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Osteopoikilosis as an Uncommon Cause of Extremity Pain: A Case Report

Ekstremite Ağrısının Nadir Bir Nedeni Olarak Osteopoikilozis: Olgu Sunumu

Emre Fidan[1], Adil Can Karaoğlu[2], Ahmet Özdemir[2]

[1]Balıkesir University Faculty of Medicine, Balıkesir, Türkiye
[2]Balıkesir University Faculty of Medicine, Department of Neurosurgery, Balıkesir, Türkiye

Abstract

Osteopoikilosis, also known as spotted bone disease, is a rare, benign, and inherited sclerosing bone dysplasia characterized by multiple small, well-defined sclerotic foci, predominantly involving the appendicular skeleton. While the etiology remains unclear, it is believed to result from inherited defects in trabecular bone formation along stress lines. Lesions occur symmetrically around the knee joints, pelvic bones, tarsal bones, shoulder girdle, and carpal bones, while involvement of the skull and spine is considered extremely rare. The disease follows an autosomal dominant inheritance pattern and can often be confused with bone dysplasias, bone metastases and other sclerotic diseases on radiological examinations. The diagnosis is usually made incidentally on radiologic examinations. We report the case of a 40-year-old man who presented to our clinic after trauma, with pain in both wrists and the left knee. Radiologic imaging revealed widespread sclerotic lesions, notably involving the cervical, thoracic, lumbar, and sacral vertebrae-an uncommon pattern for osteopoikilosis. In addition to classical appendicular lesions, the extensive axial skeleton involvement distinguished this case from typical presentations.
Keywords: Osteopoikilosis, bone dsyplasia, radiographic sclerosis

Öz

Osteopoikilozis, benekli kemik hastalığı olarak da bilinen, nadir, benign ve kalıtsal bir sklerozan kemik displazisidir. Bu hastalık, çoğunlukla apendiküler iskeleti tutan, çok sayıda küçük, iyi sınırlı sklerotik odaklarla karakterizedir. Etiyolojisi tam olarak aydınlatılamamış olmakla birlikte, stres hatları boyunca trabeküler kemik oluşumundaki kalıtsal defektlerden kaynaklandığı düşünülmektedir. Lezyonlar simetrik olarak diz eklemleri, pelvis kemikleri, tarsal kemikler, omuz kuşağı ve karpal kemikler çevresinde görülürken, kafa ve omurga tutulumunun son derece nadir olduğu kabul edilmektedir. Hastalık, otozomal dominant bir kalıtım modeli izler ve radyolojik incelemelerde sıklıkla kemik displazileri, kemik metastazları ve diğer sklerotik hastalıklarla karıştırılabilir. Tanı genellikle radyolojik tetkiklerde tesadüfen konur. Bu olgu sunumunda, travma sonrası her iki el bileğinde ve sol dizinde ağrı şikayetiyle kliniğimize başvuran 40 yaşında bir erkek hastayı bildirmekteyiz. Radyolojik görüntülemede, servikal, torakal, lomber ve sakral vertebraları da içeren yaygın sklerotik lezyonlar saptanmış olup, bu durum osteopoikilozis için alışılmadık bir tutulum paternidir. Klasik apendiküler lezyonlara ek olarak, aksiyel iskeletteki bu geniş tutulum, olgumuzu tipik osteopoikilozis sunumlarından ayıran en belirgin özelliktir.
Anahtar kelimeler: Osteopoikliosis, kemik displazisi, radyolojik sklerozis

Introduction

Osteopoikilosis (OPK) (OMIM #166700) is an exceptionally uncommon hereditary bone disorder first described by Albers Schönberg in 1915 (1,2). OPK is an osteochondrodysplasia characterized by an increase in bone density without any accompanying changes in bone structure. OPK often does not cause any symptoms; however, in rare cases, it may manifest with dermatological signs. Dermatological manifestations may appear as small, yellowish papules or subcutaneous nodules, which can develop into keloid-like lesions resembling scleroderma. These sores tend to get keloid formations that look like scleroderma. Occasionally, these symptoms may cause pain and effusion in larger joints, although they do not result in any deformities (3). Since most patients don't show symptoms, we can establish the diagnosis using an imaging method for an unrelated cause (4). Radiological imaging reveals the presence of several uniformly distributed sclerotic lesions with distinct boundaries and higher

