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OSTEOPOIKILOSIS: AN INCIDENTAL RADIOLOGICAL DIAGNOSIS IN A WOMAN PRESENTING WITH DIFFUSE BONE PAIN – A CASE REPORT

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AUTHOR AND AFFILIATION

Ibtissam El Bqaq ¹, Sarah Lembarki ¹, Salma El Kadiri ¹, Hassan Ennouali ¹, Jamal El Fenni ¹, Rachida Saouab ¹

¹ Department of Radiology, Mohammed V Military Teaching Hospital (Hôpital Militaire d'Instruction Mohammed V), Rabat, Morocco

Corresponding author: Ibtissam El Bqaq

ABSTRACT

Osteopoikilosis is a rare, benign sclerosing bone dysplasia characterised by multiple small foci of cortical (lamellar) bone within the medullary cavity, typically discovered incidentally on imaging. Although classically asymptomatic, it may occasionally be associated with non-specific musculoskeletal pain, and its radiological appearance can closely mimic osteoblastic bone metastases, creating a significant diagnostic challenge. We report the case of a 53-year-old woman with no significant past medical history who presented with diffuse bone pain.

She had no cutaneous lesions suggestive of an associated connective tissue disorder.

Computed tomography, performed as part of the diagnostic work-up, revealed innumerable well-defined sclerotic lesions of variable size distributed throughout the axial and appendicular skeleton. No conventional radiograph was available for comparison. Laboratory investigations were unremarkable. Given the diffuse nature of the findings and the absence of a baseline radiograph, a bone biopsy was performed to exclude a malignant sclerotic process; histopathological examination confirmed a benign lesion, consistent with osteopoikilosis. The patient was managed conservatively with clinical surveillance. This case underscores the importance of recognising the characteristic imaging pattern of osteopoikilosis to avoid unnecessary invasive investigations, while also illustrating that tissue confirmation may still be warranted when the diagnosis is made on cross-sectional imaging alone, without a corroborating radiograph.

KEYWORDS

Osteopoikilosis, Sclerosing bone dysplasia, Bone islands, Enostosis, LEMD3

MAIN ARTICLE

INTRODUCTION

Osteopoikilosis is a rare, benign sclerosing bone dysplasia first described by Albers-Schönberg in 1915, characterised by multiple discrete foci of compact (lamellar) bone, known as bone islands or enostoses, scattered throughout the skeleton [1]. It is inherited in an autosomal dominant pattern and results from heterozygous loss-of-function mutations in the LEMD3 (MAN1) gene, which encodes an inner nuclear membrane protein that antagonises transforming growth factor- β (TGF- β) and bone morphogenetic protein (BMP) signalling [2]. The same gene defect underlies Buschke-Ollendorff syndrome, in which osteopoikilosis coexists with cutaneous connective tissue naevi (dermatofibrosis lenticularis disseminata); however, isolated osteopoikilosis without skin involvement is more commonly encountered in clinical practice [2,3].

The condition is usually asymptomatic and discovered incidentally on imaging performed for unrelated reasons, though mild arthralgia or bone pain has occasionally been reported [3,4]. Its principal clinical importance lies in its radiological resemblance to osteoblastic bone metastases, a distinction that carries significant implications for patient management [5]. We report a case of osteopoikilosis diagnosed on computed tomography (CT) in a patient presenting with diffuse bone pain, in whom the absence of a corroborating radiograph prompted histological confirmation.

CASE PRESENTATION

A 53-year-old woman with no significant past medical history presented with diffuse, longstanding bone pain. She reported no history of trauma, fever, weight loss, or other constitutional symptoms. Physical examination was unremarkable, and in particular no cutaneous lesions suggestive of connective tissue naevi or dermatofibrosis were identified. There was no family history of a similar bone disorder.

Laboratory investigations, including full blood count and serum calcium-phosphate metabolism, were within normal limits.

Computed tomography, performed as part of the diagnostic work-up for her bone pain, revealed innumerable well-defined, round to oval sclerotic lesions of variable size, diffusely distributed throughout the axial and appendicular skeleton (Figure 1). No conventional radiograph had been performed prior to the CT scan, and none was available for comparison. Given the diffuse skeletal involvement and in the absence of a baseline radiograph to support a confident diagnosis of osteopoikilosis on typical morphological grounds alone, a bone biopsy of one of the sclerotic lesions was performed under CT guidance (Figure 1E, F) to formally exclude a malignant sclerotic process, particularly osteoblastic metastatic disease. Histopathological examination revealed no evidence of malignancy, confirming the benign nature of the lesion and supporting a diagnosis of osteopoikilosis.

