MYOSITIS OSSIFICANS PROGRESSIVA (MUNCHMEYER' DISEASE)
(A case report)

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MYOSİTİS OSSİFİKANS ,PROGRESİVA, MUNCHMEYER HASTALIĞI (Vaka Takdimi)

ÖZET

Fibrodysplasia (myositis) ossificans progressiva nadir görülen herediter bir bağ dokusu hastalğıdır.Hastalık iskelet kaslarının bağ dokusu içerisinde başlangıçta ağrılı olan, progressif ektopik ossifikasyonlar ve simetrik iskelet malformasyonları ile karakterizedir. Hastalıkta ilk belirtiler çocukluk yaş gurubunda başlar. Nadir, otozomal dominant geçişli konjenital bir hastalıktır.Halen etkin bir tedavisi yoktur. Bu yazımızda büyük eklemlerinde hareket kısıtlılığı ile kkiniğimize başvuran boyun, sırt, omuzlar, kalçalar ve dizlerde kas içerisinde ektopik ossifikasyonları ve başparmaklarında kısalığı bulunan ekstensif tutulumlu 23 yaşındaki bir erkek hastayı literatür bilgileri ışığında sunduk.

Anahtar Kelimeler: Myositis ossifikans , Erişkin, Tanı

SUMMARY:

Fibrodysplasia ossificans progressiva is a rare hereditary connective tissue disorder. Patients with fibrodysplasia ossificans progressiva develop progressive ossification of muscle and connective tissue associated with pain and disability. Onset is typically in childhood, and congenital feet anomalies are the early signs of this condition. Pain and stiffness of the spine or an inflammatory mass are common presenting features of fibrodysplasia ossificans progressiva . Studies on twins and families suggest that fibrodysplasia ossificans progressiva is a genetically inherited autosomal dominant trait with complete penetrance but variable expressivity. Unfortunately, effective therapy is unavailable. We present a 23 years old man with fibrodysplasia ossificans progressiva with a review of the literature on the clinical, radiographic, and genetic manifestations of this disorder.

Key Words: Myositis ossificans , Adult , Diagnosis,

Myositis ossificans progressiva, recently known as Fibrodysplasia Ossificans Progressiva, is a rare hereditary mesodermal disorder. It is characterized by congenital skeletal malformation of great toes and severe progressive ossification of the soft tissues, with an onset in late childhood. There is a tendency to heterotopic bone formation in soft tissues such as muscles, tendons, ligaments, fasciae, aponeuroses, joint capsules and occasionally in skin. The diagnosis of Fibrodysplasia ossificans progressiva is based on clinical and radiographic findings. There is no effective treatment, although administration of diphosphonates , simple surgical removal of the ectopic bones, genetic counseling has been advocated. Here we present a 23 years old case and review the literature.

CASE:

A 23 year old male patient admitted with complaints of severe movement limitation of the big joints. In his medical history, he developed a painful lump in his neck at the age of 4 and in the interscapular region at the age of 8, which rapidly changed to bony lesions, like bone. Same type of lesions occurred in his pelvic region, thoracic and lumbar regions, and shoulder girdle at the age of 14, which eventually caused severe movement limitation with almost complete spontaneous relief of pain in a short period of time. At the age of 20, he had an operation of resection of the bony tissues from the hip and interscapular region, which resulted in much more bone formation at the operation sites.

On his admission, he had a right sided tilt in his neck, walking like a "stone man", with hips 10° abduction and 15° external rotation without any movement in the hips and the shoulder girdles. His mental status was normal.

On his physical examination, he had limited movements in temporomandibular joint, a decrease in the lordosis and the kyphosis of the

vertebral column. There was no movement in the entire vertebral column, except 5° of extension flexion and 45° of lateral rotation of the neck. He had 60° abduction ve 60° flexion with limited amount of rotations. He was able to flex his elbows about 70° and able to rotate about 30° inward and outward. There was no limitation in the movements of the wrist and the phalanges. There was only 45° of flexion in both knees, with full ankylosis of the hips. On palpation his lumbar, gluteal and interscapular regions were as hard as a stone block.

