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Поражение костей таза, позвоночника и рёбер при остеопойкилии: клинический случай

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АННОТАЦИЯ

Остеопойкилия — редкая форма наследственной доброкачественной дисплазии костей, случайно обнаруживаемая при рентгенографии. Характеризуется специфической рентгенологической картиной — диффузными склеротическими участками кости круглой или овальной симметричной формы, определяемыми по всему скелету. Правильная постановка диагноза очень важна, поскольку поражения такого типа схожи с костными метастазами.

В данной статье представлен случай остеопойкилии у пациентки, обратившейся в нашу клинику с жалобой на кратковременную потерю сознания без признаков онемения, покалывания, слабости в ногах или других частях тела. Компьютерная томография показала множественные мелкие склеротические очаги, рассеянные по грудному и поясничному отделу позвоночника, рёбрам, тазовым костям, крестцу и проксимальному отделу бедренных костей с обеих сторон. При остеосцинтиграфии всего тела с применением технеция-99м повышения накопления препарата не выявлено. У пациентки были диагностированы характерные рентгенологические признаки остеопойкилии, после чего она находилась под наблюдением.

Ключевые слова: остепойкилия; дисплазия костей; клинический случай.

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ЭКО•ВЕКТОР

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Osteopoikilosis in the ribs, pelvic region and spine: a case report

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ABSTRACT

Osteopoikilosis is a rare inherited benign bone dysplasia incidentally found on radiological exams. It is characterized by a specific radiological pattern: diffuse, round or oval, symmetrically shaped sclerotic bone areas distributed throughout the skeleton. It is essential to do a correct diagnosis because these lesions could be easily confused with bone metastasis.

We reported a case of an osteopoikilosis patient presenting to our clinic with transient loss of consciousness and without any numbness, tingling and weakness in the legs or other parts of the body. The computed tomography scan showed multiple small sclerotic foci bone islands, scattered throughout the thoracic and lumbar spine, ribs, pelvic bone, sacrum and bilateral proximal femur. No significant increase in the activity was detected in technetium-99m whole-body bone scintigraphy. The patient was diagnosed with characteristic radiological findings of osteopoikilosis and was followed up.

Keywords: osteopoikilosis; bone dysplasia; clinical case.

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肋骨、骨盆区和脊柱脆性骨硬化：一份病例报告

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摘要

脆性骨硬化是一种在放射学检查中偶然发现的罕见遗传性良性骨发育不良。其特征是具有特殊的放射学表现：分布于整个骨骼的弥漫性、圆形或椭圆形、形状对称的骨硬化区。这些病变很容易与骨转移瘤相混淆，因此做出正确诊断至关重要。

本文报告了一例脆性骨硬化患者，其因一过性意识丧失前来我们门诊就诊，双腿或身体其他部位无任何麻木、麻刺感和虚弱。计算机断层成像扫描示多发小面积硬化性骨岛，散布于胸腰椎、肋骨、骨盆、骶骨和双侧股骨近端。锝-99m全身骨显像未检测到活性显著增加。

患者被诊断为脆性骨硬化典型放射学表现，并接受随访。

关键词：脆性骨硬化；骨发育不良；临床病例。

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BACKGROUND

Osteopoikilosis is a rare benign bone dysplasia that affects about one in every 50,000 people, usually with no age or gender differences [1].

It is characterized by numerous circular or ovoid sclerotic bone lesions symmetrically distributed throughout the skeleton [2]. Lesions are frequently found incidentally on imaging studies for unrelated complaints [3]. Histologically, the lesions are thicker trabeculae of lamellar osseous tissue with haversian systems within the cancellous structure; they are most likely foci of bone that did not become cancellous throughout growth and differentiation. The condensation of cancellous bone in osteopoikilosis consists of a peripheral area of trabeculae in which osteocytes are scant, and there are no osteoblasts or osteoclasts (both are present in the central core of irregular trabeculae) [4,5].We report a case of osteopoikilosis patient who presented to our clinic for a syncope.