Corresponding Author/Sorumlu Yazar: Emre Fidan MD, Balıkesir University Faculty of Medicine, Balıkesir, Türkiye
E-mail: emreefidan12@gmail.com **ORCID ID:** orcid.org/0000-0002-7871-771X
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radiodensity (5). The main objective of diagnosis is to exclude OPK as a potential cause, particularly when considering more severe cases such as osteoblastic metastases (6).

Case Report

A 40-year-old man was admitted to our clinic with complaints of pain in both wrists and the left knee after a fall two days ago. He stated that the pain was aggravated by movement and partially relieved by rest and that there was no increase in his complaints at night. No systemic symptoms such as fever, weight loss, or night sweats were reported by the patient. Physical examination revealed tenderness on palpation in both wrists and metacarpophalangeal joints, especially in the left one. Swelling was noted in the left wrist with a positive distal radioulnar joint (DRUJ) test. Range of motion at the wrists and knees was slightly limited due to pain, but no deformity or muscle weakness was noted.

Imaging of the patient showed a cervical spine radiograph showing multiple osteosclerotic lesions involving vertebral bodies and spinous processes, consistent with OPK (Figure 1A). Thoracic and lumbar spine radiograph showing multiple sclerotic foci in the vertebral bodies, indicating axial skeletal involvement (Figure 1B), radiographs of the left hand and wrist showing sclerotic lesions in the carpal bones, metacarpals, and phalanges (Figure 1C). Bilateral knee radiographs show diffuse osteosclerotic areas in the bones surrounding both knee joints, typical of OPK (Figure 1D). A radiograph of the lumbar spine shows prominent sclerotic lesions in the vertebral bodies, more prominent in the thoracic region (Figure 1E). The patient's clinical history, physical examination, and radiologic imaging studies were evaluated together, and a diagnosis of OPK, which is not one of the musculoskeletal diseases we frequently encounter in clinical practice, was made. No surgical procedure was considered; painkillers were prescribed, and rest was recommended.

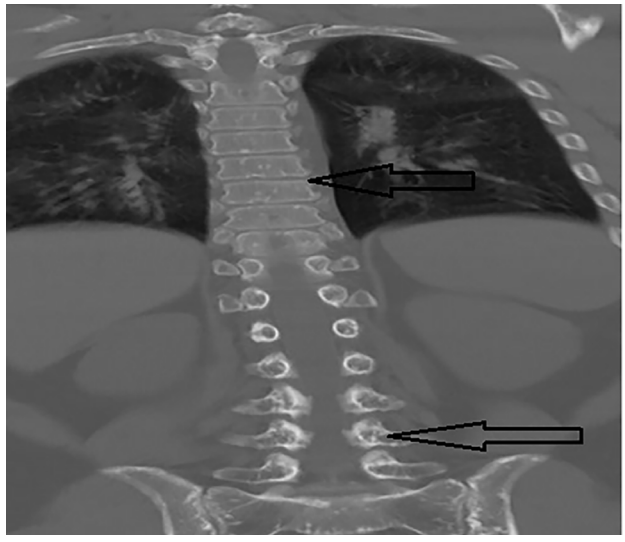

Figure 1B. Thoracic and lumbar spine radiograph showing multiple sclerotic foci in the vertebral bodies, indicating axial skeleton involvement

