OLGU SUNUMU CASE REPORT

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Melorheostosis: A Rare Disease

Melorheostozis: Nadir Bir Hastalık

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ABSTRACT Melorheostosis is a rare nongenetic developmental anomaly first described in 1922 by Leri and Joanny. Its etiology is unknown. Patients present at any age, and both sexes are affected equally. Onset is usually insidious, with pain, stiffness and limitation of motion at the affected areas. The characteristic radiographic appearance consists of irregular hyperostotic changes of the cortex, generally on one side of the bone, resembling melted wax dripping down one side of a candle. There is usually a demarcation line between the affected and normal bone. Dense, sclerotic linear areas are seen mainly in the cortex but also extending into the cancellous bone. Melorheostosis affects mainly the long bones of the upper and lower limbs, but also the short bones of the hand and foot and, rarely, the axial skeleton may be affected. Bone scintigraphy is positive and shows moderately increased uptake of tracer in all three phases. Computed tomography and magnetic resonance imaging can further characterize the lesion, but rarely contribute to the diagnosis. Treatment of this chronic condition consists of conservative therapies or surgical soft-tissue procedures and even, in very severe cases, amputation. In this case report, a 53 years old male patient with pain and morning stifness in his left hand for 6 months who has been diagnosed as melorheostosis by radiographic appearence is presented.

Key Words: Melorheostosis; rare diseases; chronic pain; rehabilitation

ÖZET Meloreostoz ilk 1922'de Leri ve Joanny tarafından tanımlanan nadir nongenetik gelişimsel bir anomalidir. Etiyolojisi bilinmemektedir. Hastalar herhangi bir yaşta prezente olabilirler ve her iki cinsiyet eşit olarak etkilenmektedir. Başlangıcı genellikle sinsidir, etkilenen bölgelerde ağrı, sertlik ve hareket kısıtlılığı ile ortaya çıkar. Karakteristik radyografik görünüm özellikle kemiğin tek tarafında irregular hiperosteotik korteks değişikliklerinden oluşur, erimiş mum görüntüsüne benzer. Genellikle etkilenen ve normal kemik arasında bir demarkasyon hattı bulunur. Dens sklerotik lineer alanlar asıl kortekste görülür fakat ayrıca süngerimsi kemiğe uzanabilir. Melorheostozis esas olarak üst ve alt ekstremitenin uzun kemiklerini etkiler fakat ayrıca el ve ayağın kısa kemikleri ve daha nadiren aksiyel iskeleti etkiler. Kemik sintigrafisi pozitiftir ve her üç fazda orta derecede artmış uptake tutulumu gösterir. Bilgisayarlı Tomografi ve Manyetik Rezonans Görüntüleme daha fazla karakterize lezyonu gösterir fakat nadiren tanıya katkıda bulunur. Bu kronik hastalığın tedavisi konservatif tedaviler veya cerrahi yumuşak doku prosedürlerinden oluşmaktadır hatta bazı şiddetli olgularda ampütasyon uygulanmaktadır. Bu olgu raporunda altı aydır sol elinde ağrı ve sabah tutukluğu olan ve radyografik görünümü ile meloreostoz tanısı konulan 53 yaşında erkek hasta sunulmuştur.

Anahtar Kelimeler: Meloreostoz; nadir hastalıklar; kronik ağrı; rehabilitasyon

Melorheostosis, also known as Leri disease or syndrome, is a nongenetic developmental anomaly, one of a group of sclerosing dysplasias of bone; described first by Leri and Joanny in 1922.[1] The etiology of this disorder is unknown.[2] It is often detected on plain radiographs incidentally.[1] In this case report, 53 years old male patient who admitted to our outpatient clinic with pain and morning stifness in his left hand, and who had radiological findings consistent with melorheostosis is presented.

CASE REPORT

A 53 years old male patient who is a dentist admitted to outpatient clinic with complaints of pain and morning stifness lasting 15-20 minutes in his hands for 6 months. He had no history of joint swelling or artralgia. His rheumatologic history was negative and there was no family history or comorbid diseases. The patient's physical examination was normal except a slight thoracolomber scoliosis with a Cobb angle less than 10 degree. There was no swelling, increase in temperature, tenderness, limitation of range of motion, deformity or color change in his left hand, and other joints.

Serum alkaline phosphatase, phosphorus, calcium levels, erythrocyte sedimentation rate (ESR), white blood cell (WBC) in laboratory tests were normal. To rule out rheumatoid arthritis or other inflammatory joint disease which may also be reason of pain and stiffness of the joints; anti-CCP, rheumatoid factor and antinuclear antibodies were tested, and all were negative. 25(OH) vitamin D was low (15.20 ng/mL). In the radiological examination ulnar side of the left 2. metacarpal bone revealed a sclerotic linear area and resembled melted wax dripping down one side of a candle (Figure 1). The patient was diagnosed as melorheostosis based on this characteristic radiographic image. The patient was asked to have a nuclear scintigraphy for advanced imaging but he refused to do it. The patient's informed consent was obtained. He was prescribed vitamin D and 500 mg naproxen two times a day for 10 days. At his first visit (on 1 month control), the patient was pain free and reported that he had no morning stiffness in his hand.

