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FIBRODYSPLASIA OSSIFICANS PROGRESSIVA: A CASE REPORT

PROGRESİF FİBRODİSPLAZİ OSİFİKANS: BİR OLGU SUNUMU

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SUMMARY

Fibrodysplasia Ossificans Progressiva (FOP) or Miyositis Ossificans Progressiva is a genetic disorder with unknown cause. Disease is characterized by heterotopic ossifications of connective tissue and congenital malformations of distal part of extremities.

Our case was a 27 years-old woman who had restrictions of range of motion and disability of daily living aktivities . We have diagnosed according to our physical examination, x-rays and whole body bone scintigraphy as Fibrodysplasia Ossificans Progressiva and rewieved literature about FOP in light of this case.

Key Words: *Fibrodysplasia Ossificans Progressiva, Miyositis Ossificans Progressiva*

ÖZET

Fibrodysplasia Ossificans Progressiva (FOP) or Miyositis Ossificans Progressiva , nedeni bilinmeyen genetic bir hastalıktır. Hastalık, konnektif dokunun heterotopik ossifikasyonları ve extremitelerdeki kongenital malformasyonlar ile karakterizedir.

Bizim vakamız, eklem hareket kısıtlılıkları ve günlük yaşam aktivitelerinde yetersizlik olan 27 yaşında bir kadındı. Biz bu vakaya fizik muayene bulguları, kısıtlı eklemlerin direkt röntgenogramları ve tüm vücut kemik sintigrafisi ile Fibrodysplasia Ossificans Progressiva tanısı koyduk ve bu vaka ışığında konuyla ilgili literatürleri gözden geçirdik.

Anahtar Kelimeler: *Fibrodisplazi Osifikans Progresiva, Miyozitis Osifikans Progresiva*

INTRODUCTION

Fibrodysplasia Ossificans Progressiva (FOP) or Miyositis Ossificans Progressiva is an extremely rare genetic disorder which is inherited autosomal dominantly (1). The cause of FOP is unknown. It is first described by Guy Patin in 1648 (2). Disease is characterized by heterotopic ossifications of connective tissue and congenital malformations of distal part of extremities. Diagnosis is based on clinical observations and radiological findings. There is often a significant delay between the onset of the disease and its diagnosis because it may be confused with infection, bruising or tumor (3). Disease is frequently seen in adolescents and young adults with male predominance (4). No effective medical treatment is available. Surgical treatment is almost always contraindicated, since new heterotopic ossification can develop (5) .

We report a 27 year-old woman with Fibrodysplasia Ossificans Progressiva and rewiev literature about FOP in light of this case.

CASE

We report a 27 year-old woman who admitted to our outpatient clinic with complaints of restrictions of motion in almost all of her joints and being not to be able to sit due to these restrictions. From her history, we have learned that she was able to do her activities of daily living indepently up to 10 years of age. While she was 10 years old , she had fallen on her right arm and her right humerus was fractured. Thereafter, she had developed restrictions of motion in her joints.

In physical examination, we observed ankylosis of lumbar vertebrae and bilaterally short great toes in her feet. She could walk independently , without need of any support with very short step-lenghts. All joints of her body -except wrist jointswere almost totally limited and she had pain on attempt for passive motion. Her mouth hygiene was very bad, with totally limited temporomandibular joint motion and dental carries.

We have done a series of hematologic and biochemical blood and urine analysis. All were within normal limits. We obtained

X-rays of all joints. On these roentgenograms, we have seen hypoplasic and ankylosed cervical vertebrae, ankylosed thoracic and lumbar spine.

In soft tissues around both of shoulder joints, there were new bone formations which bridge over the parts of these joints. Such bridging ossifications were also present around right iliac crest, right ankle and left knee. (Figure 1, 2, 3)

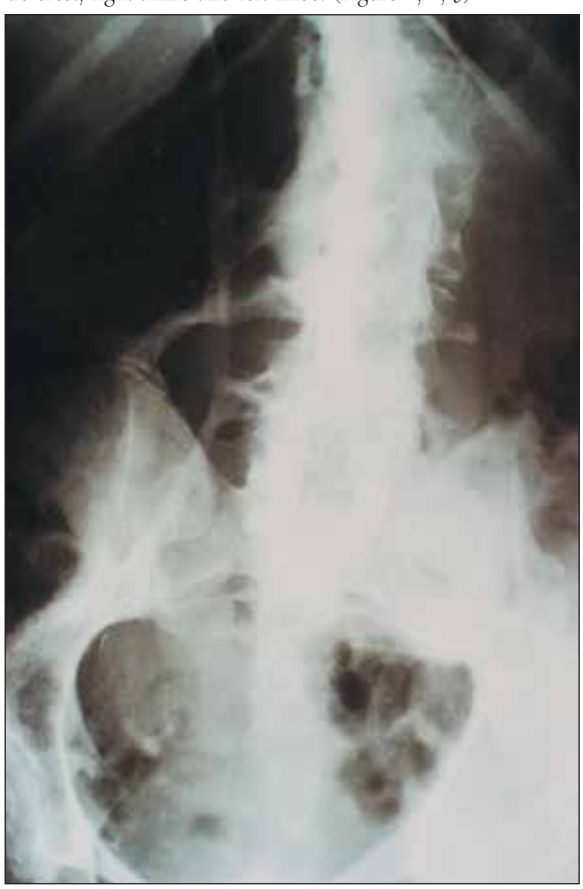

Figure 1. Bridging on right iliac crest.

