Case Report

Fibrodysplasia Ossificans Progressiva: A rare and severely disabling disease-Case Report and Review of Literature

1. Abdul Sattar 2. Azhar Mehmood Javed 3. Samreen Mushtaq 4. Maria Zahoor 5.Sadia Anjum

1. Asstt. Prof. of Radiology, NMC, Multan 2. Asstt. Prof. of Radiology, ICH, Multan 3. Radiologist, ICH, Multan 4. MO of Radiology, ICH, Multan 5. PG Registrar of Radiology, NMC, Multan.

ABSTRACT

Fibrodysplasia ossaficans progressiva is an extremely rare and severely disabling, autosomal dominant condition that effects 1 in 2 million people. We report a 3 years old female child referred to radiology department from pediatric orthopedic clinic with complaints of multiple painful swellings over the back and inability to sleep due to discomfort. Radiological investigations that include - X-Rays and C.T Scan were done that revealed heterotopic ossifications in muscles, facia and in ligaments.X-Rays revealed bilateral hallus valgus deformity with microdactyly that is characteristic of fibrodysplasia ossificans progressive.

Key words: Fibrodysplasia Ossificans Progressiva (FOP), Heterotopic Ossification, Hallus valgus deformity, 3D CT scan and Multiplanar reconstruction (MPR)

INTRODUCTION

Fibrodysplasia Ossificans Progressiva (FOP), is an extremely rare and severely disabling, autosomal dominant disease characterized by recurrent painful episodes of soft tissue swellings that lead to heterotopic ossification that is true bone formation in muscles, ligaments, fascia, tendons and joint capsules. A variety of congenital skeletal malformations of hand and feet, especially a hallus valgus deformity with microdactyly have been described as characteristic feature. [1, 3]

Incidence is 1 case per 1.64 million in United Kingdom. [4] Fewer than 600 cases have been reported in literature worldwide. [7]The condition is more common in whites, but has been reported in blacks. Females are affected more frequently than males. [2]

Pathogenesis involves over expression of bone morphogenetic protein 4 (BMP-4), which maps to 14q22-q23 [4]

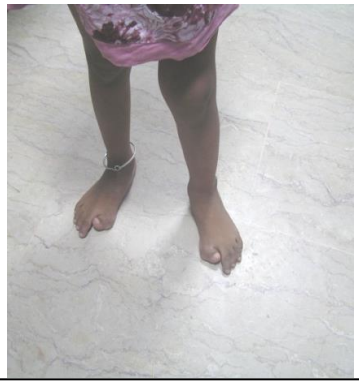
Disease process starts in early infancy, though skeletal deformities are present at birth. There is characteristic anatomical progression of heterotopic bone formation. Progressive involvement of joints leads to ankylosis. Involvement of spine causes pain and stiffness. Severe restrictive pulmonary disease may result due to involvement of muscles of chest wall. [5, 6]

CASE REPORT

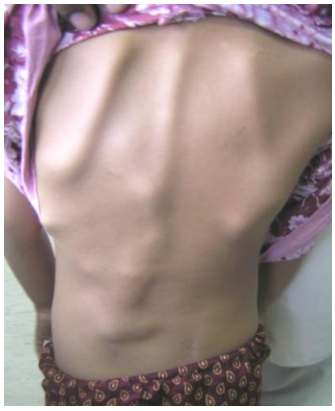
A 3 year old female child was referred to radiology department from pediatric orthopedic clinic with complaints of multiple painful swellings on the back and was unable to sleep properly due to pain.

Physical examination revealed multiple painful and hard tender swellings in paraspinal regions and along chest wall with bilateral hallus valgus deformity.

Biochemical results of mineral metabolism were with in normal limits.



