

## UNUSUAL PRESENTATION OF OSTEOPOIKILOSIS

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**We present a case of osteopoikilosis in a 74 year-old woman with hip pain, presenting multiple osteoblastic lesions of the axial skeleton including an osteoblastic large lesion of her left femur. The imaging findings on X-rays and computed tomography are provided along with the discussion of the differential diagnosis on the basis of the recent literature.**

**Key-words:** Bones, diseases.

### Case report

A 74 year-old woman was referred for a 2-month history of pain and limitation of movement of her left hip. There was no history of trauma.

X-rays revealed a single osteoblastic lesion localised at her proximal left femur (Fig. 1).

Computed tomography (CT) showed a 2 cm large osteoblastic lesion of left proximal femur with multiple radial peripheral spiculae and other smaller osteoblastic lesions of the hip and the proximal femora not detectable on X-rays (Fig. 2).

Technetium-99m (<sup>99m</sup>Tc) bone scan (not shown) and the <sup>18</sup>F-fluorodeoxyglucose (FDG) positron emission tomography combined with computed tomography (FDG-PET/CT) (not shown) were unremarkable.

Due to the appearance and localization of the osteoblastic lesions, a diagnosis of osteopoikilosis was suspected.

A biopsy of the proximal left femur confirmed the diagnosis.

### Discussion

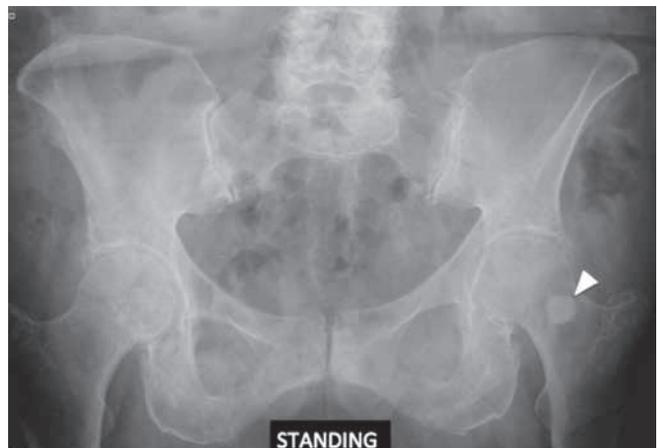
Osteopoikilosis (OPK) is a rare and benign dystrophic disease of the bone (1).

In OPK multiple osteoblastic bone lesions are found within the trabecular bone.

OPK is typically localised at the metaphyses and epiphyses of the appendicular skeleton (2, 3). The axial skeleton and skull are usually spared. OPK can mimic osteoblastic bone metastases (4, 5).

The estimated prevalence of the OPK is 1 in 50 000 individuals. OPK is incidentally found in subject of all ages with no sex predilection (4).

The OPK is due to a mutation of LEMD3 gene (6). Most cases of OPK are sporadic. Nevertheless, an autosomal dominant inheritance has been reported (6). Rarely inherited



*Fig. 1.* — X-rays of the hip and proximal femora: a femoral osteoblastic solitary bone of 2,2 cm lesion is noted in the left femoral neck (white arrowhead).

OPK can be associated with connective tissue nevi (Buschke-Ollendorff syndrome) (6).

In most clinical settings, OPK is asymptomatic and bone changes are incidentally detected on imaging. However, mild pain has been reported in up to 15-20% of cases (7).

On X-rays and Computed Tomography (CT), OPK presents as multiple appendicular enostosis measuring <1 cm (1). The osteoblastic lesions observed in OPK are round or oval-shaped with spiculated margins in continuity with the nearby trabecular bone, the elementary lesion sharing the same appearance of a classical bone island. No transitional zone is observed around the osteoblastic spots (4).

Bone lesions of OPK have usually a symmetrical distribution at the epiphyses and metaphyses of the appendicular skeleton. This pattern of distribution is virtually diagnostic of OPK (3).

The main differentials diagnosis of OPK include osteoblastic metastases, bone mastocytosis, Bourneville's disease and bone sarcoidosis (1).

Osteoblastic metastases are usually located in the axial skeleton. They may be heterogeneous and a transitional surrounding zone of abnormal can be observed.

In systemic mastocytosis multiple osteoblastic lesions are found in late phase of the bone involvement and they are located at the metaphyses and the diaphyses of the appendicular skeleton. An irregular thickening of the cortical is usually associated (8).

Bone lesions in Bourneville's disease, bone lesions are associated with a periosteal reaction and different degrees of hyperostosis. Bone lesions of Bourneville's disease can occur anywhere in bones including the axial skeleton and the extremities. Calvarium is typically involved (4).

A bone scan can be performed in case of doubt, as in our case, when lesions larger than 1 cm are found (1).

Osteoblastic lesions of OPK are generally small and Technetium-99m (<sup>99m</sup>Tc) bone scan doesn't show any tracer uptake. However, a positive bone scan does not rule out OPK (10).