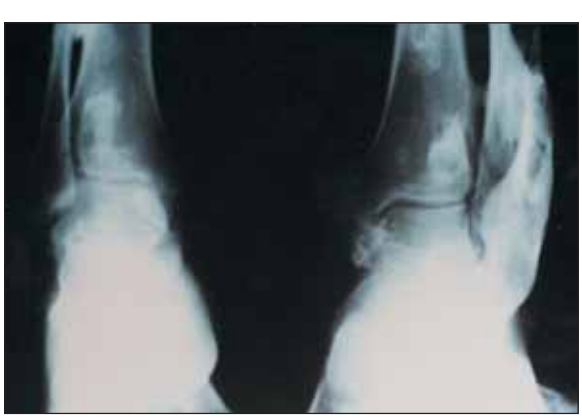

Figure 2. Ossifications on right ankle.

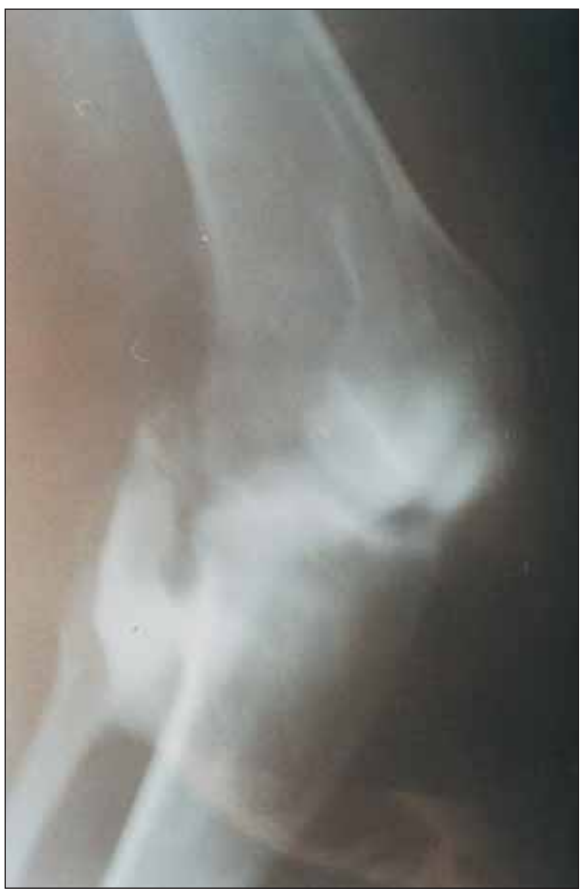

Figure 3. Ossifications on left ankle.

In total body scintigraphy, by using Tc 99M MDP as contrast, we observed increased non homogenous osteoblastic activity around left shoulder joint, 1/3 distal part of right humerus, lower part left scapula and superior part of right iliac crest, in views taken 3 hours after injection of contrast. (Figure 4)

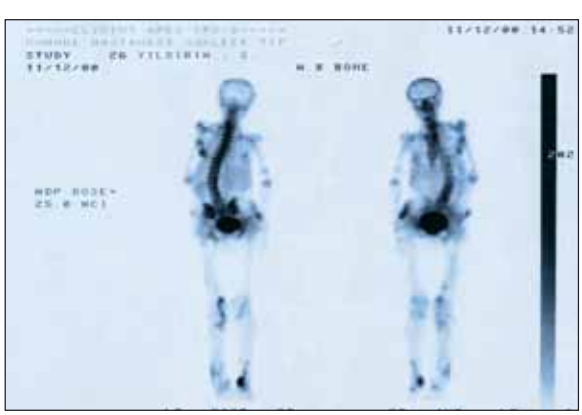

Figure 4. Increased non homogenous osteoblastic activity.

DISCUSSION

With these findings, we have diagnosed the case as Fibrodysplasia Ossificans Progressiva and rewieved literature about FOP in light of this case.

Although FOP is a relatively rare condition, it is well described with characteristic clinical, radiologic and pathologic features (4,6). Disease is actually characterized by progressive replacement of fascia, muscles, tendons and ligaments by bone tissue (3) and is associated with skeletal malformations. The pathogenesis of FOP is not well understood. Trauma has been associated with the majority of cases as in our case , but there are reported cases without an antecedent injury (4).

Although signs may be present at birth, the first appearance of ectopic bone typically occurs in early childhood. As the disease advances, mobility becomes restricted and, affected individuals are limited to bed or wheel-chair by their early 30s (7).

Congenital malformations of great toes is the earliest phenotypic feature of FOP and is present in nearly all affected individuals. The most important abnormality is bilateral short great toes with hallux valgus (8,9,10,11).

Patients have two skeletons: a normotopic one formed during embriyogenesis and a heterotopic one formed after birth. As heterotopic skeletogenesis progresses in characteristic anatomic patterns, maturing heterotopic bone generally forms rigid synostoses with the normotopic skeleton, thus further restricting motion and enhanced disability (12).

Radiographic evaluations are important in diagnosis of disease. But, it has been reported that, radiographs may underestimate the extent and progress of the disease process. By bone scintigraphy , we may evaluate not only the extent and progression of the disease but also the metabolic activity of the already formed bone (3,13,14).

Treatment consists of supportive care, genetic counselling and education regarding the importance of avoiding contact sports and surgical/dental procedures. Corticosteroids, etidronate, radiotherapy and surgery have been used with limited efficacy. Etidronate has been used to prevent recurrence of ectopic ossification after removal of bone, but the author's experience suggests that this is not useful.(2)

FOP is a rare disabling disease and the diagnosis can be made on clinical and radiological findings. In presence of bilateral short great toes with hallux valgus associated with heterotopic ossifications of connective tissue, one should consider FOP as a diagnosis.

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