Osteopoikilosis of the axial skeleton and pelvis: A rare cause of back pain

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Abstract:
Osteopoikilosis (OPK) is an autosomal dominant and rare skeletal condition characterized by discrete ovoid or round, multiple radiodense lesions scattered predominantly in the appendicular skeleton. On radiographs, bone metastases and sclerotic bone dysplasias may mimic OPK. However, OPK is an incidental diagnosis. OPK is an asymptomatic condition but rarely may present with slight joint pains and effusions. Radionuclide bone scans have the propensity to distinguish OPK from osteoblastic bone metastases. Here, we present a 49-year-old female with chronic backache, and imaging revealed OPK of the axial skeleton and pelvis.

Keywords:
Enostosis, imaging, osteoblastic metastases, osteopoikilosis, sclerosing bone dysplasia

Osteopoikilosis (OPK) is a benign, autosomal dominant condition categorized as a sclerosing bone dysplasia. Radiographically, OPK demonstrates multiple radiodense lesions which are asymptomatic. This rare sclerosing bone dysplasia was first described by Albers-Schonberg in 1915.[1] Incidence rates are identical in both sexes and may occur in any age group. Pathogenesis of OPK is related to abnormal bone maturation process. Well-defined ovoid and round, symmetric, small, and homogeneous radiodense lesions are the radiological findings of OPK. Pelvis, carpal and tarsal bones, epiphyses of long bones, and scapula are the commonly involved sites.[2] OPK is asymptomatic, but pain could also be a presenting symptom as described in our case report. Principal differential considerations of OPK are lymphoma, Paget’s disease, and osteoblastic metastasis.[3] Our case report reveals that OPK should be kept as a differential diagnosis in patients presenting with diffuse low-back pain to avoid misdiagnosis and invasive diagnostic procedures. Here, we present a 49-year-old female who presented with chronic backache, and imaging revealed OPK of the axial skeleton and pelvis.

Case Report

A 49-year-old female patient complaining of pelvic and back pain persisting for 8 years was referred to the orthopedic department of our hospital. She reported the presence of diffuse back pain mainly in the lower dorsal and lumbar sacral regions and pelvis. Back pain was evaluated clinically which included history and physical examination to evaluate for red flag signs or symptoms of cauda equina syndrome that indicate the need for immediate imaging and further evaluation. Later laboratory tests and imaging were done as a part of the evaluation. She experienced continuous low-quality pain, which worsened on doing physical activities such as climbing stairs and long distance walking. Low-intensity pain rated as 4–5 severity on the Visual Analog Scale. She reported no history of recent or past trauma or the presence of systemic disease. Physical examination revealed no tenderness at the hip joints.
and normal range of motion. There was no visible postural deviation or gait abnormality. Palpatory examination revealed sacroiliac joint tests to be normal with tenderness in the region of symphysis pubis. Laboratory findings for alkaline phosphatase, rheumatoid factor, C-reactive protein, erythrocyte sedimentation rate, calcium, phosphate, complete blood count, serum electrolytes, and thyroid function tests were normal. Inflammatory markers were done to rule out seropositive spondyloarthropathies such as rheumatoid arthritis, polymyalgia rheumatica, and systemic lupus erythematosus as well as seronegative spondyloarthropathies such as ankyllosing spondylitis and psoriatic arthritis. There were no clinical stigmata of inflammatory diseases such as redness, swelling, early morning stiffness, or involvement of small joints of hands and feet. The plain radiograph demonstrated symmetric, sclerotic foci involving the pelvis and lumbar spine without involvement of bone cortex [Figure 1]. Differential diagnoses of sclerotic lesions on plain radiographs are mastocytosis, tuberous sclerosis, striated osteopathy, melorheostosis, osteopetrosis, and osteoblastic metastasis, and in cases with persisting pain, computed tomography (CT) helps in differentiation as OPK is essentially a benign painless condition. On CT imaging of pelvis dorsolumbar spine, the sclerotic lesions were found to be well defined, numerous, 2–10 mm in size with a symmetric distribution. The sclerotic foci were located bilaterally in the epiphyses and metaphyses of long bones in the inner bone cortex. No objectified lumbar spinal stenosis was identified [Figures 2, 3a-f, 4a and b]. The radiological findings were the key findings for the diagnosis of OPK in our case. Treatment required analgesics such as nonsteroidal anti-inflammatory drugs for improvement of pain. She requires pain medications to continue her daily activities to the current day.

### Discussion

OPK is an incidental radiological diagnosis and is characterized by abnormal bone maturation process. The lesions are symmetrically distributed predominantly involving appendicular skeleton, pelvis, carpal, tarsal bones, and periarticular sites of knee and shoulder. Mandible, vertebral bodies, and ribs are less commonly affected with cranial vault involvement being the rarest. Altered osteogenesis is responsible for OPK lesions. Lesions of OPK are asymptomatic; however, mild joint tenderness and effusions have been reported in 15%–20% of patients. Our patient presented with complaints of persistent pain in the lower dorsal and lumbosacral regions and pelvis. Few hypotheses were postulated to give an insight into the mechanisms causing joint pain in patients with OPK- localized increase in bone metabolism at the site of the sclerotic lesion, venous stasis resulting in increased intraosseous pressure at the location of lesion, and joint capsule distension by OPK lesions. Bone scan findings are a reflection of active osseous remodeling resulting in slightly increased activity. Periosteal reaction, variable size, asymmetry, and predominant axial skeleton involvement help differentiate osteoblastic metastases from benign OPK.

