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Case Report

A Rare Case of Fibrodysplasia Ossificans Progressiva in a Resource Limited Setup: A Case Report

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Abstract

Fibrodysplasia Ossificans Progressiva (FOP) is a rare genetic disorder characterized by congenital malformation of the great toe and heterotopic ossification in the connective tissue of muscles, tendons, ligaments, fascia, and aponeuroses. It has an incidence of 1 in 2 million, with less than 700 reported cases worldwide to date. The aim of this case report is to highlight clinical features of FOP so that early diagnosis will be made and the impact of its delayed diagnosis will be minimized. We report a case of a 42-year-old man who presented with multiple soft tissue swellings of 30 years duration with later inability to move. Physical examination findings are firm to hard tender masses over bilateral forearms, left arm, anterior and posterior chest, bilateral hip, and bilateral distal thigh with bilateral subtle hallux valgus. He had been visiting nearby health centers for the last 30 years, but a diagnosis was not made until the current visit. With clinical finding of hallux valgus and radiologic finding of extraskeletal ossifications, diagnosis of FOP was made. He is on analgesics and a short course of corticosteroids during flare-ups. Even though it is rare, primary care physicians should consider FOP as a possibility in patients presenting with soft tissue ossifications to minimize suffering and disability due to delayed diagnosis. Even though the confirmatory diagnostic modality for FOP is genetic test, for low-income countries where the genetic test is not easily accessible, clinical consideration should be made to prevent unnecessary disease progression.

Keywords

Fibrodysplasia Ossificans, Heterotopic Ossification, Stoneman Disease, Case Report

1. Introduction

Fibrodysplasia ossificans progressiva (FOP) is an extremely rare genetic disorder characterized by congenital skeletal malformation and heterotopic ossification [1, 3]. Classically, it is characterized by congenital malformation of the great toe and bone formation at extraskeletal sites [1-3]. Even though the majority of cases of FOP are due to sponta-

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neous mutation, there are also cases with autosomal dominant inheritance [2-4]. The most frequently mutated gene in FOP is Bone morphogenic protein (BMP) type 1 receptor (ACVR1), which has an osteogenic effect [1]. FOP has an overall incidence of 1 in 2 million in the population [1-6]. Not more than 700 cases are reported to date [6].

Patients with FOP become symptomatic in the first decade of life. Hallux valgus malformation of the great toe is a universal finding in FOP, and it usually appears at birth [5]. The typical clinical features of FOP are malformation of the great toe and gradually increasing extraskeletal ossification [3]. Analgesia with non-steroidal anti-inflammatory drugs and a short course of steroids during a flare-up is currently considered a supportive medical treatment [1, 3, 9, 11].

We present a case of a 42-year-old male patient who presented to Aksum University Referral Hospital, diagnosed with FOP after 30 years of complaints of multifocal painful body swellings. To the best of our knowledge, this is the third reported case in Ethiopia, a country with a population of more than 120 million.

2. Case Presentation

We present a case of a 42-year-old male Ethiopian who presented to our hospital with a complaint of multiple body swellings of 30 years duration. The swellings are located at the chest, back, bilateral upper extremities, right hip, and bilateral thigh. The swellings started at the age of 12 year, which initially appeared at the back and chest; later, the upper extremity, hip, and thigh swellings appeared sequentially. Associated with the swellings, he also complained of pain at the swelling sites, which was insidious in onset. The pain gradually worsened to the extent of causing him difficulty walking and limiting movement at hip and elbow joints. For the body swelling and pain, he visited traditional healers and the traditional healer, removed the swellings at the chest and back 20 years back. He also had frequent visits at nearby health centers and primary hospitals, at which he was treated with unspecified intravenous and oral medications, but still the diagnosis was not settled. Two years back from the current visit, he had a falling down accident and sustained a left humerus fracture, for which he was treated at a traditional bone settler.

He is the second child in his family, born from non-consanguineous parents after term pregnancy. No similar illness in his siblings and whole family. Now he is married and a father of a 2-year-old boy. His son was also examined for the presence of similar lesions, and no hallux valgus or soft tissue swellings were detected. No significant past medical, surgical, or psychosocial history.

On examination, multiple tender firms to hard masses were identified in bilateral forearms, left arm, bilateral hip, and bilateral distal thigh with multiple scar makings over his chest and paraspinal areas. Bilateral subtle hallux valgus malformation is also noted. He has a limited range of motion at bilateral elbow and right hip joints (Figures 1-5).

