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Case Report

Osteopoikilosis in a patient with familial adenopolyposis : A case report ☆

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ABSTRACT

Osteopoikilosis, a sclerosing bone dysplasia, is an asymptomatic incidental finding characterised by multiple bone islands. Although it requires no treatment there can be diagnostic uncertainty as appearances can be similar to osteoblastic metastases or metabolic disorders such as Paget disease. We present a case of osteopoikilosis in a patient with familial adenopolyposis and discuss the clinical presentation, image findings and key considerations in diagnosis of this benign entity.

Introduction

Osteopoikilosis was first described in 1915 and has a prevalence of approximately 1 in 50,000 individuals [1]. The condition has autosomal dominant inheritance with variable penetrance [2]. It is a benign, usually asymptomatic disease which is often detected as an incidental finding. There is no age or sex predilection and the condition may rarely in 15%-20% of patients present with slight juxta articular pain. However there are no specific clinical features [3].

By definition the disease is characterized by multiple bone islands (enostoses). It is characterized by symmetrically distributed sclerotic lesions centered in epiphysis and metaphysis of long bones, in a periarticular location. In rarer cases it may be diffuse throughout the axial and appendicular skeleton [3].

Bone islands often have characteristic features on imaging. They can occur almost anywhere in the skeleton, usually <1 cm in size and have characteristic peripheral radiating spicules. Mean CT attenuation values are usually >885 Hounsfield units (HU) [4]. There is absence of uptake on a bone scan. Bone islands are usually stable in size over time. Important negatives include absence of pain, no cortical destruction and no periosteal reaction.

There is no documented association between familial adenopolyposis and osteopoikilosis. Gardner syndrome is a genetic disorder characterized by familial adenopolyposis in association with other abnormalities. It results from mutation of the tumor suppressor adenomatous polyposis coli (APC)

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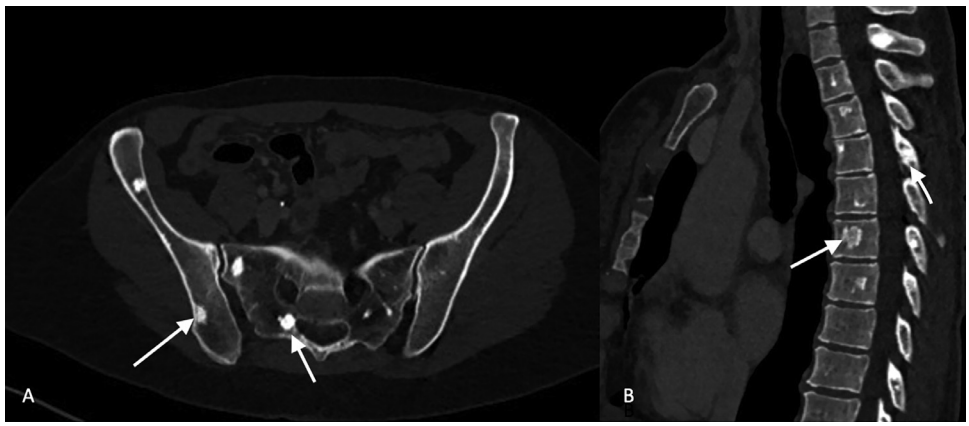

Fig. 1 – Axial (A) and sagittal (B) reformatted CT images of the axial spine in bone windows. There are multiple well defined sclerotic lesions within the pelvis and thoracic vertebrae (white arrows). There is no cortical destruction or periosteal reaction. The lesions had peripheral radiating spicules and had mean attenuation values of >885 Hounsfield units.

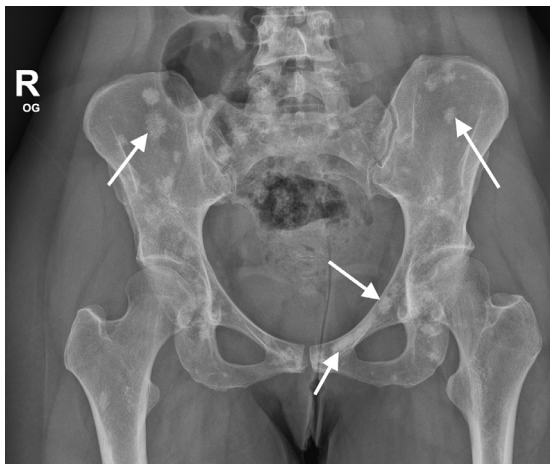

Fig. 2 – Plain film of the pelvis. There are multiple intramedullary radiodense lesions that involve the pelvis and both proximal femurs (white arrows). There is no fracture or bony destruction. The lesions have a spiculated appearance.

gene [5]. We present a rare case of osteopoikilosis in a patient with familial adenopolyposis and discuss the clinical presentation, imaging appearances and key considerations in diagnosis of this benign entity.

