

Osteopoikilosis: A Cause of Elevated Bone Mineral Density on Dual X-Ray Absorptiometry Measurement in a Young Woman: Case Report

Osteopoikiloz: Genç Bir Kadında DXA Ölçümünde Artmış Kemik Mineral Yoğunluğunun Bir Nedeni: Olgu Sunumu

Asylbek Kaparov, Murat Uludağ*, Hidayet Sarı, Ülkü Akarımak

Istanbul University Cerrahpaşa Medical Faculty, Department of Physical Medicine and Rehabilitation, Istanbul, Turkey

*Harran University Medical Faculty, Department of Physical Medicine and Rehabilitation, Şanlıurfa, Turkey

Summary

Osteopoikilosis (OPK) is an asymptomatic, rare bone dysplasia. It causes an increase in bone density. The etiology and pathogenesis is unknown. OPK is generally diagnosed incidentally on plain radiographies which were performed for other locomotor system symptoms. Diagnostic lesions of OPK are typically diffuse, round, symmetrically shaped sclerotic bone areas. Laboratory findings and bone scintigraphy are usually normal. OPK should be considered in the differential diagnosis of osteoblastic bone disorders. OPK is a benign disease and invasive diagnostic procedures as well as aggressive treatment modalities should be avoided. In young individuals who have elevated scores on dual-energy X-Ray absorptiometry measurement, OPK as well as other sclerosing bone disorders would be considered. (From the World of Osteoporosis 2010;16:25-8)

Key words: Osteopoikilosis, bone mineral density, DXA

Özet

Osteopoikiloz (OPK) nadir görülen, asemptomatik ve kemik yoğunluğunda artışa neden olan bir kemik displazisidir. OPK genellikle diğer lokomotor sistem şikayetleri için yapılan röntgen incelemeleri sırasında tesadüfen saptanır. OPK'nın tanı koydurucu lezyonları tipik olarak yaygın, yuvarlak, simetrik olarak yerleşmiş sklerotik kemik bölgeleridir. Laboratuvar bulguları ve kemik sintigrafisi genellikle normaldir. OPK osteoblastik kemik hastalıklarının ayırıcı tanısında düşünülmelidir. OPK selim bir hastalıktır ve agresif tanı ve tedavi yöntemlerinden kaçınılmalıdır. DXA kullanılarak yapılan kemik yoğunluğu ölçümünde artmış değerler saptanan genç bireylerde diğer sklerotik kemik hastalıkları gibi osteopoikiloz da akılda tutulmalıdır. (Osteoporoz Dünyasından 2010;16:25-8)

Anahtar kelimeler: Osteopoikiloz, kemik mineral yoğunluğu, DXA

Introduction

Osteopoikilosis (osteopathia condensans disseminate, spotted bone; OPK) is an asymptomatic, rare bone dysplasia. This disorder was first described by Alberg Schönberg in 1915. The etiology and pathogenesis remain unclear, but it has been documented to occur as an autosomal-dominant trait (1,2).

OPK is generally diagnosed incidentally on plain radiographies. These diagnostic lesions are typically diffuse, round and symmetrically shaped sclerotic bone areas.

OPK is also a dysplasia characterized with increased bone mineral density (BMD) (3).

Many methods for BMD evaluation exist, with dual-energy radiograph absorptiometry (DXA) being the one used routinely and widely accepted as an accurate, and noninvasive predictor of bone density but it is not economic. DXA can be used to determine the density of various bones in the body but the lumbar spine (L1-L4) and the neck of the femur are most frequently used and validated (4). Falsely high BMD on DXA may be caused by a vertebral wedge or crush fracture, degenerative diseases, Paget's

disease, sclerotic metastases, vertebral hemangioma and other sclerotic bone disorders (5).

We present a case of OPK with elevated BMD of lumbar spine and femur neck on DXA scan in a young female patient.

