Osteopoikilosis ("Spotted Bone Disease"): A Benign Bone Finding. A Case Report

Víctor A. Avendaño Arango,[*] Ricardo Londoño García,[*] Gustavo A. Molina,[**] Laura Moreno,[#] Miguel A. Murcia Hernández[**]
[*]*Universidad Pontificia Bolivariana, Medellín, Colombia*
[**]*Oncologic Orthopedics Department, Hospital Pablo Tobón Uribe, Medellín, Colombia*
[#]*Universidad Pontificia Javeriana, Cali, Colombia*

ABSTRACT
We report the case of a woman with osteopoikilosis who was evaluated by the orthopedic oncology service due to blastic lesions in the left proximal femur identified on pelvic radiography and increased uptake on bone scintigraphy. The patient reported a one-year history of polyarthralgia of unknown origin, with pain in the spine, hips, and knees, which worsened during the menstrual cycle and when ascending or descending stairs. Magnetic resonance imaging of the pelvis and left femur was performed, confirming the findings. **Conclusion:** Osteopoikilosis is a rare benign sclerosing bone dysplasia; however, lack of awareness of this condition may lead to unnecessary invasive studies and emotional distress. This case is presented to raise awareness of its existence and its importance as a differential diagnosis of malignant diseases.
Keywords: Osteopoikilosis; Albers-Schoenberg disease; benign bone tumor.
Level of Evidence: IV

Osteopoiquilosis, "la enfermedad de los huesos manchados": un hallazgo óseo benigno. A propósito de un caso clínico

RESUMEN
Se comunica el caso de una mujer con osteopoiquilosis, evaluada en el Servicio de Ortopedia Oncológica, por presentar lesiones blásticas en el fémur proximal izquierdo detectadas en una radiografía de pelvis e hipercaptación en la gammagrafía ósea. La paciente refirió que, desde hacía un año, tenía poliartralgias de origen desconocido, dolor en la columna, las caderas y las rodillas, que se exacerbaba con el ciclo menstrual, y al bajar y subir escaleras. Se solicitó una resonancia magnética de pelvis y de fémur izquierdo, con la que se confirmaron los hallazgos. **Conclusiones:** La osteopoiquilosis es una displasia ósea esclerosante benigna con una baja incidencia; sin embargo, desconocer esta enfermedad lleva a indicar estudios invasivos y a generar un malestar emocional. Se presenta este caso clínico con la intención de concientizar sobre su existencia y la importancia como diagnóstico diferencial de enfermedades malignas.
Palabras clave: Osteopoiquilosis; enfermedad de Albers-Schoenberg; tumor óseo benigno.
Nivel de Evidencia: IV

INTRODUCTION

Osteopoikilosis, also known as disseminated condensing osteopathy, is an autosomal dominant disorder associated with heterogeneous mutations in the LEMD3 gene, which encodes a protein of the inner nuclear membrane. It was first described by Heinrich Albers-Schönberg in 1915 and is currently recognized as a rare condition, with an estimated prevalence of 1 in 50,000 individuals.[1] In some studies, mutations have been identified in familial cases of osteopoikilosis.[2] Its incidence is similar in both sexes, it can occur at any age, and it primarily affects the epiphyses of long bones. It has three patterns of presentation: spotted, striated, and mixed.

Most patients are asymptomatic; only 20% experience symmetric joint pain and edema.[2,3] Additionally, 25%

Received on October 18th, 2024. Accepted after evaluation on May 2nd, 2025 • Dr. VÍCTOR A. AVENDAÑO ARANGO • avendano7245@gmail.com https://orcid.org/0000-0002-2976-3269
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may present with other conditions, such as cardiac disease, dacryocystitis, and renal and endocrine malformations. It may occur in isolation or in association with melorheostosis (hyperostosis of the cortical bone of tubular bones resembling dripping candle wax over the surface of long bones, usually unilateral and asymmetric), dermatofibrosis lenticularis disseminata, forming Buschke-Ollendorff syndrome,[4] or with Gardner syndrome, which includes osteopoikilosis and colonic polyposis.[5]

From a radiological standpoint, it is characterized by multiple small (2–3 mm), well-defined, round or ovoid radiodense sclerotic lesions. Histologically, focal areas of compact lamellar bone are observed within cancellous bone.[3,5]