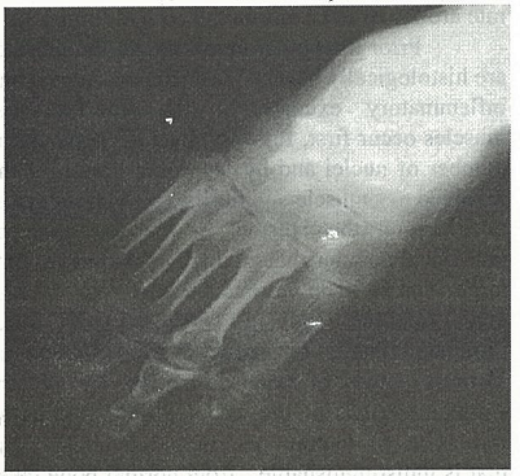

Figure 1:Radiogram of the foot revealed microdactily

On his radiological examination, disseminated multiple, irregular bone tissues within the soft tissue were observed from occiput to sacrum, in the intercostal spaces, hips and thighs and crura. His feet graphs revealed symmetrical microdactily of the great toes and the hypoplasia of the others. Additionally, broadening of the first metatarsal bones, with the aplasia of the proximal phalanges were observed. His 3-D CT revealed heterotopic ossifications along the facial borders of the muscles, located in the regions mentioned above. (Figure 1,2,3,4)

Hearing and cardio-pulmonary functions tests were within normal limits. Upon the certain diagnosis, based on then history, physical examination and radiological findings, the patient was acknowledged about his illness and its prognosis. He was discharged with the necessary recommendations, especially about his pulmonary functions.

DISCUSSION:

Myositis ossificans progressiva, recently known as Fibrodysplasia Ossificans Progressiva, is a rare hereditary mesodermal disorder. There have been at least six sets of homozygous twins with the disease published in literature. It is contributed to be an autosomal dominant disease, either it may be inherited or it may arise as a spontaneous mutation , with full penetrance (no skipped generations) but variable expressivity (variable phenotypic expression of the gene in affected members of the same family) and no sexual or ethnic predilection.(1,2,3,4).

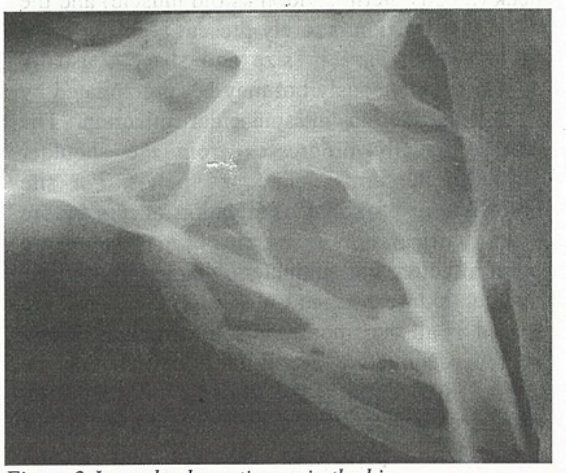

Figure 2:Irregular bone tissues in the hip

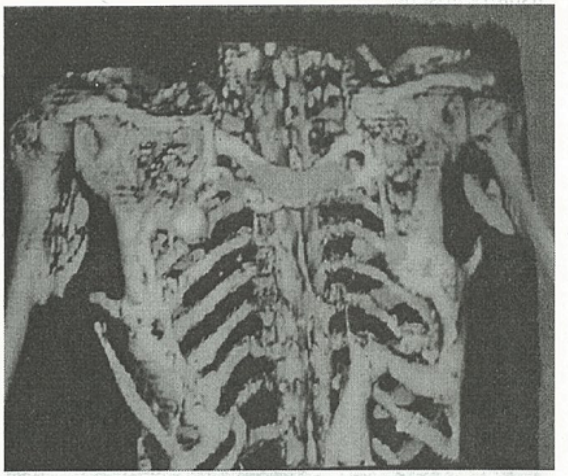

Figure 3:3 D CT of the thorax

Characteristic skeletal abnormality of the feet include microdactyly of the first toes with hypoplasia or synostosis of the phalanges or both. Other congenital anomalies of the skeleton associated with myositis ossificans progressiva are hallux valgus deformities, clinodactyly, broad femoral neck, bilateral thickening of the medial