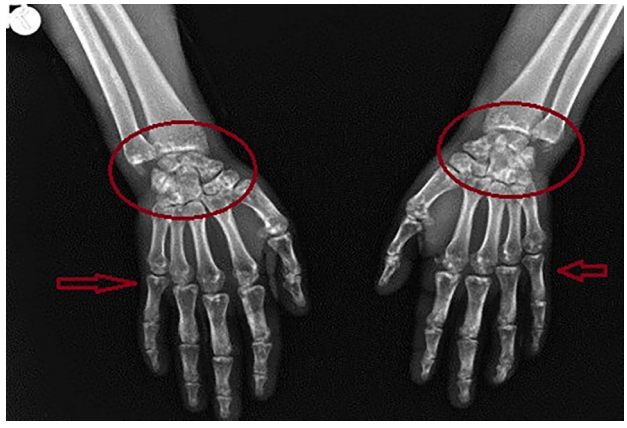

Figure 1C. Radiograph of the left hand and wrist revealing sclerotic lesions in the carpal bones, metacarpals, and phalanges. A positive distal radioulnar joint test was noted on the left side

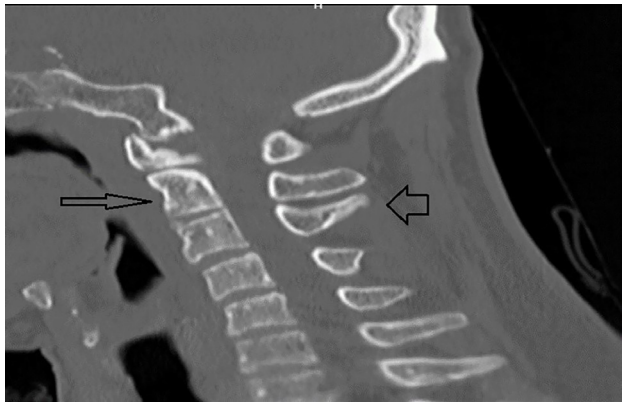

Figure 1A. Cervical spine radiograph demonstrating multiple osteosclerotic lesions involving the vertebral bodies and spinous processes, consistent with osteopoikilosis

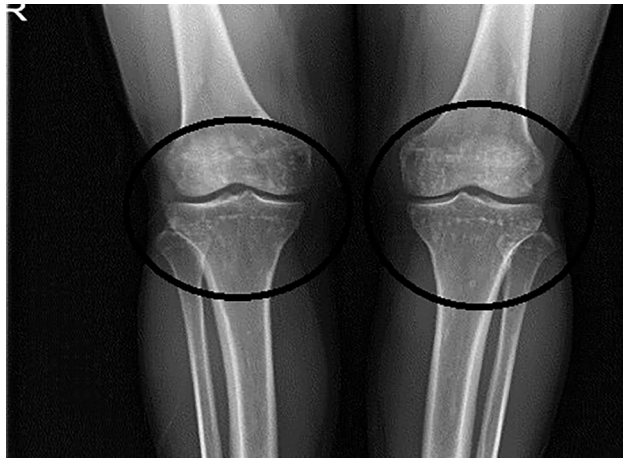

Figure 1D. Bilateral knee radiographs showing diffuse osteosclerotic areas in the bones surrounding both knee joints, typical of osteopoikilosis

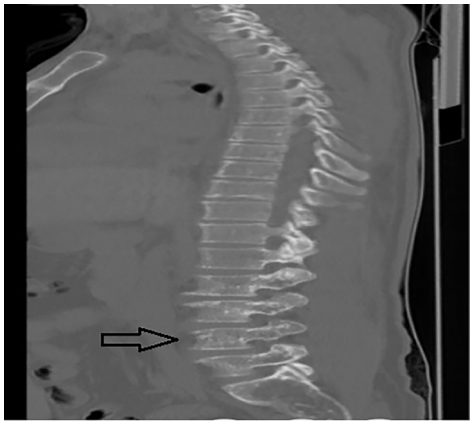

Figure 1E. Lumbar spine radiograph with prominent sclerotic lesions in the vertebral bodies, more pronounced than in the thoracic region