For this reason, the diagnosis is usually incidental. It should be emphasized that the condition does not undergo malignant transformation or affect bone strength, and it does not require specific treatment.[1,6] To differentiate it from metastatic disease, its distribution must be considered, as it involves long bones as well as phalanges, carpal bones, metacarpals, tarsal bones, and the pelvis, with a symmetrical pattern but uneven distribution, and only rarely involves the skull, ribs, clavicles, and vertebrae. In addition, there is no evidence of bone destruction.[5,6] Its course is typically benign, and most patients remain asymptomatic, with diagnosis often being incidental. However, associations between osteopoikilosis and other malformations, such as duplicated ureter, coarctation of the aorta, precocious puberty, and exostoses, have been reported.[7]

The aim of this article is to highlight the characteristics of this condition and how it differs from metastatic disease, with emphasis on avoiding unnecessary invasive procedures, which may cause significant emotional distress for patients and impose an economic burden on the healthcare system.

CASE REPORT

A 30-year-old woman presented with bilateral knee pain radiating to the left thigh, with a one-year history. She also reported occasional thoracolumbar spine pain that worsened during the menstrual cycle and when climbing or descending stairs. Additionally, she described a nonspecific sensation of decreased strength.

Bilateral radiographs of the pelvis and femurs were obtained, revealing rounded, bead-like sclerotic lesions in both femurs, without signs of malignancy (Figure 1).

Figure 1. Anteroposterior radiograph of the pelvis showing multiple rounded sclerotic foci measuring <1 cm in the cancellous bone of the proximal femur and pelvis, sparing the cortex, with no periosteal reaction.

Magnetic resonance imaging showed multiple millimetric punctate lesions, hypointense on both T1- and T2-weighted sequences, predominantly visible on T1-weighted images. These lesions were bilateral and relatively symmetrical, involving the lower right limb, mainly at the periarticular level in the distal knee region. They had ill-defined margins, with no associated edema, expansile effect, or contrast enhancement (Figures 2 and 3).

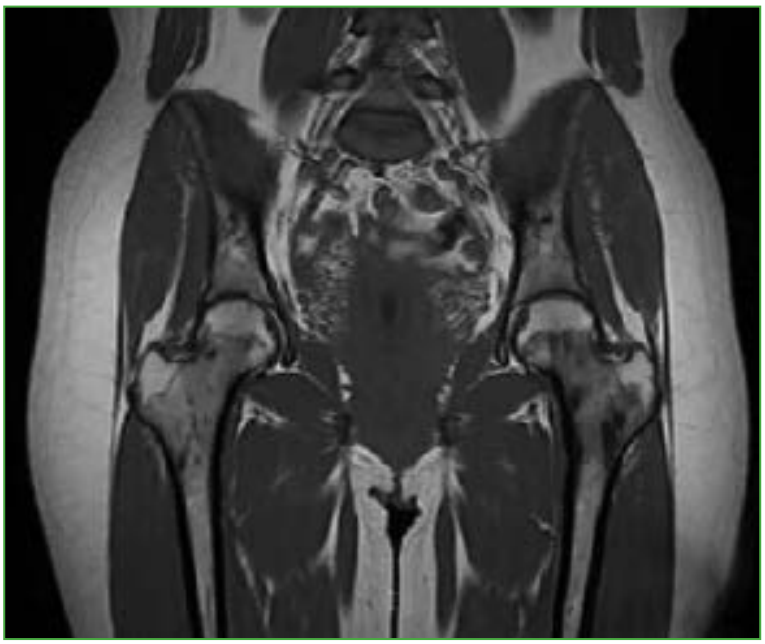

Figure 2. Magnetic resonance imaging of the pelvis, coronal section, T1-weighted sequence, showing multiple small hypointense lesions in the proximal femur.

