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Clinical Image

Osteopoikilosis: spotted bones

Süleyman Yalçın[1*]

[1]Department of Orthopedics and Traumatology, University of Health Sciences Kayseri City Training and Research Hospital, 38080, Kayseri, Turkey

[*]**Corresponding author:** Süleyman Yalçın, MD, Department of Orthopedics and Traumatology, University of Health Sciences Kayseri City Training and Research Hospital, 38080, Kayseri, Turkey; syalcinn00@gmail.com

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Abstract

Osteopoikilosis is a rare, benign sclerosing bone dysplasia characterized by multiple, symmetric, well-defined sclerotic lesions primarily located in the epiphyseal and metaphyseal regions of long bones, pelvis, and hands. Although usually asymptomatic and incidentally discovered, its radiographic appearance may mimic osteoblastic metastases, leading to diagnostic confusion. Accurate recognition of imaging features is essential to avoid unnecessary invasive procedures and to provide appropriate patient reassurance.

Keywords: Osteopoikilosis, spotted bones, melorheostosis, Leri's disease, Buschke-Ollendorff syndrome

Description

Osteopoikilosis (OPK) is a rare, benign sclerosing bone dysplasia first described by Albers-Schönberg in 1915 [1,2]. It is commonly diagnosed incidentally during radiographic evaluation [1-3]. The disorder is defined by numerous, small (typically 2–10 mm), round or ovoid sclerotic lesions that are symmetrically distributed in the epiphyseal and metaphyseal regions of long bones, pelvis, scapula, hands, ankle, and feet [1-3]. The estimated prevalence is approximately 1 in 50,000 individuals [1-3].

Genetics and Pathogenesis

OPK is usually inherited in an autosomal dominant pattern. The underlying pathophysiology involves loss-of-function mutations in the LEMD3 gene, which encodes an inner nuclear membrane protein that regulates TGF-β and BMP signaling pathways [1-3]. Mutations in this gene are also associated with melorheostosis (Leri's disease) and Buschke–Ollendorff syndrome [1-4].

Melorheostosis is an extremely rare sclerosing hyperostosis that typically affects the appendicular skeleton in a limited, segmental manner [3]. It sometimes occurs in the setting of benign generalized sclerosing bone disease known as osteopoikilosis, caused by germline mutations in LEMD3, which encodes the inner nuclear membrane protein MAN1, which regulates TGFβ/bone morphogenetic protein signaling [3].

Buschke-Ollendorff syndrome (BOS) is an autosomal dominant connective tissue disorder characterized by numerous subcutaneous nevi or nodules [4]. Histological examination reveals lesions rich in elastin (elastoma) or rich in collagen (dermatofibrosis lenticularis disseminata) [4]. Lesions are usually non-tender and firm. Affected individuals also have osteopoikilosis (OPK) [4].

Clinical Features

Most individuals with OPK are asymptomatic [1-3]. When present, clinical manifestations may include joint pain or decreased range of motion. In rare cases, dermatologic findings such as dermatofibrosis lenticularis disseminata (as seen in Buschke–Ollendorff syndrome) may coexist [4]. Very rarely, spinal canal stenosis, enthesopathy, or dermatomyositis-like symptoms may occur [3,4].

Imaging Findings

The hallmark of OPK is its distinct radiographic appearance [1-3]. Plain radiographs typically reveal multiple, well-circumscribed, homogeneous, symmetric sclerotic lesions ranging from 2–10 mm in diameter [1-4]. These are usually found in the epiphyseal and metaphyseal regions of long bones, pelvis, hands, and feet [1-4]. Computed tomography (CT) and magnetic resonance imaging (MRI) demonstrate dense bone islands without associated soft tissue mass or bone marrow edema. The phalanges, carpal bones, femur, and pelvis are among the most frequently involved sites [1-4].

Diagnosis and Differential Diagnosis

Diagnosis of OPK is based on its characteristic imaging features [1-3]. Bone scintigraphy is typically normal, a key finding that helps to distinguish OPK from osteoblastic metastases [3]. Differential diagnoses include melorheostosis, osteopathia striata, tuberous sclerosis, and Paget's disease [1-4].