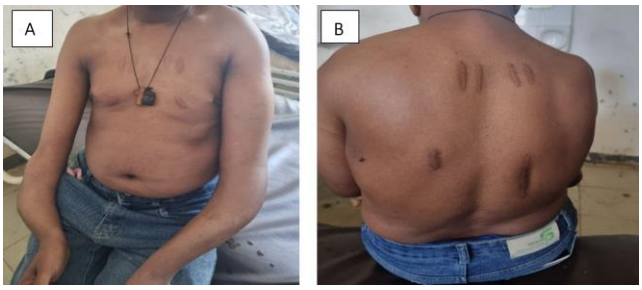

Figure 1. *Clinical photography. A. showing multiple hard masses over bilateral upper extremities and anterior chest with scar markings from traditional treatment. B. Multiple paraspinal hard swellings and scar markings from traditional treatment to remove the swellings.*

Routine laboratory studies were within normal range. Conventional radiological examination revealed widespread heterotopic ossification of soft tissues of the hip, thigh, left arm, and bilateral forearm with evidence of hallux valgus malformation of bilateral great toe (Figures 2-5). Computed tomography (CT) scans and genetic tests were not done because of the availability in our setup and the patient's financial problem.

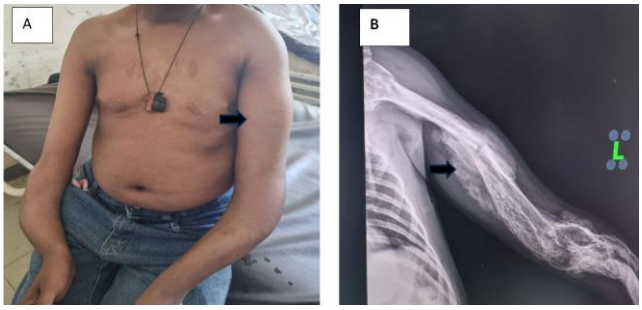

Figure 2. *(A) Clinical photography showing swelling over his right distal arm and forearm, left arm (arrow) and forearm. (B) X-ray of the left arm showing left humerus healed fracture and ossification of soft tissue surrounding the humerus (arrow).*

Figure 3. *Clinical photographs and radiologic findings showing bilateral subtle hallux valgus.*

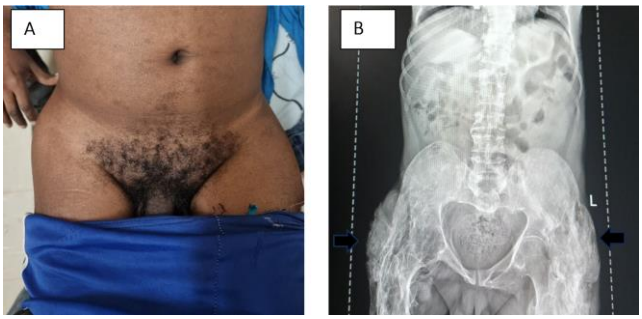

Figure 4. *Clinical photography showing bilateral hip swellings (A) and x ray finding (B) ossification of the soft tissue of hip (arrows).*

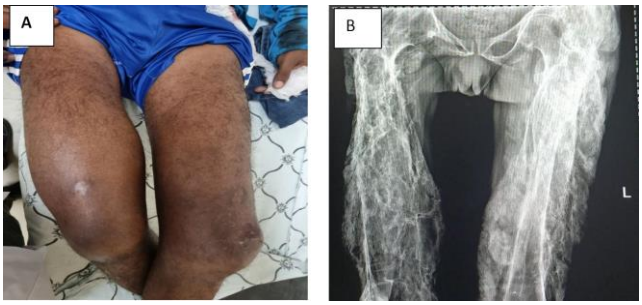

Figure 5. *Clinical photography showing thigh swellings (A) and radiology findings (B) of diffuse ossification of soft tissue of the thigh.*

Based on the history, physical examination, and radiologic findings, the clinical diagnosis of Fibrodysplasia Ossificans Progressiva was made. He was counselled about the etiology, natural course, and prognosis of the disease. He is also strictly advised to avoid any invasive procedure and to be protected from physical trauma as much as possible. He is currently on follow-up with intermittent courses of non-steroidal anti-inflammatory drugs and steroids during flare-ups. He claimed that there are no changes in the size of the masses, and he is still in difficulty of motion at his hip and elbow joints. But he stated that the non-steroidal ant-inflammatory drugs are effective when the pain begins. He did not report any complication against the use of steroids or non-steroidal anti-inflammatory drugs.