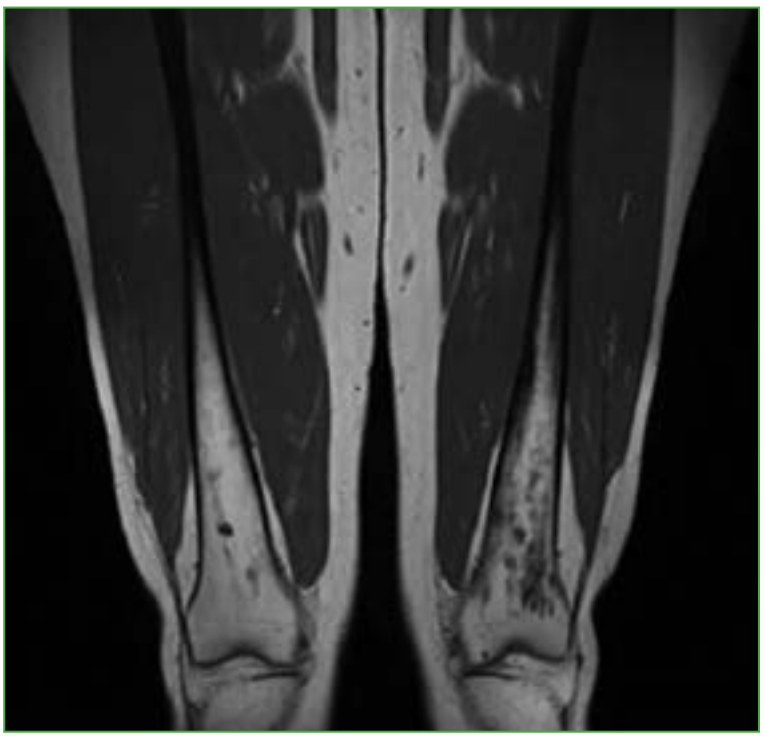

Figure 3. Magnetic resonance imaging of the distal femur, coronal section, T1-weighted sequence, showing multiple small hypointense lesions.

A bone scintigraphy was also performed, demonstrating areas of increased uptake consistent with osteopoikilosis (Figure 4).