DESCRIPTION OF THE CASE

A 43-year-old female patient was taken to the emergency room by ambulance after experiencing transient loss of consciousness. The initial evaluation consisting of history, physical examination, 12-lead electrocardiogram and laboratory tests did not reveal any abnormalities; thus, a total-body computed tomography (CT) was performed. The CT scan showed multiple small sclerotic foci bone islands, scattered throughout the thoracic (Figure 1a) and lumbar spine (Fig. 1b), ribs, pelvic bone (Fig. 2), sacrum (Fig. 3) and bilateral proximal femur (Fig. 4). All bones were free of any cortical erosion or periosteal reaction. No other signs, such as rubor or edema, were noticed; moreover, the patient did not describe any numbness, tingling and weakness in the legs or other parts of the body.

The CT pattern was suspicious for osteopoikilosis. The relative clinical and laboratory tests, such as routine blood count, erythrocyte sedimentation rate, serum electrolytes, tumor markers, alkaline and acid phosphatase, ANA and anti-DS-DNA were negative for any type of arthritis, infection or osteoblastic bone metastases, which were in the differential diagnosis. No significant increase in the activity was detected in technetium-99m whole-body bone scintigraphy. The patient was diagnosed with typical radiological findings of osteopoikilosis by excluding other differential diagnoses and was followed up.

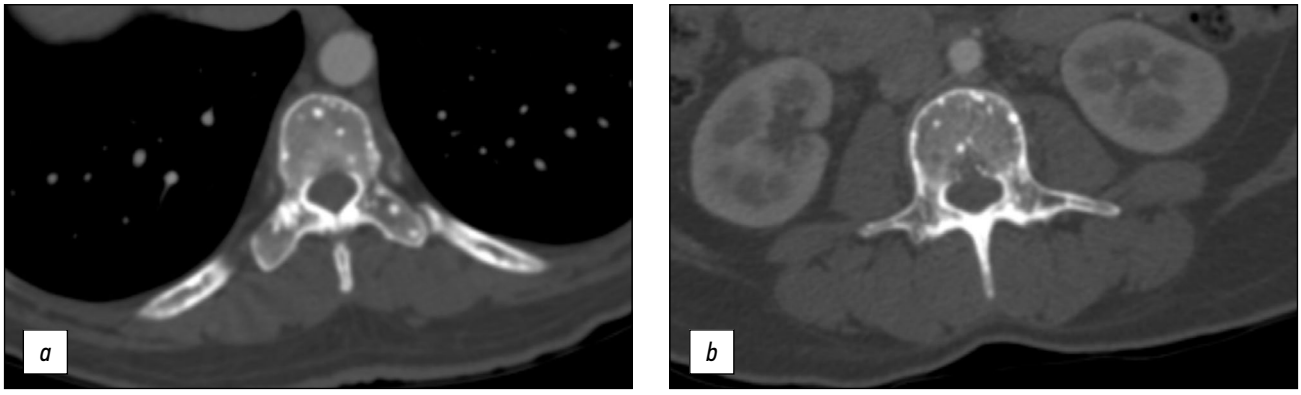

Fig. 1. Transverse cross-section computed tomography scan passing through the thoracic (*a*) and lumbar (*b*) spine. It shows numerous, well-defined, homogeneous, circular, hyperdense foci in spinous processes and vertebral arches**.**

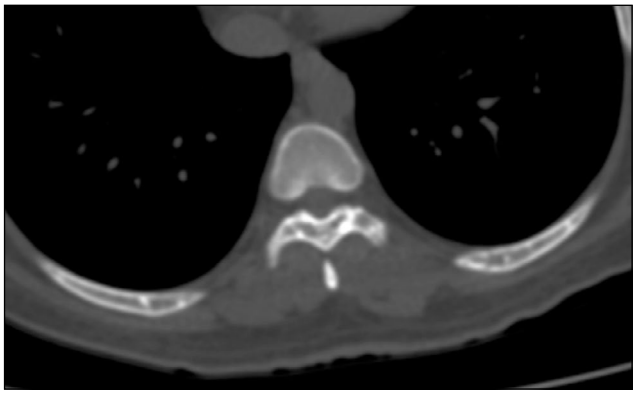

Fig. 2. Transverse cross-section computed tomography scan passing through the seventh rib. It shows numerous hyperdense lesions; these are well-circumscribed and are measured in millimeters.