The <sup>18</sup>F-Fluoro-deoxyglucose Positron Emission Tomography (<sup>18</sup>FDG-PET) was unremarkable in our patient but not enough data are

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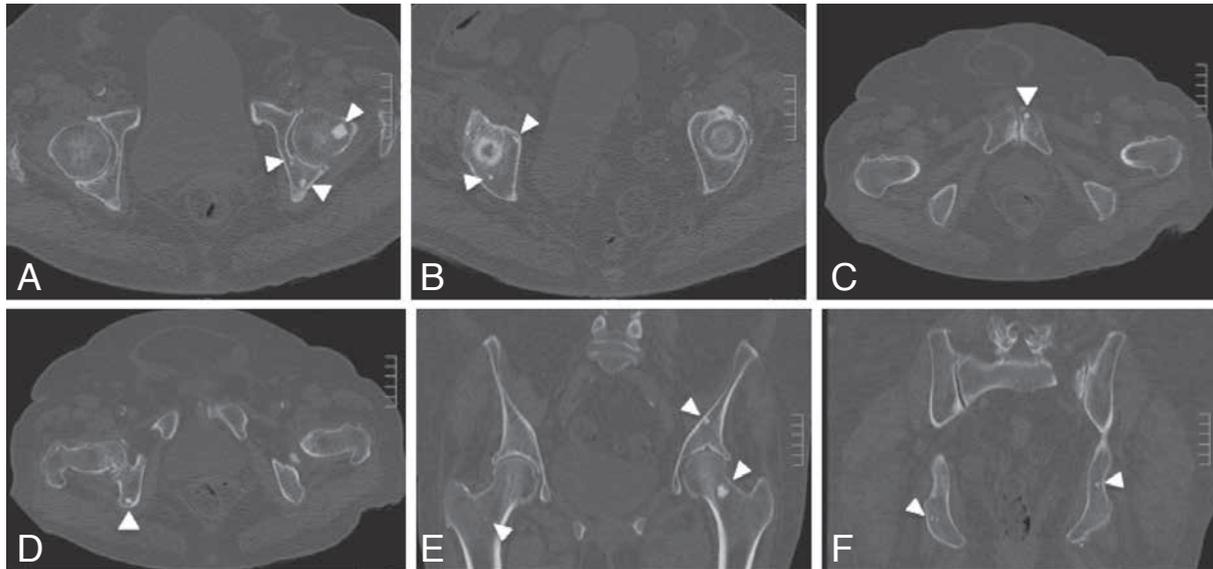


Fig. 2. — Computed tomography (CT) (A-D axial slices, thickness 1 mm; E-F coronal reformatted images, slice thickness 1,5 mm): CT revealed other osteoblastic spots of the hip bone and the proximal femora of less than 1 cm (white arrowheads). Like the large lesion of the left femur, these smaller osteoblastic lesions appear well margined and speculated. No transitional zone is observed.

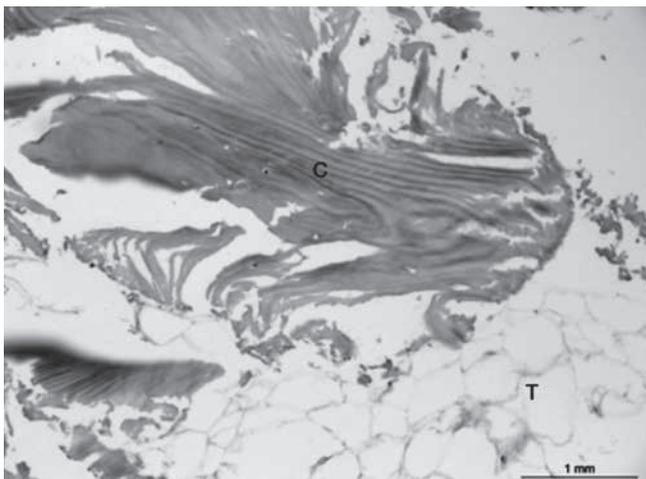


Fig. 3. — Histology specimen of the lesion of the left femoral neck (H&E stain; magnification x20). The cortical bone (C) is within the trabecular bone (T). Absence of periosteum covering the cortical bone.

available in literature about OPK appearance on  $^{18}\text{F}$ FDG-PET.

As in our patient, histology can corroborate the diagnosis of OPK in case of the doubt (Fig. 3). Histology shows the islands of mature cortical bone with thickened sclerotic irregular laminae interspersed in the cancellous bone (2). The absence of periosteum around the cortical bone confirms its intramedullary origin (Fig. 3).

In conclusion, OPK is an uncommon benign dystrophic disease of the bone. The imaging findings are quite typical in vast majority of cases. Bone scan and biopsy can be performed in atypical presentations to

rule out bone metastases. Bone metastases and rare conditions such as bone mastocytosis and Bourneville's disease should be taken into account in the differential diagnosis.

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