cortices of the tibiae, an abnormal carrying angle at the elbow, and increased frequency of spina bifida. (4,5,6) The limbs may be short , but this deformity is less prevalent than the toe anomalies. The congenital skeletal abnormalities cause few problems, and the affected child remains asymtomatic until heterotopic ossification begins. This generally begins by age 10 (the average age of onset is 4 years) with a series of firm , painful , asymmetric soft tissue lumps in the muscles of the neck (mostly sternocleidomastoid muscle) and the back. Ossification is rarely present at birth. These lumps which vary in size and shape , usually appear spontaneously but may be precipitated by a trauma like an intramuscular injection. The disease usually progresses from the shoulder girdle to the arms, spine and pelvis. For site predilection, proximal ossification occurs before distal ossification. The muscles of mastication are often affected, but sphincters, diaphragm, larynx, tongue, extraocular muscles and the heart are spared as all the smooth muscle structures. There may be conductive hearing loss, which may be due to ossification of the stapedius muscle or an associated genetic defect. Ocular problems do not occur but premature baldness and mental retardation may sometimes be observed. Systemic sign of disease , such as fever and malaise , are usually absent.

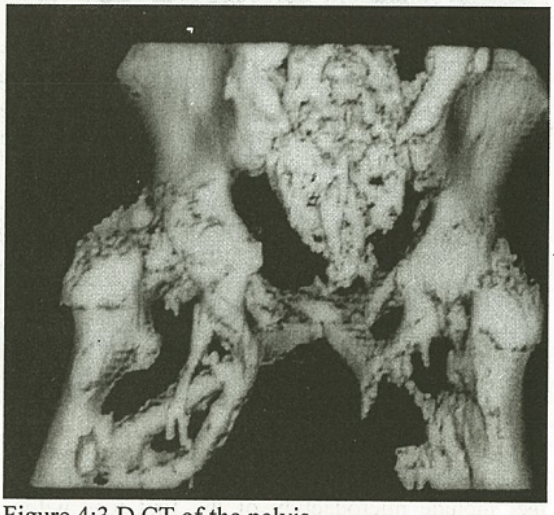

Figure 4:3 D CT of the pelvis

As heterotopic ossification develops in the soft tissues throughout the body , extraarticular ankylosis of the joints occurs, beginning proximally and axially, then progressing distally throughout the appendicular skeleton. Although longitudinal growth is normal, it may be masked by deformities caused by bony ankylosis of the spine and the limbs. Paradoxically, osteoporosis resulting from immobilization may occur as the disease progresses, most notably about ankylosed joints. Fractures of the osteoporotic bone or the heterotopic new bone occur occasionally. (7)

Although clinical and radiographic findings are striking, blood chemistry values, serum alkaline phosphatase concentrations, renal function, parathyroid hormone, serum calcium, phosphate levels, and erythrocyte sedimentation rate are found to be within the normal limits.

Prior to clinical involvement, the muscles are histologically normal. Spontaneous edema and inflammatory exudate of the interfascicular muscles occur first, followed by a decrease in the number of nuclei and hyalen degeneration of the involved muscle fibrils. Mesenchymal proliferation then results in the formation of multifocal nodules that gradually coalesce to form large masses. The lesions become less cellular and replaced by a process of either intramembranous or endochondral ossification. Various stages of bone formation are present simultaneously in the same lesion; finally, formation of mature heterotopic lamellar bone that is indistinguishable from normal bone takes place. The disease process is true ossification not calcification. A dramatic progression takes place beginning with spinal and shoulder movement limitation by 10 years of age, and severe hip movement limitation by the second decade of life. Most patients are confined to a wheelchair by age 30. The average life span is about three or four decades and pneumonia is the usual cause of death. (5,6)

The diagnosis of Fibrodysplasia ossificans progressiva is based on clinical and radiographic findings. There is no effective treatment, although administration of diphosphonates has been advocated. However, this treatment merely delays the mineralization of bone rather than impairing the production of heterotopic osteoid.(8,9) Even simple surgical removal of the ectopic bones causes additional ossification in the scar tissue. Operative intervention may only help a joint to fuse in more functional position. Patients with Fibrodysplasia ossificans progressiva may present problems to the anesthesiologist, including difficulties with tracheal intubation, restrictive pulmonary disease and abnormalities of cardiac conduction.(10) Even in the late stages

of the disease, patients should be considered severely disabled rather than ill. Genetic counseling should be provided to families in which the disease occurs.

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