OPK is characterized by multiple, <10 mm in size, symmetric round- or oval-shaped radiodense lesions involving metaphyses and epiphyses of long bones, which appear as radio-opaque spots in cancellous bone tissue. It is diagnosed by radiographies with characteristic numerous, symmetric, homogeneous, and circular or ovoid bone lesions. Patients are usually asymptomatic with laboratory tests and bone scintigraphy normal in most cases. OPK commonly requires no treatment other than education and reassurance for patients but should be identified to prevent unnecessary invasive testing and distress.
Bone islands or enostosis are benign, sclerotic, well-defined, and rounded lesions that are frequently isolated and typically small (<10 mm). The margins at the periphery generally tend to merge with the underlying bony trabeculae. The shape of enostosis is not specific, but most commonly the lesions are oval or round. Other sclerotic bone dysplasias must be considered as differentials despite having characteristic peripheral margins, if the lesions are larger than 10 mm in size. The diagnosis of OPK or enostosis may be unlikely when sclerotic lesions are large in size or numerous.

Multiple nonhomogeneous lesions are a major diagnostic dilemma as they may demonstrate osteoblastic activity such as metastasis related to marrow replacement of the involved bones. Metastases do not have a periarticular distribution, may affect any bone, and predominantly tend to involve the axial skeleton. Pelvis and spine are more frequently affected by metastases. Reviewing prior imaging studies is necessary to demonstrate the appearance of a new lesion or whether a lesion has remained stable over time. Bone scintigraphy studies have a crucial role in distinguishing osteoblastic bone metastases from OPK in patients with a known or suspected primary malignancy, when previous images are not available. OPK lesions appear classically inactive on scintigraphy although they may have the appearance of sclerotic metastatic on radiographs and CT.

Sclerotic bone dysplasias of endochondral bone formation such as enostosis, pyknody sostosis, osteopathia striata (Voorhoeve disease), and osteopetrosis (Albers-Schönberg disease) are considered as differential diagnoses of OPK. However, every sclerotic bone condition has varying and characteristic radiologic appearances. OPK is generally asymptomatic and is incidentally diagnosed. OPK may be confused with...
sclerosing bone dysplasias and sclerotic bone metastases. Nevertheless, the tendency for metaphyseal and epiphyseal involvement, the uniform size of the lesions, and symmetric distribution are unique characteristics for diagnosing OPK. Besides, bone scintigraphy of OPK is commonly normal, while slightly higher activity of the bone metabolism can be detected sometimes. Tuberculous sclerosis and skeletal metastasis are characterized by usual involvement of axial skeleton, especially spine, asymmetric distribution, diversity in size, and common osseous destruction with positive scintigraphic findings, which means that bone scintigraphy is usually crucial for distinguishing osteoblastic bone metastases and primary bone tumors from OPK.[11] Our patient did not undergo bone scintigraphy because of typical imaging features of OPK. Due to the benign nature of OPK, complications are very rare. Possible complications described in the literature are osteosarcoma, giant cell tumor, and chondrosarcoma.[12]

**Conclusion**

OPK is characterized by circular or oval-shaped sclerotic bone lesions that most frequently involve the hands, feet, pelvis, ends of long bones, and rarely, spine. OPK may sometimes present with pain in patients with characteristic radiologic findings; however, it is considered a benign condition with asymptomatic natural variation. It can be mistaken for metastatic bone tumors or other diseases because of similar imaging characteristics, which may result in distress to the treating physician and patient. Radionuclide bone scans play a crucial role in distinguishing OPK from metastatic bone tumors.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

**References**

1. Ayling RM, Evans PE. Giant cell tumor in a patient with osteopoikilosis. Acta Orthop Scand 1988;59:74-6.
2. Agostinelli JR. Osteopoikilosis. A case report. J Am Podiatry Assoc 1983;73:529-31.
3. Young LW, Gershman I, Simon PR. Radiological case of the month. Osteopoikilosis: Familial documentation. Am J Dis Child 1980;134:415-6.
4. Benli IT, Akalin S, Boysan E, Muncu EF, Kiş M, Türköghlu D. Epidemiological, clinical and radiological aspects of osteopoikilosis. J Bone Joint Surg Br 1992;74:504-6.
5. Roberts NM, Langtry JA, Branfoot AC, Glesson J, Staughton RC. Case report: Osteopoikilosis and the Buschke-Ollendorff syndrome. Br J Radiol 1993;66:468-70.
6. George P, Athanasios R, Marios S, Sofia S, Basileios P. Osteopoikilosis: A case report of a symptomatic patient. Musculoskelet Radiol 2009;3:38-43.
7. Borman P, Ozoran K, Aydoğ S, Coşkun S. Osteopoikilosis: Report of a clinical case and review of the literature. Joint Bone Spine 2002;69:230-3.
8. Lagier R, Mbakop A, Bigler A. Osteopoikilosis: A radiological and pathological study. Skeletal Radiol 1984;11:161-8.
9. Mungovan JA, Tung GA, Lambiasse RE, Noto RB, Davis RP. Tc-99m MDP uptake in osteopoikilosis. Clin Nucl Med 1994;19:6-8.
10. Khot R, Sikarwar JS, Gupta RP, Sharma GL. Osteopoikilosis: A case report. Indian J Radiol Imaging 2005;15:453-4.
11. An YS, Yoon JK, Lee MH, Jou CW, Yoon SN. Abnormal bone scan in an adult with osteopoikilosis. Clin Nucl Med 2004;29:856-8.
12. Mindell ER, Northup CS, Douglass HO Jr. Osteosarcoma associated with osteopoikilosis. J Bone Joint Surg Am 1978;60:406-8.