3. Discussion

FOP, which is also called stoneman syndrome or munchmeyer's disease, is a rare genetic disorder characterized by soft tissue calcification [8]. Most cases of FOP are the result of sporadic mutation, but autosomal dominant mutation is also reported [2-4, 8]. The most commonly mutated gene is the BMP type 1 receptor, which is called Activin A receptor type 1 (ACVR1), whose net effect is exaggerated ossification at ectopic sites [1, 3, 6-8, 10, 11].

FOP classically presents with bilateral congenital malformation of the great toe, which is the earliest manifestation, and hallux valgus is the commonest and most typical phenotype [11]. The other manifestations that develop during the first decade of life are painful inflammatory soft tissue swelling in the connective tissue of aponeurosis, skeletal muscle, tendons, ligaments, and fascia, which latter transforms to a bony mass [3, 11]. Flare-ups in FOP occur secondary to minor traumas like intramuscular immunizations and blunt muscle trauma from bumps and falls. Influenza-like illnesses can also trigger flare-ups of FOP [3, 11]. Surgical excision of the bony masses for therapeutic or diagnostic biopsy purposes can lead to the explosion of bone growth [11].

The principal radiographic findings of FOP are joint malformations and extraskeletal ossifications [3, 11]. Malformation of the great toe, thumbs, cervical spine, and proximal femur with proximal medial tibial chondromas is helpful to support the diagnosis of FOP [3]. Routine laboratory test results are usually within the normal range, but during flare-ups elevated values of alkaline phosphatase [3, 11] and erythrocyte sedimentation rate [6] can be seen.

Delayed diagnosis is common in patients with FOP because of the fact that it's a rare, unfamiliar nature with primary care physicians. FOP is mostly mistakenly diagnosed as cancers, scleroderma, and fibromatosis [11]. In our case, fibromatosis, scleroderma, and soft tissue malignancy were considered as differential diagnoses along with FOP. But the prolonged duration of the complaint, the presence of hallux valgus malformation of the great toe, and the radiologic finding of extraskeletal ossification all together excluded the other differentials. Our patient had been suffering from the illness for the last 30 years, and because of the delayed diagnosis, he was exposed to treatments by traditional healers, which aggravated the swellings.

There is no known definitive therapy for FOP [6]. Analgesia with non-steroidal anti-inflammatory drugs and a short course of steroids during a flare up is currently considered a supportive medical treatment [1, 3, 9, 11].

Generally, as in our case, delayed and inappropriate diagnosis results in pain, suffering, and disability for the patient as well as the family and the society at large. This case report is aimed at alarming the primary caring physicians and other healthcare professionals to consider FOP as a possible differential diagnosis in patients presenting with soft tissue swellings and congenital malformation of the great toe so that the suffering and disability will be minimized. FOP can result in significant morbidity and disability, but early diagnosis can help in minimising the morbidity by counselling on the natural course of the disease.

4. Conclusion

For patients presenting with congenital malformation of the great toe and soft tissue swellings with radiologic evidence of extraskeletal calcification, FOP should be considered as a top differential. Early diagnosis, counselling on the nature of the disease, and advising on precautions are significant in minimising suffering and disability in patients. Even though the

confirmatory diagnostic modality is a genetic test, for low-income countries where the genetic test is not easily accessible, clinical consideration should be made to prevent unnecessary disease progression.

Abbreviations

FOP	Fibrodysplasia Ossificans Progressive
BMP	Bone Morphogenic Protein
ACVR1	Activin A Receptor Type 1

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Author Contributions

Birhanu Kassie Reta: Conceptualization, Data curation, Project administration, Visualization, Writing – original draft, Writing – review & editing

Kibrom Hadush Seyum: Writing – review & editing, Data curation, Investigation

Million Abraha Zeray: Supervision, Writing – review & editing

Tsegay Birhane Bisrat: Visualization, Writing – review & editing

Yohannes Gebremedhin Berhe: Data curation, Writing – review & editing

Hindeya Hailu Hagos: Supervision, Data curation, Writing – review & editing

Yordanos Birhane Gebrecherkos: Supervision, Visualization

Ethical Approval

Ethical approval for this study is obtained from Aksum university ethical review committee.

Consent for Publication

Written informed consent was obtained from the mother to publish this case report and accompanying images.

Availability of Data and Materials

The patient's data and file are available.

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Conflicts of Interest

The authors declare no conflicts of interest.

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