Case

A 25-year-old woman was admitted to our outpatient clinic with complaints of right wrist and low back pain for 3-4 years. Her pain deteriorated with standing and doing housework. No swelling or redness was found in her right wrist. The patient was overweight (Body mass index (BMI): 29.71 kg/m²). On musculoskeletal examination, range of motion (ROM) of neck was normal. Right wrist dorsiflexion and palmar flexion were painful at the end of the range and Finkelstein test was positive. Tinel and Phalen tests were normal. Lower lumbar interspinous space and bilateral facet joints (L4-5 and L5-S1) were painful with palpation. ROM of low back was minimally restricted and was more painful in extension. FABER, FADIR, sacroiliac compression, femoral, and sciatic stretching tests were normal. Remaining joints were normal. Neurologic examination and muscle strength testing were normal. Complete blood count, erythrocyte sedimentation rate, rheumatoid factor, C-reactive protein, thyroid, kidney and liver function tests, serum calcium, phosphorus, magnesium, alkaline phosphatase, sex hormones, 25 (OH) D₃, and parathyroid hormone levels were normal.

Roentgenograms (X-rays) revealed numerous small round foci of bony sclerosis on radius, ulna, carpal and metacarpal bones (Figure 1) as well as in the pelvis and upper heads of the femur and humerus (Figure 2,3). Magnetic resonance imaging of the right hand showed numerous sclerotic lesions in the distal radius, ulna, carpal bones and metacarpal head.

Bone scintigraphy for whole body with Tc-99m was normal.

BMD measurement was performed using a Hologic QDR and the results were expressed according to the manufacturer's reference range. Bone density of the lumbar spine reveals an L1-L4 measurement of 1.202 g/cm², which is 116% of the age matched normal. This finding corresponds to a T-score of +1.47. Bone density of neck of the left hip is 1.040 g/cm². This finding represents 116% of the age matched normal value. This finding corresponds to a T-score of +1.47.

The patient was treated with nonsteroid anti-inflammatory drug, lumbar flexion exercise and wrist splint with great improvement of her symptoms.

Discussion

OPK is a dysplasia characterized with increased bone mineral density; however a study about quantity of this elevation has not been reported yet. In our patient, DXA scan in the lumbar spine and proximal femur were made and BMD was detected as elevated. In fact, whole body BMD measurement on DXA was more ap-

propriate than in the spine and femur BMD in our young patient.

DXA is an important non-invasive method of bone densitometry measurement. It provides precise and accurate measures of BMD in the spine, hip, wrist, and calcaneus (5). It was accepted as gold standard in BMD measurement, although DXA is not an economic method. To our knowledge, DXA scan has not yet been reported in osteopoikilosis disease.

Intense osteoblastic and osteoclastic activity lead to density changes in affected bone by Paget's disease. Vertebra and/or femur involved by Paget's disease may be detected in patients referred for BMD using DXA (6). Paget's disease as well as OPK also might appear with elevated BMD as related to involved bone.



Figure 1. Anteroposterior roentgenogram of the hands demonstrating small radiodense foci in the proximal radius and ulnae, carpal, metacarpal and proximal phalangeal bones



Figure 2. Bilateral X-ray film of the shoulder showing multiple ovoid radiodense sclerotic foci of the proximal humerus and scapula adjacent to the glenohumeral joint

Vasireddy et al (7) reported three patients who were referred for bone densitometry, in whom a diagnosis of Paget's disease was made after the investigation of significantly increased BMD of a single vertebral body. Shanmugam et al (8) reported a 78-year-old man who has falsely elevated bone density of the total right hip secondary to the presence of Paget disease.

OPK is rare, inherited sclerosing bone dysplasia caused by failure of resorption of secondary spongiosa, which predominantly involves the appendicle and rarely the axial skeleton. The lesions appear to be metabolically active; they become denser with time, but later their size may change (1).

Dysplasia of endochondral ossification, affecting secondary spongiosa, are characterized by errors in resorption or remodeling of secondary spongiosa, which results in focal densities and/or striations within the trabecular bone (3). Some cases are spontaneous, and there is no family history. Prevalence has been estimated 1/50.000. Male and female are equally affected and it may occur at any age (1).