Case report

A middle-aged female patient presented with a 6-month history of vomiting, nausea, abdominal cramps, and weight loss in addition to a 4-week history of bilateral hip and groin pain. Past medical history included familial adenopolypisis whereby a prophylactic laparoscopic colectomy and ileorectal anastomosis was performed 1 year prior to presentation. She also had hypertension.

On examination the patient walked with normal stable gait with same push-off on both sides. On inspection there was no obvious leg length discrepancy or deformity. There was no tenderness on palpation over the hip joints bilaterally and no tenderness over the knee joints. There was mild tenderness on palpation over the anterior superior iliac spine, mild tenderness on palpation over the gluteus medius and gluteus maximus. There was significant tenderness on palpation over the sacroiliac joints. Mild tenderness on palpation over L5 and S1 vertebrae. The patient had good range of motion of the hip and knee joints bilaterally. The neurological examination did not reveal any deficit.

A CT chest, abdomen, and pelvis with contrast was performed to rule out malignancy. There was no evidence of visceral malignancy on the CT. There were inflammatory/infectious nodular changes in the left lung which resolved on subsequent imaging as well as an incidental endometrial polyp.

The CT scan demonstrated multiple well defined sclerotic lesions throughout the axial skeleton (Fig. 1). The mean attenuation values of the lesions were well above 1000 HU and there were peripheral radiating spicules. There was no cortical destruction, soft tissue component or periosteal reaction. The initial impression was these lesions appeared benign, however given the history of familial adenopolyposis and clinical presentation multidisciplinary team discussion was recommended.

The patient also had a plain film of the pelvis following this which demonstrated similar appearances (Fig. 2).

The patient was discussed at the orthopedic meeting where a SPECT CT from the vertex to the mid-thighs (Fig. 3) was recommended to confirm the benign nature of the lesions. There was no abnormal focal area of increased tracer uptake throughout the skeleton.

The combination of the CT and SPECT CT findings confirmed the diagnosis of osteopoikilosis. The patient required no follow up or further investigations.

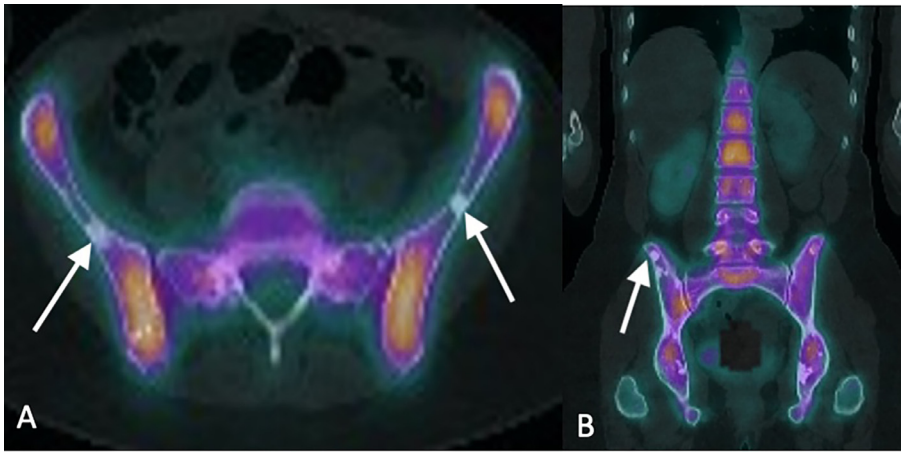

Fig. 3 – Axial (A) and coronal (B) images of SPECT CT after the injection of 99mTc HDP. There is no abnormal focal tracer uptake in the sclerotic lesions seen on CT (white arrows).

Discussion

This case is an excellent depiction of the typical findings seen in osteopoikilosis which have been corroborated in current literature. Firstly the lesions are sclerotic with a radiodense central appearance and peripheral spiculated margins. Secondly these lesions demonstrated no abnormal tracer uptake on the bone scan, in keeping with current literature. However low-grade activity can be seen in histologically proven bone islands, particularly if the lesions are larger than 1 cm [6]. Atypical features include the presence of pain or significant change in size, in these cases a bone biopsy may be warranted.

There are divided opinions with regards to the use of CT attenuation cut off values in differentiating bone islands from untreated osteoblastic metastases. There are 2 studies which have reported a high diagnostic accuracy in differentiating these pathologies using mean and maximum CT attenuation values [4,7]. Ulano et al. [4] compared the attenuation values of 126 bone islands in 30 patients with 153 osteoblastic metastases in 25 patients. They found that using a cut off of >885 HU for mean attenuation the sensitivity was 95% and specificity was 96%. However Azar et al. [8] found CT attenuation values do not reliably differentiate benign sclerotic lesions from osteoblastic metastases in patients who underwent bone biopsy.