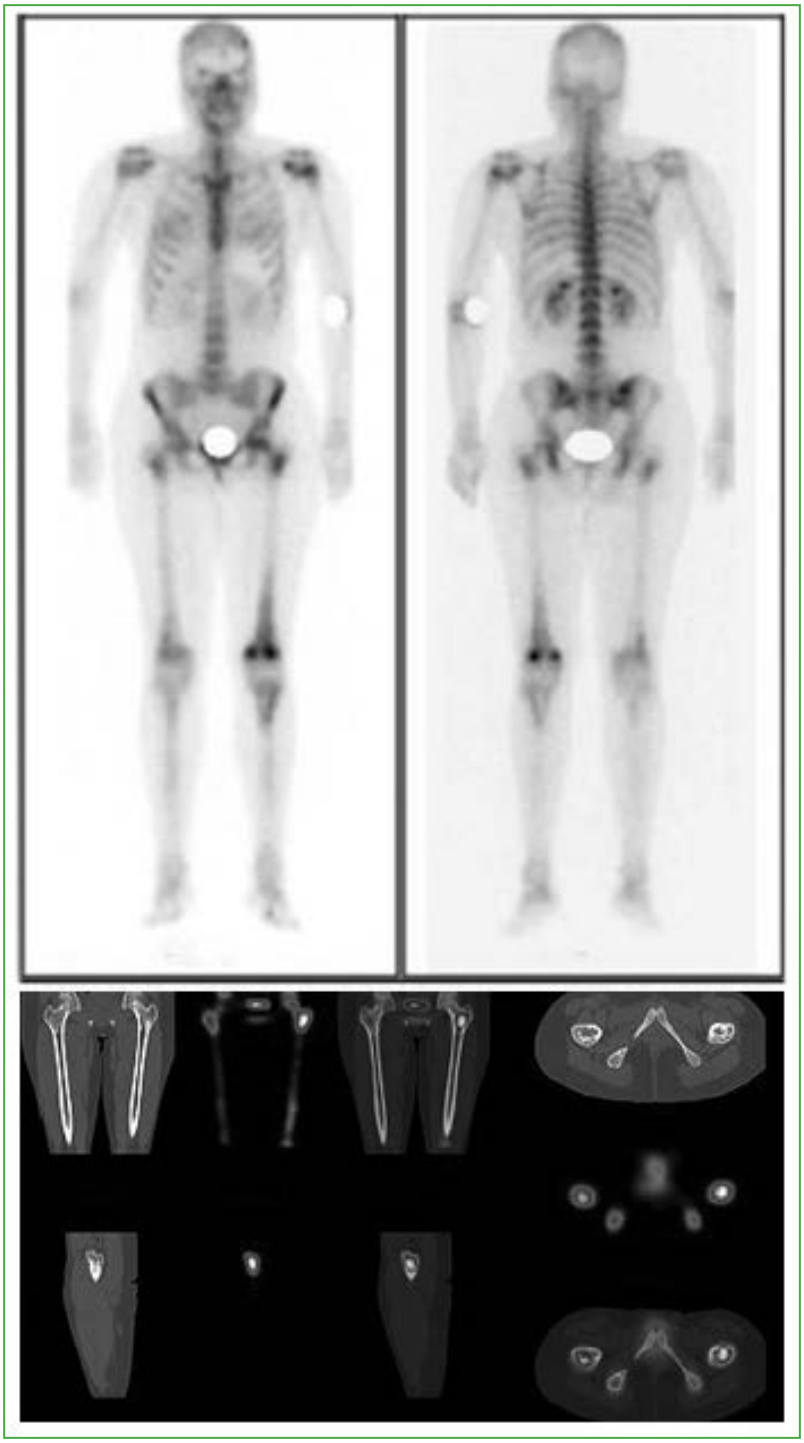

Figure 4. Bone scintigraphy showing areas of increased uptake in the bilateral proximal humeri, pelvis, and proximal and distal femora.

Based on these findings, the patient was evaluated by the Orthopedic Oncology Service, where left patellofemoral pain and hip pain on rotation were identified, along with right-sided hyporeflexia.

A discrepancy between the clinical presentation and imaging findings was noted. A consultation with the Rheumatology Department was requested, and comparative follow-up radiographs of the spine, pelvis, and femurs were obtained. These showed normal bone density, preserved joint relationships, and well-defined radiodense lesions in the distal femur and bilateral proximal tibia, more pronounced on the left side. There was no bone expansion, cortical disruption, or soft tissue component, and no changes compared with the previous study.

DISCUSSION

On magnetic resonance imaging, osteopoikilosis may appear as multiple benign bone islands distributed throughout the axial and appendicular skeleton, appearing as small hypointense lesions on both T1- and T2-weighted images.[6] These findings are attributed to a failure of resorption of secondary cancellous bone and are typically clustered around large joints.[7,8] In our patient, the main characteristic features (symmetrical involvement of long bones) were present, and clinical suspicion was confirmed by the imaging studies. However, one of the most important differential diagnoses is osteoblastic metastases, as well as Erdheim-Chester disease.[7,8] Therefore, bone scintigraphy was required to rule out findings such as osteolysis or periosteal reaction suggestive of malignancy and to exclude these diagnostic possibilities.[9] It should also be emphasized that the radiological findings of osteopoikilosis are sufficiently characteristic to avoid misdiagnosis and prevent unnecessary invasive procedures, such as biopsy.[10]

Patients with osteopoikilosis may also be associated with autoimmune disorders, as the LEMD3 gene influences the expression of transforming growth factor β1, a modulator of immune responses. Therefore, a thorough clinical examination and detailed medical history are warranted, along with referral to the Rheumatology Department to rule out such conditions. In our patient, the clinical findings did not correlate with the imaging findings, which prompted interdisciplinary evaluation.

It should be noted that this condition may predispose to excessive fibrous tissue formation, which may increase the risk of joint stiffness and disability following surgical procedures. Therefore, early diagnosis and appropriate follow-up are essential.[6]

Currently, there is no consensus regarding treatment. Some studies suggest the use of nonsteroidal anti-inflammatory drugs for pain management. Analgesics, such as acetaminophen, and opioids may also be used.[11]

CONCLUSIONS

It is essential to recognize osteopoikilosis to avoid misdiagnosis as a malignant condition, and to ensure timely referral for multidisciplinary evaluation to rule out associated autoimmune disorders.

Conflicts of interest: The authors declare no conflicts of interest.

R. Londoño García ORCID ID: https://orcid.org/0000-0002-6568-9166
G. A. Molina ORCID ID: https://orcid.org/0009-0007-3351-2298
L. Moreno ORCID ID: https://orcid.org/0009-0006-6429-7722
M. A. Murcia Hernández ORCID ID: https://orcid.org/0009-0006-8611-8625

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