Management and Follow-up

OPK is a benign and non-progressive condition. It does not carry any risk of malignant transformation [1-4]. Treatment is not required in asymptomatic cases. For patients experiencing pain, conservative management with NSAIDs or physical therapy

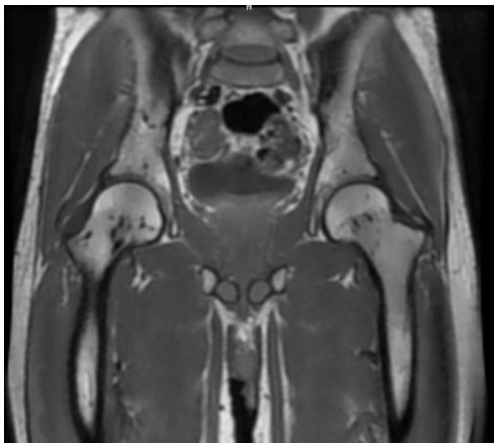

Figure 1. Coronal T1-weighted magnetic resonance imaging (MRI) demonstrates multiple, well-defined, hypointense sclerotic lesions involving the epiphyseal and metaphyseal regions of both proximal femurs and the pelvis. These lesions are characteristic of osteopoikilosis and were initially misinterpreted as potential osteoblastic metastases due to their distribution and density.

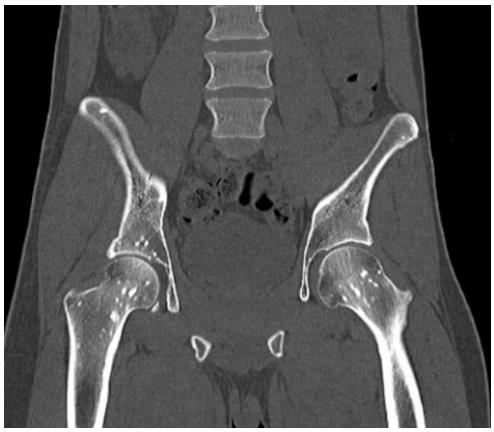

Figure 2. Coronal computed tomography (CT) image of the pelvis demonstrates numerous, small, well-demarcated, high-attenuation sclerotic bone islands located in the epiphyseal and metaphyseal regions of both proximal femurs and acetabula. These findings are typical for osteopoikilosis and contributed to the initial radiologic suspicion for osteoblastic metastases in this patient.

may be considered [1-4]. Accurate diagnosis is crucial to prevent unnecessary invasive procedures and misclassification as metastatic disease [1-4].

Case Presentation

A 27-year-old male presented to a physical medicine and rehabilitation clinic with complaints of persistent lower back and hip pain for several months. His personal and family history was negative for skeletal dysplasias or malignancy. MRI and CT scans revealed multiple sclerotic bone islands predominantly in the pelvis and proximal femur, raising initial suspicion for osteoblastic metastases (Figures 1, 2). Physical examination revealed full range of motion in all joints, and no neurological