Familial adenopolyposis is characterized by the presence of hundreds or thousands of colonic adenomatous polyps. These intestinal polyps carry a 100% risk of malignant change, so early diagnosis is crucial [9]. There is an association between familial adenopolyposis and osteomas in the head and neck, this is termed Gardner syndrome. Gardner syndrome is associated with other abnormalities including epidermal cysts, dental anomalies, desmoid tumors and malignancies of the small bowel and thyroid.

To our knowledge there is no association between familial adenopolyposis and osteopoikilosis, as seen in our case. There have been case reports which describe bone and dental abnormalities as being first signs of familial Gardner syndrome [9,10], however these bone abnormalities are separate entities to osteopoikilosis. Osteopoikilosis itself is an inherited condition with evidence suggesting it is caused by a mutation in the LEMD3 gene [11]. It is associated with other musculoskeletal conditions including melorheostosis [11]. Therefore there is no genetic overlap or other cases at present to hypothesise familial adenopolyposis and osteopoikilosis are associated conditions.

Conclusion

Osteopoikilosis is a rare benign entity. CT and bone scan demonstrate the characteristic appearances of peripheral spicules, CT mean attenuation >885 HU and no abnormal tracer uptake. Knowledge of these typical features will aid the clinician in reliably distinguishing this sclerosing bone dysplasia from osteoblastic metastases and metabolic bone disorders.

Although familial adenopolyposis can be associated with osteomas in the head and neck (Gardner syndrome) there is no evidence in the current literature to suggest this can be associated with osteopoikilosis. Therefore our case likely represents incidental unrelated dual pathological processes.

Patient consent

Verbal and written informed consent for the publication of this case report was obtained from the patient.

REFERENCES

[1] Hill CE, McKee L. Osteopoikilosis: an important incidental finding. Int. J. Care Injured 2015;46:1403–5. doi:10.1016/j.injury.2015.02.005.

[2] Benli IT, Akalin S, Boysan E. Epidemiological, clinical and radiological aspects of osteopoikilosis. J Bone Joint Surg Br 1992;74(4):504–6. doi:10.1302/0301-620X.74B4.1624505.
[3] Panchal SR, Gawhale S, Shah, et al. Case report of osteopoikilosis: sparse cause of bone pain and mimicker of metastasis on radiographs. J Orthopaed Case Rep 2021;11(3):98–101 [online]. doi:10.13107/jocr.2021.v11.i03.2106.
[4] Ulano A, Bredella M, Burke P, et al. Distinguishing untreated osteoblastic metastases from enostoses using CT attenuation measurements. AJR Am J Roentgenol 2016;207(2):362–8. doi:10.2214/AJR.15.15559.
[5] Brueckl WM, Ballhausen WG, Förtsch, et al. Genetic testing for germline mutations of the APC gene in patients with apparently sporadic desmoid tumors but a family history of colorectal carcinoma. Dis Colon Rectum 2005;48(6):1275–81. doi:10.1007/s10350-004-0949-5.
[6] Greenspan A, Stadalnik R. Bone Island: Scintigraphic findings and their clinical application. Can Assoc Radiol J 1995;46(5):368–79.
[7] Sala F, Dapoto A, Morzenti C, Firetto MC, Valle C, Tomasoni A, Sironi S. Bone islands incidentally detected on computed tomography: frequency of enostosis and differentiation from untreated osteoblastic metastases based on CT attenuation value. Br J Radiol 2019;92(1103):20190249. doi:10.1259/bjr.20190249.
[8] Azar A, Garner H, Rhodes N, Yarlagadda B, Wessell D. CT attenuation values do not reliably distinguish benign sclerotic lesions from osteoblastic metastases in patients undergoing bone biopsy. AJR Am J Roentgenol 2021;216(4):1022–30. doi:10.2214/AJR.20.24029.
[9] Yu D, Ng Cw B, Zhu H, Liu J, Lin Y. Bone and dental abnormalities as first signs of familial Gardner's syndrome in a Chinese family: a literature review and a case report. Med Sci (Paris) 2018;34 Focus issue F1:20–5. doi:10.1051/medsci/201834f104.
[10] Antal G, Zsigmond A, Till Á, et al. Case report: Initial atypical skeletal symptoms and dental anomalies as first signs of Gardner syndrome: the importance of genetic analysis in the early diagnosis. Pathol Oncol Res 2024;30:1611768. doi:10.3389/pore.2024.1611768.
[11] Bansal A. The Dripping Candle Wax Sign. Radiology 2008;246(2):638–40. doi:10.1148/radiol.2462050537.