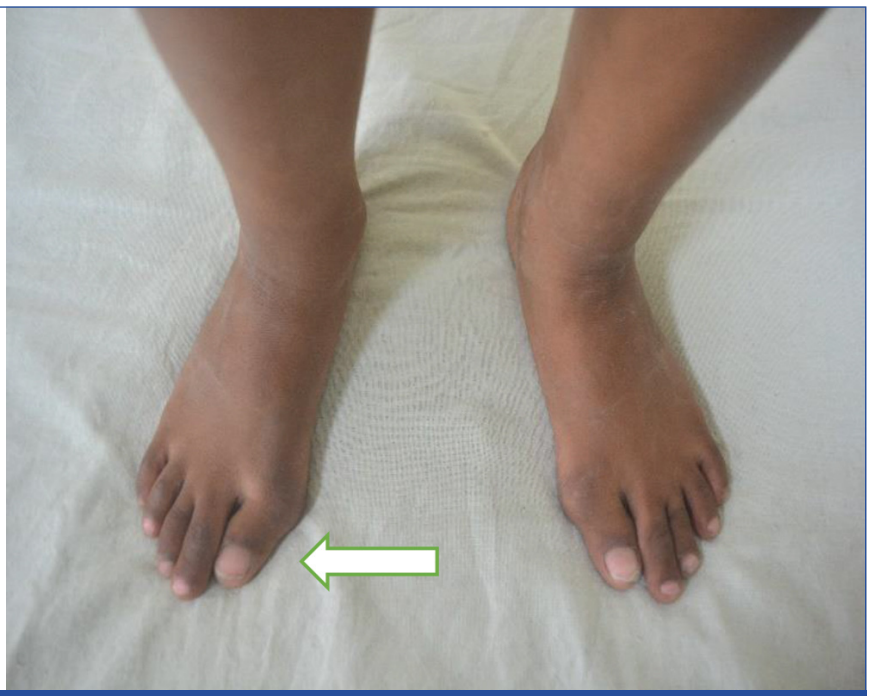
A 5-year-old female child presented in the Outpatient Department at the tertiary care hospital with the complaint of painless swelling since the age of 3 years. Initially, it was small in size and localise around the sacral region, now it had extended to the level of ribs along the spine. There was no history reported of trauma, fever, systemic illness, or prior admission to the hospital.

She was born at term age in a hospital with no significant perinatal event. No other members of the family group from the paternal or maternal side had similar complaints. Her developmental milestones were appropriate for her age and her vaccination status was upto date. She was playful and active upto the age of 3 years.

At the time of examination, the swelling was noted along the spine from the D11 vertebrae to the sacral region [Table/Fig-1]. It was firm, immobile, hard in consistency, not attached to superficial skin and painless to palpation, and showed no active sign of inflammation. There was a gross restriction on the range of motion of the lumbar spine level. Local examination of the lower limb revealed bilateral big toe deformity (hallux valgus) [Table/Fig-2,3].

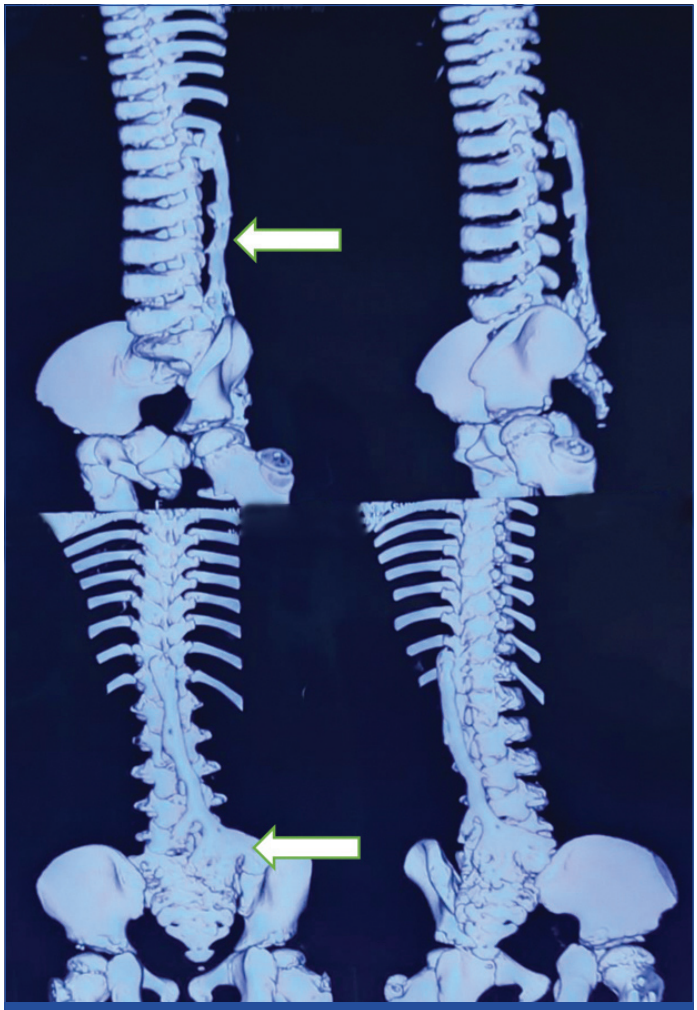
The differential diagnosis were rhabdomyosarcoma, posttraumatic myositis, lymphadenopathy, Tuberculosis (TB), scleroderma, calcinosis interstitialis ossificans, pseudohypoparathyroidism, hypervitaminosis D, and dermatomyositis of childhood. After clinicopathological and radiological investigation diagnosis of Fibrodysplasia Ossificans Progressive (FOP) was made [Table/Fig-3]. A pulmonary function test and cardiac evaluation was done to rule out any syndromic involvement. As there is no definitive treatment available till now so family members were counselled for the same. Supportive medical management was done as she had intermittent