Loss of function mutations of the LEMD3 (MAN1) gene were shown to underlie disorders characterized by increased bone density, namely osteopoikilosis, Buschke-Ollendorff syndrome (BOS), and melorheostosis (9,10). Osteopoikilosis can occur either as an isolated anomaly or in association with other abnormalities of skin and bone. BOS is an autosomal dominant disorder that results from the association of osteopoikilosis with disseminated connective-tissue nevi and manifests as skin lesions. Melorheostosis (often also associated with osteopoikilosis) is characterized by a 'flowing' (rheos) hyperostosis of the cortex of tubular bones and has a 'dripping wax' appearance (9). Our patient was 25 year-old female and was not in contact with any of her family thus the family members could not be traced.



Figure 3. X-ray of the pelvis shows small, roundish, sclerotic lesions at ilea, sacrum, femoral head and around the acetabulum of the pelvis and symphysis pubis without abnormality of the intervening joint spaces

Osteopoikilosis results in numerous round 2-10 mm densities of oval or rounded shape (2,11). Osteopoikilosis is generally diagnosed incidentally on plain radiographies which were performed for other locomotor system symptoms. On radiography, small, well-defined, ovoid foci of increased radio density are clustered in periarticular osseous regions. These little deposits of bone are essentially multifocal bone islands. On histology these foci are formed by dense trabeculae of spongiosa bone, sometimes forming a nidus without communication with bone marrow (11,12).

Sites of predilection include phalanges (100%), carpal bones (97.4%), metacarpals (92.5%), foot phalanges (87.2%), metatarsals (84.4%), tarsal bones (84.6%), pelvis (74.4%), femur (74.4%), radius (66.7%), ulna (66.7%), sacrum (58.9%), humerus (28.2%), tibia (20.5%), and fibula (2.8%) in one study of 4 families. Involvement of the ribs, clavicles, skull and vertebrae is rare and, when present, is less marked (11). In our patient phalanges, metacarpals, carpal bones, pelvis, femur, radius, ulna, sacrum and lumbar spine were involved.

Patients with osteopoikilosis are typically asymptomatic although up to 20% may have mild articular pain and joint effusion (12). Our patient was complained of right wrist and low back pain for 3-4 years. Her pain was deteriorated with standing and housework. We diagnosed tendinitis of the wrist extensors and the mechanical low back pain because improve with conservative treatment.

Cutaneous lesions may be evident in approximately 25% of cases, consisting of closely situated, elevated, whitish fibrocollagenous infiltrations, a predisposition to keloid formation, and skleroderma-like lesions (13). Various developmental malformations have been reported to be associated with osteopoikilosis, including: coarctation of the aorta, double ureter, pubertas praecox, urogenital defects, growth abnormalities, peptic ulcer, diabetes mellitus at the endodermal strata level; arthritis, exostoses, osteitis condensans ilii, Klippel-Feil Syndrome, melorheostosis, spinal stenosis, cervical myelopathy, dacryocystitis, giant cell tumor, fibrous dysplasia, chondrosarcoma, osteosarcoma, synovial chondromatosis at the mesodermal level; facial abnormalities, hare lip, dental abnormalities, dermatofibrosis lenticularis disseminata, keloid formation, plantar and palmar keratomas at the ectodermal level (14-16). We observed none of these malformations in our patient.

The major differential diagnostic considerations in cases of widespread focal round or oval radiodense lesions are osteopoikilosis, osteoblastic metastases, mastocytosis, and tuberous sclerosis. The symmetric distribution, the propensity for epiphyseal and metaphyseal involvement, and the uniform size of the foci are features that suggest osteopoikilosis. A radionuclide bone scan is essential in distinguishing OPK from primary bone tumors or osteoblastic bone metastases. Scan findings are usually normal in patients with OPK but may reveal slightly increased activity (13,17). Bone scintigraphy for whole body with Tc-99m was normal in our patient.

Several rheumatologic pathologies have been reported to coexist with OPK, such as rheumatoid arthritis, lupus erythematosus, reactive arthritis, ankylosing spondylitis and familial Mediterranean fever (13,18-23) but these were not seen in our patient.

Conclusion

Osteopoikilosis is a benign disease and invasive diagnostic procedures as well as aggressive treatment modalities should be avoided. In young persons who have elevated scores in DXA measurement, OPK as well as other sclerosing bone disorders should also be considered. This is the first report of OPK revealing increased spine and femoral neck BMD on DXA measurement.

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