Discussion

When someone has OPK, they will have dense radiolucent spots that are spread out evenly across the epiphyses and metaphyses of long bones. It has a prevalence of roughly 1 in 50,000. Patients typically receive a diagnosis of this condition between the ages of 15 and 60. The lesions are characterized by several circular or oval-shaped patches that are 1-10 mm in diameter and have a hardened texture. These areas can appear in anyone, regardless of age or gender (3,7). Osteopoikilosis is characterized by the presence of a concentrated area of bone that resembles cortical bone and contains Haversian canals. The spongiosa, located directly below the cortex, typically displays this area (1). A lot of the time, osteopoikilosis, osteopathia striata, and melorheostosis happen together. This makes it possible that they are all different symptoms of the same disorder, which is called mixed sclerosing bone dysplasia (8). Researchers have demonstrated that loss-of-function mutations in the *LEM domain containing 3* (*LEMD3*) gene cause conditions characterized by elevated bone density, such as osteopoikilosis, Buschke-Ollendorff syndrome, and melorheostosis (9). A mutation in the *LEMD3* gene genetically links osteopoikilosis (3). While the inheritance of this condition is well-established, there have also been rare instances recorded. Plain radiography and computed tomography scans typically show bone islands near the joints in osteopoikilosis. The orientation of these islands aligns with the surrounding trabecula. Visualization on magnetic resonance imaging. The lesions appear as tiny, dark areas on both T1- and T2-weighted imaging due to their composition of mature, thick bone (4,8). While OPK is a rare condition, it is important to consider other disorders such as tuberous sclerosis, osteopathy striata, osteoblastic metastases, and Ollier disease when making a differential diagnosis. The most crucial concern for OPK is the presence of bone metastases, which are the most prevalent form of malignant bone tumors (5). The epiphyseal and metaphyseal areas of long bones are the most often affected anatomical sites in osteoporosis. The most frequently affected locations include the phalanges (100%), carpal bones (97.4%), metacarpals (92.3%), foot phalanges (87.2%), metatarsal (84.4%), tarsal (84.6%), pelvis (74.4%), femur (74.4%), radius (66.7%), ulna (66.7%), and tibia (20.5%); the ribs are less commonly affected (2,3). The key diagnostic markers include asymmetry, axial skeletal involvement, bone degeneration, and a positive bone scan. Typically, individuals with OPK do not show any symptoms. However, in certain circumstances, patients may experience pain and joint swelling without any specific deformity or malfunction. It does not require any special handling (3,10). Minor joint pain, which may or may not be associated with effusion, is the most commonly reported clinical finding in OPK. In some cases, it may be the sole symptom present. We use non-steroidal anti-inflammatory medicines to alleviate pain (4). Although complications of OPK are uncommon, they may include the development of osteosarcoma, chondrosarcoma, and giant cell tumors. However, it's crucial to acknowledge the lack of a definitive association (2,3).

Conclusion

OPK, also known as spotted bone disease, is an exceptionally uncommon genetic disorder affecting the bones. Several patches of excessive bone growth, typically found near the joints, characterize this condition. The inheritance pattern for this condition is autosomal dominant (6).

However, cutaneous manifestations of OPK may be associated with other conditions like rheumatic or skeletal problems (2). "Although OPK is generally asymptomatic, its radiologic appearance may mimic more serious conditions such as osteoblastic metastases".

In our case, the unusual involvement of the entire axial skeleton, including thoracic, cervical, lumbar, and sacral vertebrae, distinguished it from typical cases of OPK. However, following trauma, finding no other findings except for a fracture of the ulnar styloid, injury to the left radius DRUJ, and pain in the wrist upon palpation. In a patient presenting with pain over the extremities, you can keep in mind the possibility of osteopetrosis and diagnose this patient by direct radiography.

Ethics

Informed Consent: Informed consent was obtained.

Footnotes

Authorship Contributions

Surgical and Medical Practices: A.C.K., Concept: A.Ö., Design: E.F., Data Collection or Processing: E.F., Analysis or Interpretation: E.F., Literature Search: E.F., Writing: E.F.

Conflict of Interest: No conflict of interest was declared by the authors.

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