[Table/Fig-1]: Swelling at the back along the spine. **[Table/Fig-2]:** Bilateral hallux valgus deformity. (Images from left to right).

low backache and her haematological investigation were within normal limits and her vitamin D level was low [Table/Fig-4].



[Table/Fig-3]: Radiological image showing bony growth along Lumbosacral spine.

Parameters	Results
Haemoglobin	11.1 mg/dL
Platelet count	3,32,000/cumm
White blood cell count	10,600/cumm
Differential blood count	Neutrophil-62 Lymphocyte-30 Monocyte-5 Eosinophill-3 Basophill-0
Erythrocyte sedimentation rate	22
C-Reactive protein	1.5 mg/dL
Serum calcium	10.3 mg/dL
Serum vitamin D	23 ng/mL
Serum alkaline phosphate	563 IU/L
Serum urea	36 mg/dL
Serum creatinine	0.9 mg/dL
Pulmonary function test	Normal
Echocardiogram finding	Normal

[Table/Fig-4]: Relevant investigations.

The patient was advised to take rest and parents were counselled to take special care of the child during high-risk games/contact games and exercises. Her parents were also informed to avoid any invasive procedure and report immediately to the nearest hospital in case of a traumatic event.

DISCUSSION

In history, the first case of FOP has been discussed by Guy Patin in 1648 with a low incidence rate [1]. The inheritance of FOP is an autosomal dominant type with most patients having a new mutation of a bone morphogenic protein type 1 receptor (ACVR1) which results in activation of osteogenesis in ectopic sites [2]. It is a rare musculoskeletal disorder that gradually increases with age. The clinical hallmark of FOP is malformations of the great toes since birth and extraskeletal ossification that gradually involves the adjacent skeleton resulting in restriction of range of motion and eventually leading to disability and morbidity [3]. In affected individuals, Heterotopic Ossification (HO) starts around the mean age of 3 to 5 years and is visible in almost all patients less than 15 years of age. Patients become bedridden around the fourth decade of life and death happens due to cardiorespiratory involvement.

FOP primarily involves the axial spinal musculature, but extraosseous bone formation also involves the ligaments, fascia, aponeuroses, tendons, and joint capsules [1]. The progression of heterotopic ossification in FOP generally follows a pattern in which the body is affected in an axial-to-appendicular, cranial-to-caudal, and proximal-to-distal sequence. In the present case, it started at the sacrum and then migrated to the cephalic direction. Malformation of the great toes usually presents in most of the cases with the most common valgus position [4]. Bilateral hallux valgus deformity was also present in this case. Malformed great toes are not limited to FOP but it should always be one of the differential diagnosis. Hand deformities may be present as short first metacarpal and brachymesophalangy of the fifth finger with clinodactyly but not found in this child. Deafness, baldness and mental retardation are rare entities and not found in this case [5].

Diagnosis of FOP is the main concern due is its less familiarity with the clinicians, and more than 80% of worldwide cases of FOP are usually misdiagnosed as carcinoma [6]. Laboratory analysis and biochemical values are usually found to be normal as in the present case. Bone scans and mutation studies could not be performed due to the financial condition of the family. Usually, a diagnosis of FOP is made on the basis of clinical and radiological findings. The diagnostic interventional procedure should be avoided because any type of musculoskeletal trauma can induce rapid ossification of the involved area. Till now, no effective conservative medical treatment is available. A short course of corticosteroids (prednisone 2 mg/kg/day) started within the first 24 hours of a flare-up, it helps in reducing the inflammation and tissue oedema seen in the early stages of the disease [7]. Treatment of choice is usually conservative. During the time of flare-ups, initial bed rest is followed by gradual mobilisation. Surgery is limited to reserved cases and is usually performed when ossification has stopped to prevent the occurrence of relapse [8]. Gene therapy is a future treatment option [9].

The patient's parents were counselled regarding the prognosis and course of the disease. The family has also been informed about various Non Governmental Organisation (NGOs) and the availability of support groups like the International Fibrodysplasia Ossificans Progressiva Association (IFOPA) [10].

CONCLUSION(S)

Fibrodysplasia Ossificans Progressive (FOP is a rare musculoskeletal disorder. The important diagnosis feature is bilateral toe deformity. Family physicians, paediatricians, and musculoskeletal experts should be aware of the early features of FOP so that it can be diagnosed before ossification. Early diagnosis will help in minimising the painful flare-up, safety from traumatic events, and unnecessary musculoskeletal interventions.

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