Following multidisciplinary discussion, no further oncological work-up was pursued, and the patient was managed conservatively with clinical surveillance. No specific treatment was indicated, consistent with the benign, non-progressive nature of the condition.

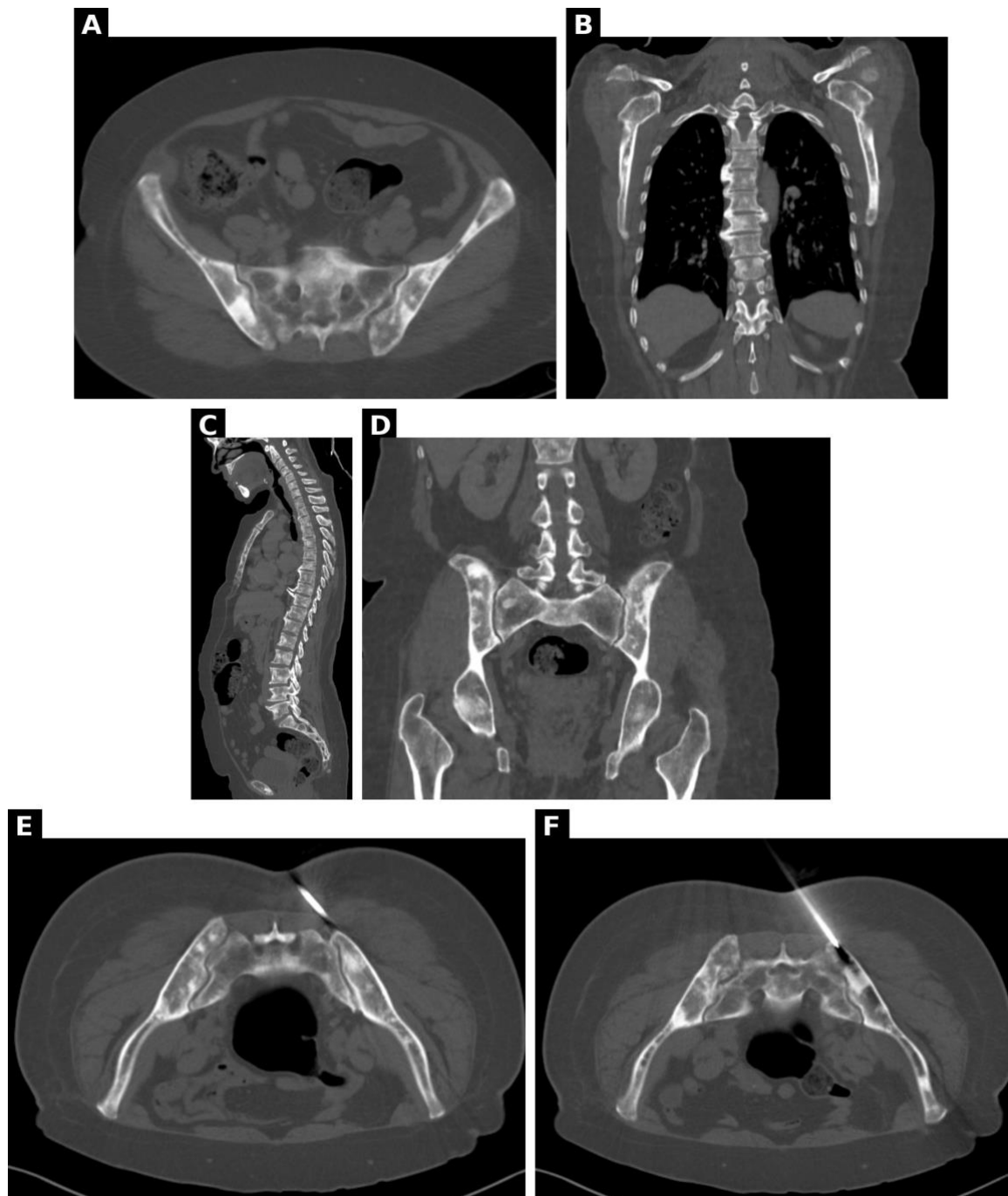


Figure 1: Contrast-enhanced CT demonstrating innumerable well-defined sclerotic foci of variable size diffusely distributed throughout the axial and appendicular skeleton. (A) Axial CT of the pelvis showing bilateral iliac sclerotic lesions. (B) Coronal CT of the thorax showing scattered sclerotic foci within the ribs and thoracic vertebrae. (C) Sagittal CT of the trunk demonstrating the distribution of sclerotic lesions relative to the vertebral column and posterior ribs. (D) Coronal CT of the pelvis confirming the diffuse, bilateral, symmetrical distribution of sclerotic lesions. (E) Axial CT of the pelvis at the level selected for biopsy, prior to needle placement. (F) Axial CT of the pelvis during CT-guided biopsy, showing the needle traversing the gluteal soft tissues to sample one of the iliac sclerotic lesions.