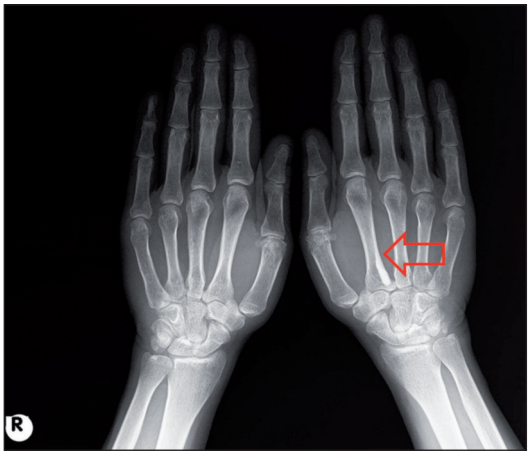

FIGURE 1: Sclerotic linear area in the ulnar side of the left 2. metacarpal bone.

DISCUSSION

Melorheostosis may present at any age between 2 - 64 years and affects both sexes in equally.[3] Onset of the condition is usually insidious and the complaints begin before 20 years old in 50% of patients.[4] The predominant symptoms are joint stiffness and pain and may include deformity of the involved extremity, limitation of motion.[5] Our case also 53 years old and applied with pain and morning stiffness in his left hand. Melorheostosis is a cause of functional morbidity and is not associated with increased mortality. Laboratory findings for serum calcium, phosphorus, and alkaline phosphatase levels are typically normal.[5] These parameters were normal in our case.

The radiographic appearance of melorheostosis is characteristic and it usually reflects developmental errors in intramembranous and endochondral bone formation sites. The appearance consists of irregular hyperostotic cortex changes usually on one side of the bone and resembles melted wax dripping down one side of a candle.[6] In our case ulnar side of the left 2. metacarpal bone revealed a sclerotic linear area and resembled flowing candle wax, the classic appearance of melorheostosis. There is usually a demarcation line between the affected bone and normal bone. The changes are seen mainly in the cortex, however the dense, sclerotic linear areas may extend to the spongiform bone. The predominantly endosteal involvement, marking long bones in a line and staining of small bones are common in children.[7] Ossifications are often observed in the soft tissues of large joints. Although long bones of the lower and upper limbs are affected more frequently, as in our case, short bones of the hands and feet may also be affected.[4,8-10] Melorheostosis rarely affects axial skeleton only.[11]

Bone scintigraphy is always positive in melorheostosis, reveals moderately increased uptake of radiopharmaceutical tracer, mainly localized to the cortex.[12] Our patient was asked to

have a nuclear scintigraphy but he refused to do it. There are very few reports on magnetic resonance imaging (MRI) findings in melorheostosis.[13] Especially in all pulse sequences, there is reduced signal intensity localized to affected bone.[14] Computed tomographic (CT) appearance is similar to the typical undulating hyperostosis seen on radiographs.[14]

The histological features of melorheostosis were reported by several investigators.[5,15] Microscopic examination of cortical specimens reveals nonspecific hyperostotic periosteal bone formation, with fibrotic changes and thickening in trabeculae in the bone marrow spaces.[5]

The differential diagnosis includes several diseases which may have similar radiographic findings such as osteosarcoma, osteomyelitis, osteopetrosis, osteopoikilosis, pycnodysostosis, and osteopathia striata.[10]

The disorder is chronic. A variety of different conservative and surgical treatments have been used in trying to treat the pain and deformities related to melorheostosis. The conservative treatments which are used include oral medications such as bisphosphonates, nonsteroidal antiinflammatory agents and nifedipine. Our case was also given nonsteroidal anti-inflammatory drugs which reduced his complaints. Other nonsurgical treatment modalities include physical therapy, manipulations, braces, serial casting, nerve block, and sympathectomies.[16] Surgical treatment includes soft-tissue procedures such as tendon lengthening, fasciotomy, capsulotomy and fibrous and osseous tissue excision. Other procedures consist of hyperostotic bone excision, corrective osteotomies and even amputation in selected cases. Conservative treatments are typically ineffective in treating severe melorheostosis-related limb deformities and surgical treatments often result in deformity recurrence.

Although the disease can be diagnosed by a plain radiography, it is generally overlooked due to low awareness of the disease. This case is reported to increase the awareness of melorheostosis.

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