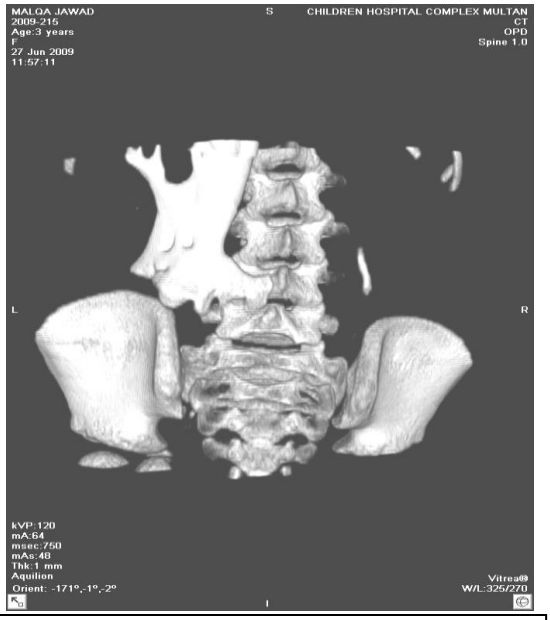
Bilateral hallus valgus deformity



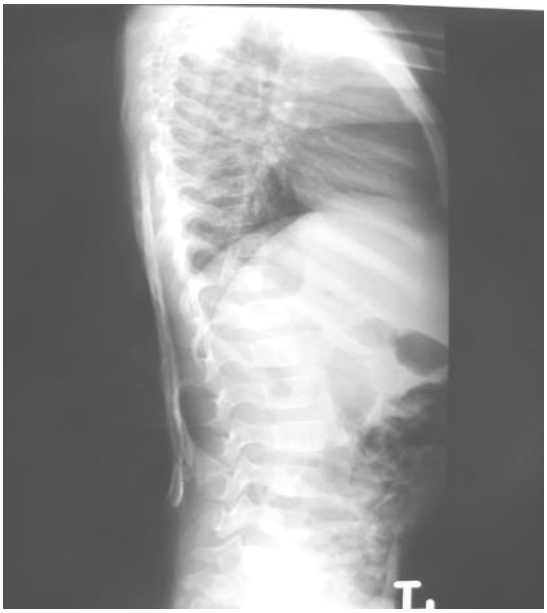
Multiple swellings on back

Digital X-rays showed heterotopic ossification along para spinal areas and chest wall with bilateral hallus valgus deformity.

CT scan spine with multiplanner and 3D reconstruction confirmed the ossification in subcutaneous location along para spinal regions and chest wall.



3 D CT scan showing heterotopic bone formation on back.



X-Ray spine showing heterotopic ossifications in paraspinal area

Patient was send back to the orthopedic surgeon with the diagnosis of FOP

DISCUSSION

FOP, though extremely rare, is by no means new, with reports in the scientific literature as early as 1692.There is no effective treatment for the heterotopic ossifications of FOP[2, 3]. Thus successful management relies on early recognition of this disorder and prevention of inciting inflammation or injury. Especially important is the avoidance of precipitating factors such as biopsies, intramuscular injection, prolonged pressure on body, aggressive physical therapy, falls etc.Soft tissue ossification occurs in response to inflammation or trauma and the pattern of heterotopic ossification that results, is unique to each patient[5]. Frequent sites of heterotopic ossification are paraspinal regions especially in thoracic and the shoulder girdle. Most patients are misdiagnosed despite the presence of pathognomonic changes of the first toe, even evident clinically at birth. [1],[3]

The combination of hallus valgus deformity of first toe and posterior thoracic soft tissue masses are highly suggestive of FOP.A mis-diagnosis rate of 87% has been reported. Soft tissue swelling and heterotopic bone was wrongly attributed to neoplasm in 32% of patients, and 67% of patients had unnecessary biopsies [4]

With awareness of characteristic radiological findings, the radiologist might be the first to recognize the constellation of findings, suggest the correct diagnosis and prevent iatrogenic injury in this rare disorder. No biopsy was done in our patient

CONCLUSION

Although FOP is a rare disease, however awareness of characteristic physical & radiological findings should able the radiologist and pediatrician to make early diagnosis that can avoid unnecessary iatrogenic injuries and flaring up of the disease process

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Address for Corresponding Author:
Abdul Sattar,
Radiology Department,
Nishtar Medical College & Hospital,
Multan, Pakistan
+92-333-4354512
E-mail. drasanjum@gmail.com