DISCUSSION

Osteopoikilosis is an uncommon sclerosing bone dysplasia, with a reported prevalence of approximately 1 in 50,000, though the true prevalence is likely underestimated given its frequently silent, incidental presentation [5,6]. It results from heterozygous loss-of-function mutations in *LEMD3*, which lead to unchecked TGF- β /BMP signalling and localised excessive deposition of lamellar bone within the medullary cavity [2]. The disorder is usually sporadic in clinical presentation despite its autosomal dominant inheritance, as many affected relatives remain undiagnosed due to the absence of symptoms [3].

Classically, osteopoikilosis is asymptomatic and identified incidentally on imaging performed for unrelated indications. However, a minority of patients report mild arthralgia or bone pain, as in the present case, although a definitive causal relationship between the bone islands themselves and pain has not been firmly established, and coexisting musculoskeletal conditions should also be considered [4,7].

The reference imaging modality for diagnosis is the conventional radiograph, which typically demonstrates multiple small (2–10 mm), round, oval, or linear sclerotic foci concentrated around the epiphyses and metaphyses of long bones, in a symmetrical distribution. Lesions characteristically involve the pelvis, the ends of long bones, and the small bones of the hands and feet, while the skull, ribs, and vertebral column are typically spared [5,8]. CT demonstrates the same lesions as discrete areas of cortical-density bone within the medullary cavity, without periosteal reaction, cortical destruction, or associated soft-tissue mass, features that are key to distinguishing osteopoikilosis from malignant sclerotic processes [5]. In our patient, the diagnosis was made on CT alone, without a corroborating radiograph; although CT is capable of demonstrating the same characteristic morphology as radiography, most previously reported cases have relied on the radiographic pattern to reach a confident non-invasive diagnosis, which may explain why histological confirmation was considered necessary in this case.

The principal differential diagnosis is osteoblastic metastatic disease, particularly from prostate or breast primary tumours, which can mimic osteopoikilosis both clinically and radiologically. Features favouring osteopoikilosis over metastatic disease include the small, uniform size of the lesions, their characteristic periarticular epiphyseal-metaphyseal distribution, symmetry, absence of cortical destruction, and a stable appearance on follow-up imaging when available [5,8]. Other differential diagnoses include mastocytosis, tuberous sclerosis complex, osteopathia striata, and, when only a single or a few larger lesions are present in a long bone,

melorheostosis, which shares the same underlying genetic pathway [2,8]. When the diagnosis remains uncertain on radiography or CT, particularly in patients with a known malignancy, bone scintigraphy is a useful adjunct: osteopoikilosis lesions characteristically show no or minimal tracer uptake, in contrast to the intense uptake typical of osteoblastic metastases [6].

Because of its benign, non-progressive nature and pathognomonic imaging appearance in typical cases, osteopoikilosis is often regarded as a skeletal "do-not-touch" lesion, and recognition of this pattern is essential to avoid unnecessary invasive investigations, patient anxiety, and healthcare costs [4,8]. Nonetheless, as illustrated by our case, biopsy or additional functional imaging may still be warranted when the diagnosis cannot be confidently established on morphological grounds alone, such as when only cross-sectional imaging without a corroborating radiograph is available, or when the clinical context raises concern for malignancy. No specific treatment is required for osteopoikilosis itself, and management consists of reassurance and, where relevant, evaluation for associated Buschke-Ollendorff syndrome if cutaneous lesions are identified [3,9].

CONCLUSION

Osteopoikilosis is a rare, benign sclerosing bone dysplasia that should be recognised by radiologists and clinicians alike, given its potential to mimic osteoblastic bone metastases. While the diagnosis can often be made confidently on the basis of the characteristic radiographic pattern, our case illustrates that when imaging is limited to CT without a corroborating radiograph, tissue confirmation may be a reasonable step to exclude malignancy, particularly in a patient presenting with bone pain. Once confirmed, osteopoikilosis requires no specific treatment beyond reassurance and routine clinical surveillance.

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