Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Radiology

Osteopoikilosis: Multiple Joint Involvement as an Incidental Imaging Finding in the Emergency Setting

M. Mekouar[1*], I. Akhiyat[1], Y. Bouktib[1], A. elhajjami[1], B. Boutakiout[1], M. Ouali Idrissi[1], N. Cherif Idrissi[1]

[1]Department of Radiology, Arrazi Hospital, Mohammed VI University Hospital Center, Marrakech, Morocco

DOI: https://doi.org/10.36347/sjmcr.2025.v13i05.122 | **Received:** 19.04.2025 | **Accepted:** 26.05.2025 | **Published:** 29.05.2025

***Corresponding author:** M. Mekouar
Department of Radiology, Arrazi Hospital, Mohammed VI University Hospital Center, Marrakech, Morocco

Abstract | **Case Report**

Osteopoikilosis is a rare, benign sclerosing bone dysplasia with autosomal dominant inheritance. Characterized by multiple small, well-defined sclerotic bone islands, it is typically discovered incidentally on imaging. This report presents a case of incidental osteopoikilosis in a patient undergoing CT for suspected pulmonary embolism.

Keywords: Osteopoikilosis, Sclerosing bone dysplasia, Incidental finding, LEMD3 gene, Periarticular lesions..

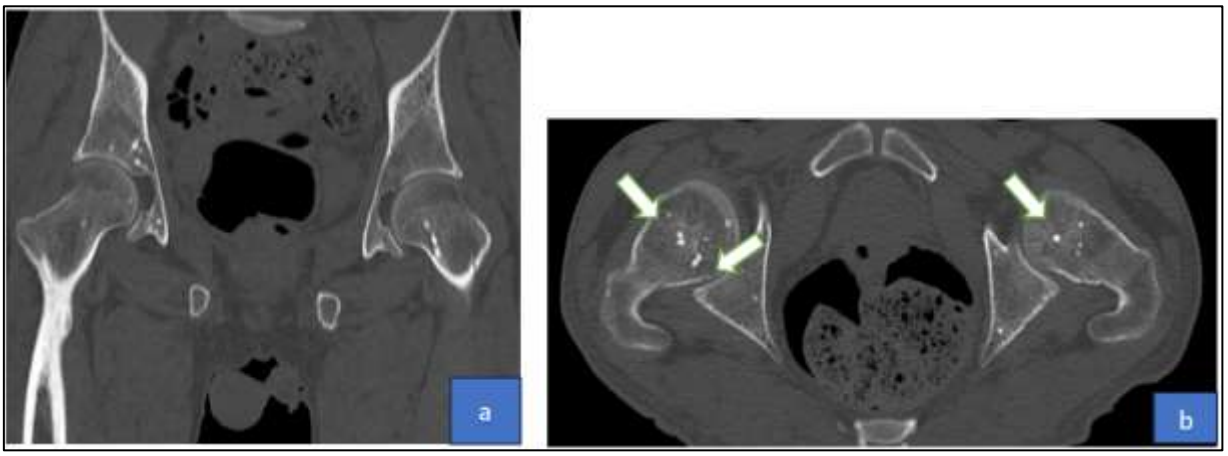
INTRODUCTION

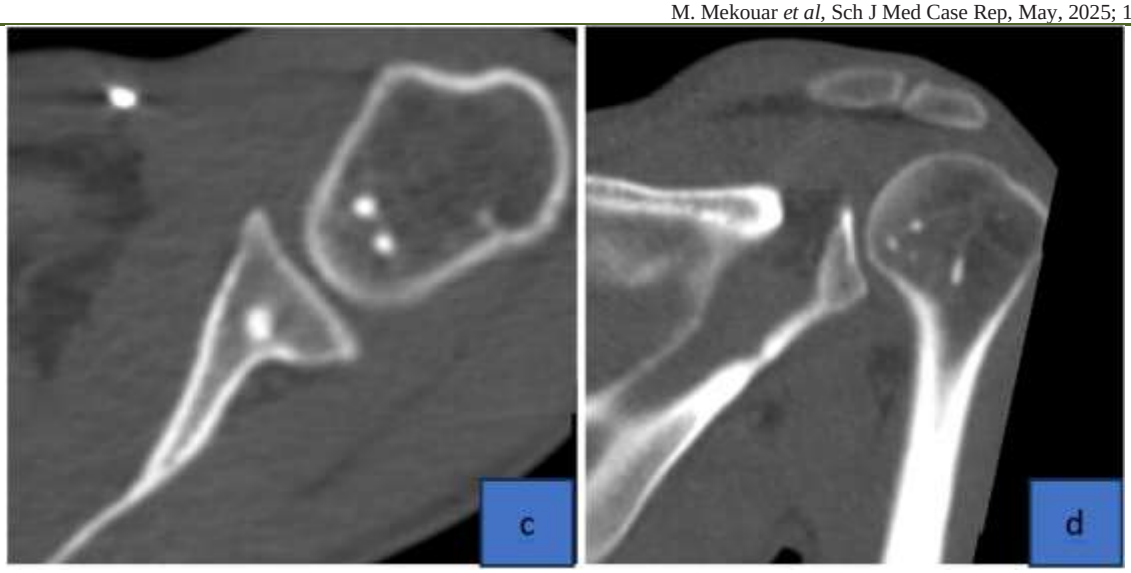
Osteopoikilosis (OP), also termed "spotted bone disease" or osteopathia condensans disseminata, is a non-progressive sclerosing bone dysplasia. Though commonly inherited, sporadic cases occur. The prevalence is about 1 in 50,000, with no sex predominance [1]. Histologically, OP involves compact lamellar bone deposits within trabecular bone [2]. Mutations in the *LEMD3* gene are implicated, disrupting normal bone remodeling [3].