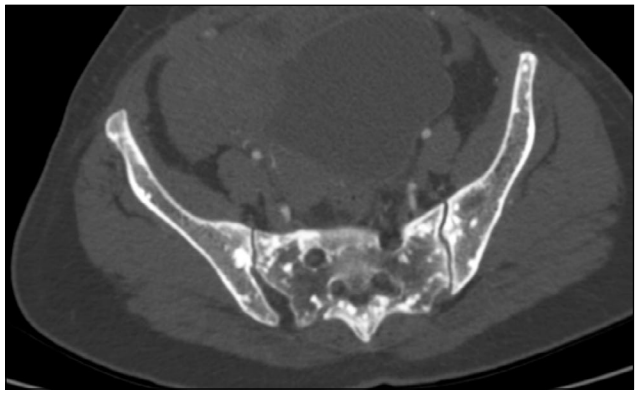

Fig. 3. Transverse cross-section computed tomography scan passing through the sacroiliac joints. It shows small, sclerotic, round opacities distributed symmetrically along sacrum, hip bone, and sacroiliac joints.

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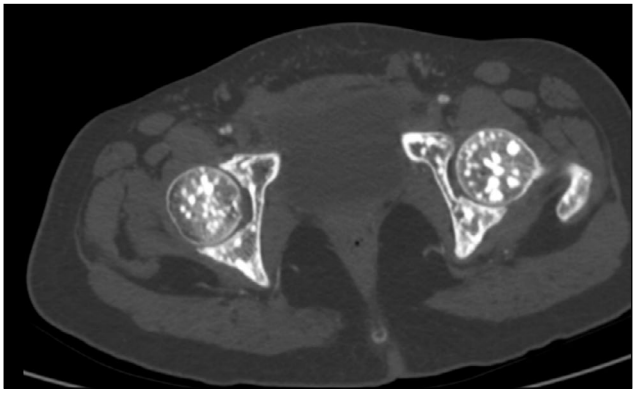

Fig. 4. Transverse cross-section computed tomography scan passing through the femoral head. It shows numerous hyperdense lesions that conform with the osteopoikilosis in the femoral head; lesions are well-circumscribed and are measured in millimeters.

DISCUSSION

Osteopoikilosis (also known as "spotted bone disease" or osteopathia condensans disseminata) is a rare bone dysplasia and was first described by Albers-Schönberg in 1915 [6]. The incidence of this disease is estimated around one in 50,000, usually without age or gender differences [1]. It is usually autosomal dominant in inheritance, but sporadic forms are also reported [1]. Current literatures suggest loss-of-function mutations of *LEM domain-containing 3* (*LEMD3*) gene located on 12q might be the cause. These mutations could also affect soft tissue and skin, causing melorheostosis that is a benign sclerosing bone dysplasia with cortical hyperostosis, thickening and fibrosis of overlying skin and Buschke–Ollendorff syndrome that comprises osteopoikilosis associated with disseminated connective tissue and cutaneous yellowish nevi [7,8]. Osteopoikilosis lesions are typically found incidentally on imaging studies done for unrelated complaints [3]. Radiological lesions of osteopoikilosis are typical: they are characterized by numerous symmetrical, homogeneous, well circumscribed, small (1–10 mm in diameter) and round or oval shaped sclerotic lesions. The most commonly affected areas are the epiphyses of short tubular bones and the metaphyses of long bones. In addition, carpal and tarsal bones, scapula, pelvis and sacrum are reported to be frequently affected [9,10]. Ribs, clavicles, spine and skull involvement is uncommon [11]. Because of their similarities, the radiological lesions of osteopoikilosis can be confused with osteoblastic bone metastases, but there are significant differences that allow us to make a differential diagnosis. In contrast to bone metastasis, the sclerotic lesions in osteopoikilosis are symmetrical, consistent in size and do not induce cortical erosion. As a result, bone scintigraphy plays an important role in definitive diagnosis; in fact, a normal radionuclide bone scan generally excludes the possibility of osteoblastic bone metastasis. Nevertheless, several cases of osteopoikilosis with an abnormal bone scan have been reported in the literature [12,13].

CONCLUSION

Although osteopoikilosis is a rare condition, it can be easily diagnosed through its typical radiological findings. Therefore, clinicians must be aware of and recognize this image pattern in order to make an accurate diagnosis and prevent further examinations and aggressive treatments.

ADDITIONAL INFORMATION

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Consent for publication. Written consent was obtained from the patient for publication of relevant medical information and all of accompanying images within the manuscript.

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