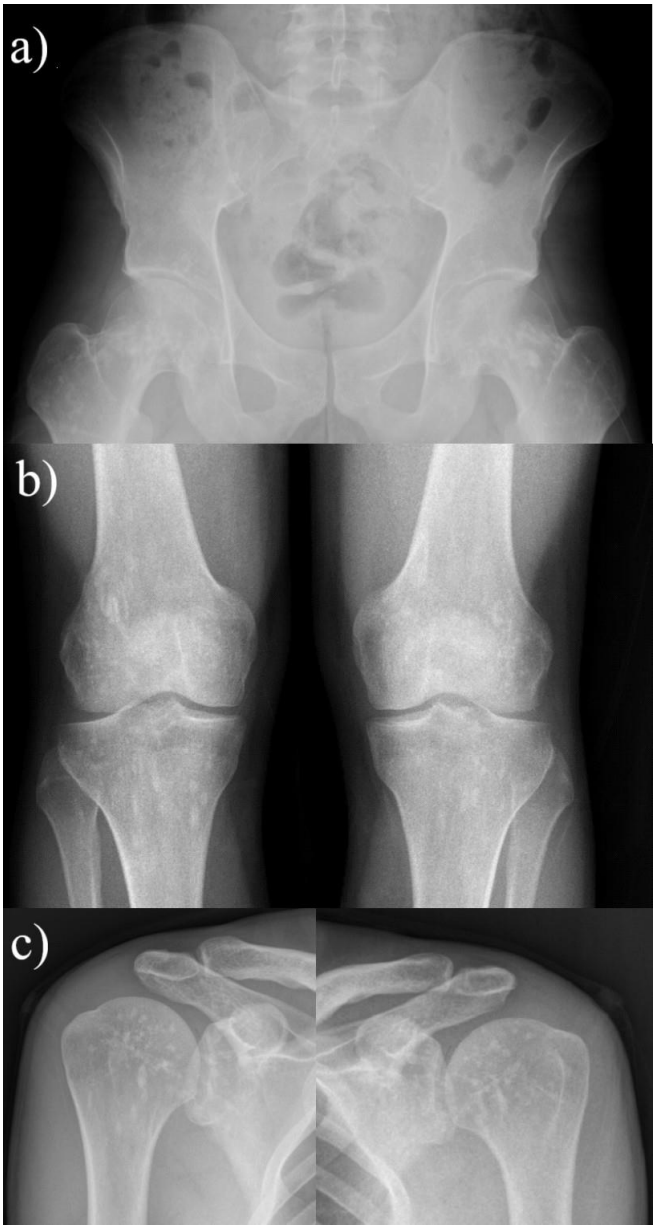

Figure 3. Representative plain radiographs from multiple anatomical regions illustrate the characteristic radiologic distribution of osteopoikilosis, **a)** anteroposterior radiograph of the pelvis reveals multiple, small, well-circumscribed sclerotic lesions symmetrically involving the iliac bones, acetabula, and proximal femurs, **b)** anteroposterior radiographs of both knees display multiple, well- defined, round-to-ovoid sclerotic bone islands in the distal femoral and proximal tibial epiphyses and metaphyses, **c)** bilateral shoulder radiographs show numerous, rounded sclerotic foci in the proximal humerus and scapula on both sides.

deficits or systemic symptoms were noted. Plain radiographs of the pelvis and long bones demonstrated numerous, symmetrically distributed, well-defined sclerotic lesions ranging from 2 to 10 mm in diameter, mostly located in the epiphyseal and metaphyseal regions (Figure 3). Bone scintigraphy was not performed due to the lack of malignancy indicators. A diagnosis of osteopoikilosis was

established based on the imaging findings and absence of systemic involvement. The patient was educated about the benign nature of the condition and scheduled for routine outpatient follow-up.

Learning points

- Osteopoikilosis is a rare but benign bone dysplasia that may radiographically mimic osteoblastic metastases, particularly when found incidentally in symptomatic patients.
- Recognition of its characteristic imaging pattern—well-defined, symmetrical, sclerotic lesions in epiphyseal and metaphyseal regions—is essential for accurate diagnosis and to avoid unnecessary invasive procedures.
- Normal bone scintigraphy and absence of systemic or laboratory findings for malignancy are critical in differentiating osteopoikilosis from metastatic disease.
- Osteopoikilosis is genetically linked to LEMD3 mutations, which are also involved in Buschke–Ollendorff syndrome and melorheostosis, and may occasionally present with dermatologic or musculoskeletal symptoms.
- Management is generally conservative, and patient education about the benign and non-progressive nature of the disease is fundamental to prevent overtreatment or anxiety.

Author contributions

Author contributed to the study conception and design. Material preparation, data collection and analysis were performed by SY. The first draft of the manuscript was written by SY and the author commented on previous versions of the manuscript. The author read and approved the final version of the manuscript.

Statements and declarations

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Conflict of Interest

The authors declare that they have no conflict of interest.

Ethical statement

The authors confirm that this retrospective study was conducted in accordance with the ethical standards set forth in the Declaration of Helsinki and its later amendments.

ORCID iD

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References

1. Panchal SR, Gawhale S, Shah NZ, Mohanty T. Case report of osteopoikilosis: rare cause of bone pain and mimicker of metastasis on radiographs. J Orthop Case Rep. 2021;11(3):98–101. doi:10.13107/jocr.2021.v11.i03.2106
2. Ruci V, Serani D. Osteopoikilosis - case report. Open Access Maced J Med Sci. 2014;2(1):132–4. doi:10.3889/oamjms.2014.024
3. Wordsworth P, Chan M. Melorheostosis and Osteopoikilosis: A Review of Clinical Features and Pathogenesis. Calcif Tissue Int. 2019;104(5):530–43. doi: 10.1007/s00223-019-00543-y.
4. Hassikou H, Tabache F, Safi S, Baaj M, Hadri L. Buschke-Ollendorff syndrome. Joint Bone Spine. 2008;75(2):212–4. doi: 10.1016/j.jbspin.2007.04.027.