Fibrodysplasia Ossificans Progressiva: A don’t Touch Syndrome

Mahmut Nedim Aytekin*1, Buğra Alpan2, Ercan Bal1, Maimaiti Mairedanjiang1 and Harzem Özger2

1Department of Orthopedics and Traumatology, Medicine Faculty, Ankara Yildirim Beyazit University, Turkey
2Istanbul Orthopedic Oncology Group, Turkey

Abstract

Fibrodysplasia ossificans progressiva (FOP) is a rare genetic disease that is characterized by the formation of heterotopic bone tissues in soft tissues, such as skeletal muscle, ligament, and tendon. The prevalence is one in two million people. During childhood, it may be asymptomatic but in later life, progressive stiffness of major joints renders movement of the individual impossible. Currently, there is no effective treatment for this debilitating disease. Here, we present a case of 32 years old male with clinical and radiological features of fibrodysplasia ossificans progressiva.

Keywords: Fibrodysplasia ossificans progressiva; Heterotopic ossification

Introduction

Fibrodysplasia Ossificans Progressiva (Munchmeyer’s disease, stoneman’s disease, Myositis Ossificans Progressiva) first described by Guy Patin in 1692 [1,2]. It is a rare progressively disabling disease characterized by formation of heterotopic bone tissues in soft tissues, such as skeletal muscle, ligament, and tendon that gradually encases the original skeleton resulting in restriction of movement and eventual immobility and mortality from cardiorespiratory complications around the fourth decade of life [1,3]. It is extremely rare with an incidence of one in two million individuals occurring without bias to gender, race or ethnicity [4]. It is an autosomal dominant disorder with most patients having a spontaneous new mutation of a bone morphogenetic protein type 1 receptor (ACVR1) which results in activation of osteogenesis in ectopic sites without ligand binding [5,6]. Familial cases have also been reported [7].

Minor trauma such as intramuscular immunizations, mandibular blocks for dental work, muscle fatigue, blunt muscle trauma from bumps, bruises, falls, or influenza-like viral illnesses can trigger painful new flare-ups of FOP leading to progressive heterotopic ossification [8]. Surgical removal of heterotopic bone provokes explosive and painful new bone growth. The correct diagnosis of FOP can be made clinically even before radiographic evidence of heterotopic ossification is seen, if soft tissues lesions are associated with symmetrical malformations of the great toes.

Definitive genetic testing of FOP is now available and can confirm a diagnosis of FOP prior to the appearance of heterotopic ossification. Clinical management of FOP remains symptomatic [8].

Case Presentation

A 32 years old male presented with pain on right knee and difficulty of walking. Clinical records reveal that at the age of about 12, he was diagnosed with congenital disease. On the physical examination: we found that he has a stiff right knee which was at the position of 70 degree flexion without any motion. The range of motion of his left hip has been decreased. His endocrinologic and systemic screening was normal except his musculoskeletal system problems. He had an excisional biopsy operation from his right hip six months ago in a medical centre. The size of excisional material was 12x4x4 cm and pathology result was; ‘sclerotic bone trabeculas + bone marrow tissues + environmental fibrous tissues’. This case has been consulted on Musculoskeletal Oncology Committee of Ankara and the decision of the committee was ‘DON’T TOUCH’.

Discussion

Fibrodysplasia ossificans progressiva (FOP), a rare and disabling genetic condition characterized by progressive heterotopic ossification (HEO) [9]. Surgical interventions are risk for new trauma-induced HO formation and release of joint contracture is generally unsuccessful [10,11]. Even A biopsy is contraindicated...
due to risk of catastrophic explosive new bone formation. Spinal bracing is ineffective and surgical intervention is associated with numerous complications [10-12]. On this report, we could give a comment to this syndrome as ‘Don’t Touch Syndrome’.

Figure 1: Plain radiograph of shoulder - anteroposterior view. Heterotopic ossification was seen in the soft tissues lateral side of the proximal humerus.

Figure 2: Plain radiograph of lumber spine – anteroposterior view. Heterotopic ossification was noted along the bilateral paraspinal area.

Figure 3: Plain radiograph of pelvis with bilateral hips– anteroposterior view. Heterotopic ossification was noted around the left hip joint.

Figure 4: Plain radiograph of both knee– anteroposterior and lateral view. Heterotopic ossification was noted medial side of the right knee as well as bilateral proximal tibia.

Acknowledgement

None.

Conflict of Interest

No conflict of interest.

References