Lesions appear as numerous small sclerotic foci in periarticular locations—most often in the appendicular skeleton, pelvis, and carpal/tarsal bones. OP is typically asymptomatic but may cause mild pain or swelling in 15–20% of patients [4].

CASE PRESENTATION

A 48-year-old male with a history of silicosis and prior pulmonary tuberculosis presented to the ED with chronic respiratory symptoms. CT pulmonary angiography excluded embolism but revealed fibrotic changes of silicosis and signs of infection. Incidental findings on the bone windows showed symmetrical, well-circumscribed sclerotic nodules in the glenohumeral, vertebral facet, coxofemoral, and sacroiliac joints. The imaging appearance was consistent with osteopoikilosis.





Bilateral hip computed tomography with coronal (a) transverse cross-section passing through the femoral head (b). White arrows show numerous hyperdense lesions that conform with the osteopoikilosis in the femoral head; lesions are well-circumscribed and have size in millimeters. Similar lesions are found in the shoulder joints (c) et (d)

DISCUSSION

Osteopoikilosis (OP) is a rare, benign, sclerosing bone dysplasia most commonly inherited in an autosomal dominant fashion, though sporadic cases also exist. It results from mutations in the *LEMD3* gene, affecting normal bone remodeling and leading to the persistence of dense lamellar bone foci within the spongiosa [1,3]. Radiographically, OP is characterized by small, well-defined, symmetrical sclerotic lesions predominantly located near joints, especially in the epiphyses and metaphyses of long bones, pelvis, and carpal/tarsal bones [2,5]. These lesions are hyperdense on CT (usually >885 HU), low signal on both T1- and T2-weighted MRI, and generally show no uptake on bone scintigraphy [4,6]. The imaging presentation may mimic osteoblastic metastases, but the absence of cortical destruction, uniform lesion size, and symmetry, along with the typical periarticular location, helps differentiate OP [8]. Other differential diagnoses include melorheostosis, osteopathia striata, Erdheim-Chester disease, Paget disease, and osteoid osteoma, each having distinctive radiologic and clinical features [7,8]. Although OP is largely asymptomatic, about 15–20% of patients may report mild joint discomfort or effusion [4]. The condition may coexist with other skeletal or dermatologic disorders such as Buschke-Ollendorff syndrome and, rarely, with malignancies [9,10]. Given its benign and stable nature, OP is considered a "don't touch" lesion that requires no treatment, and management is usually conservative, focusing on symptom control when needed. Recognizing its hallmark radiologic features is crucial to avoid misdiagnosis, unnecessary investigations, or invasive interventions.

CONCLUSION

Osteopoikilosis is a benign skeletal dysplasia often discovered incidentally. Recognition of its characteristic imaging features—symmetrical, periarticular bone islands with high attenuation and no aggressive features—is essential to distinguish it from serious conditions like metastases. Proper diagnosis can prevent unnecessary biopsies and alleviate patient concern.

REFERENCES

1. Hellemans J, Preobrazhenska O, Willaert A, et al. Loss-of-function mutations in *LEMD3* result in osteopoikilosis, Buschke-Ollendorff syndrome, and melorheostosis. Nat Genet. 2004;36(11):1213–1218.
2. Greenspan A. Sclerosing bone dysplasias—a target-site approach. Skeletal Radiol. 1991;20(8):561–583.
3. Mumm S, Wenkert D, Zhang X, et al. Deactivating mutations in *LEMD3* in osteopoikilosis and Buschke-Ollendorff syndrome. Am J Med Genet A. 2007;143A(4):367–373.
4. Yao L, Stanczyk P, Done S. Radiologic-pathologic correlation: osteopoikilosis. Radiographics. 2001;21(Spec No):S261–S264.
5. Benli IT, Akalin S, Boysan E, et al. Epidemiological, clinical and radiological aspects of osteopoikilosis. J Bone Joint Surg Br. 1992;74(4):504–506.
6. Cotten A, Flipo RM, Chastanet P, et al. Bone scintigraphy in osteopoikilosis: 4 cases. Clin Rheumatol. 1990;9(2):235–238.
7. Ramesh P, Manickam G, Venkatesan M, Rajarajan A. Osteopoikilosis: a case report and review. Int J Anat Radiol Surg. 2018;7(4):RO23–RO25.

8. Anderson SE, Huang J, Kellenberger CJ, et al. Radiographic characteristics of osteoblastic metastases: correlation with bone scan. Radiology. 2005;237(1):205–212.
9. Hamdan TA, Shousha M, Al-Ani FS, et al. Osteopoikilosis: case report and review of literature. Eur J Orthop Surg Traumatol. 2013;23(Suppl 2):S199–S203.
10. DeFazio MV, Beauchamp CP, Gitelis S. Osteopoikilosis with malignant transformation: a case report. Clin Orthop Relat Res. 2012;470(1):281–285.