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

M.T. Paparella[1], I. Gangai[1], Ch. Porro[1], L. Eusebi[2], F. Silveri[3], A. Cammarota[4], G. Guglielmi[1, 5]

[1] Department of Clinical and Experimental Medicine, Foggia University School of Medicine, Фоджа, Италия
[2] Radiology Unit, Carlo Urbani, Джези, Италия
[3] Rheumatology Unit, University of Ancona, Анкона, Италия
[4] IRCCS-CROB, Рионеро-ин-Вультуре, Италия
[5] Radiology Unit, Barletta University Hospital, Фоджа, Италия

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

Maria Teresa Paparella[1], Ilaria Gangai[1], Chiara Porro[1], Laura Eusebi[2], Ferdinando Silveri[3], Aldo Cammarota[4], Giuseppe Guglielmi[1, 5]

[1] Department of Clinical and Experimental Medicine, Foggia University School of Medicine, Foggia, Italy
[2] Radiology Unit, Carlo Urbani, Jesi, Italy
[3] Rheumatology Unit, University of Ancona, Ancona, Italy
[4] IRCCS-CROB, Rionero in Vulture, Italy
[5] Radiology Unit, Barletta University Hospital, Foggia, Italy

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

Maria Teresa Paparella[1], Ilaria Gangai[1], Chiara Porro[1], Laura Eusebi[2], Ferdinando Silveri[3], Aldo Cammarota[4], Giuseppe Guglielmi[1, 5]

[1] Department of Clinical and Experimental Medicine, Foggia University School of Medicine, Foggia, Italy
[2] Radiology Unit, Carlo Urbani, Jesi, Italy
[3] Rheumatology Unit, University of Ancona, Ancona, Italy
[4] IRCCS-CROB, Rionero in Vulture, Italy
[5] Radiology Unit, Barletta University Hospital, Foggia, Italy

简评

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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绪论

脆性骨硬化是一种罕见的良性骨发育不良，患病率为五万分之一，通常无年龄或性别差异[1]。

其特征是在整个骨骼中对称分布着大量圆形或卵圆形硬化性骨病变[2]。在对不相关疾病的影像学研究中，经常会偶然发现该病变[3]。

在组织学上，病变表现为板层骨组织小梁增厚，松质骨结构内有哈弗氏系统；它们很可能是在生长和分化过程中没有变成松质骨的骨病灶。脆性骨硬化中的松质骨凝集由骨小梁的周边区域组成，其中骨细胞很少，且无成骨细胞或破骨细胞（两者都存在于不规则骨小梁的中央核心）[4,5]。

我们报告一例脆性骨硬化患者，其因晕厥于我们门诊就诊。

病例描述

一例43岁的女性患者在出现一过性意识丧失后被救护车送往急诊室。初步评估包括病史、体格检查、12导联心电图和实验室检查，未发现任何异常。因此，行全身计算机体层成像检查（CT）。CT扫描示多发小面积硬化性骨岛灶，散布于胸椎（图1a）和腰椎（图1b）、肋骨、骨盆（图2）、骶骨（图3）和双侧股骨近端（图4）。所有骨骼均无任何皮质侵蚀或骨膜反应。未见发红或水肿等其他体征。此外，患者未诉腿部或身体其他部位任何麻木、刺痛和虚弱感。

CT表现疑似脆性骨硬化。血常规、红细胞沉降率、血清电解质、肿瘤标志物、碱性和酸性磷酸酶、ANA和抗双链DNA等相关临床和实验室检查未见任何类型的关节炎、感染或成骨性骨转移，此结果已纳入鉴别诊断。锝-99m全身骨显像未检测到活性显著增加。排除其他鉴别诊断，患者被诊断为脆性骨硬化的典型放射学表现，并接受随访。

讨论

脆性骨硬化（也称为“骨斑点症”或播散型凝集性骨病）是一种罕见的骨发育不良，由H.E.Albers-Schönberg于1915年首次描述[6]。据估计，该疾病的发病率约为五万分之一，通常不存在年龄或性别差异。该病通常为常染色体显性遗传，但也有散发型的报告[1]。

目前的文献表明，这可能是由于位于12q的LEM结构域3（LEMD3）基因的功能缺失突变导致。这些突变也可能影响软组织和皮肤，导致肢骨纹状肥大，这是一种良性硬化性骨发育不良，伴有皮质骨质增生、上覆皮肤增厚和纤维化，以及布希

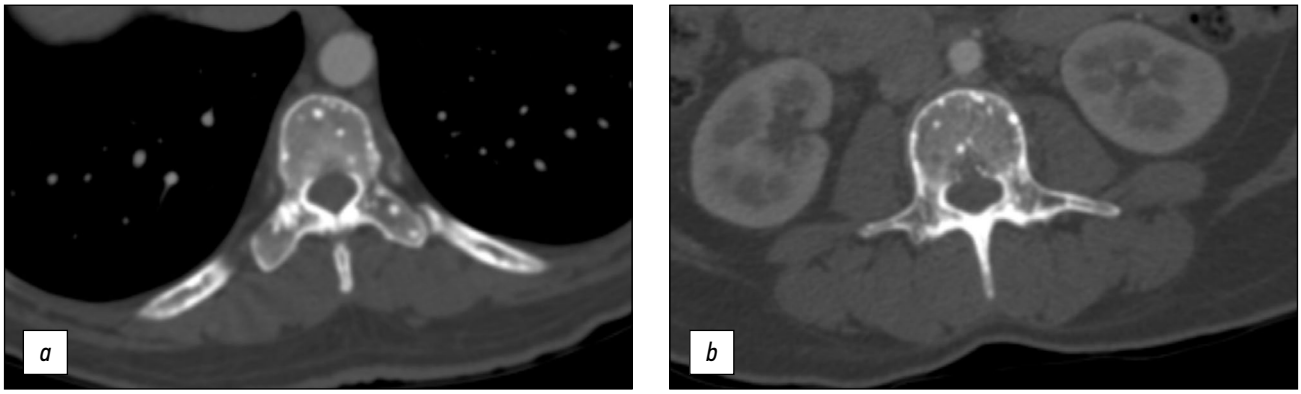

图1穿过胸椎（*a*）和腰椎（*b*）脊柱的横截面计算机断层扫描。显示棘突和椎弓根内有大量、清晰、均匀的圆形高密度病灶。

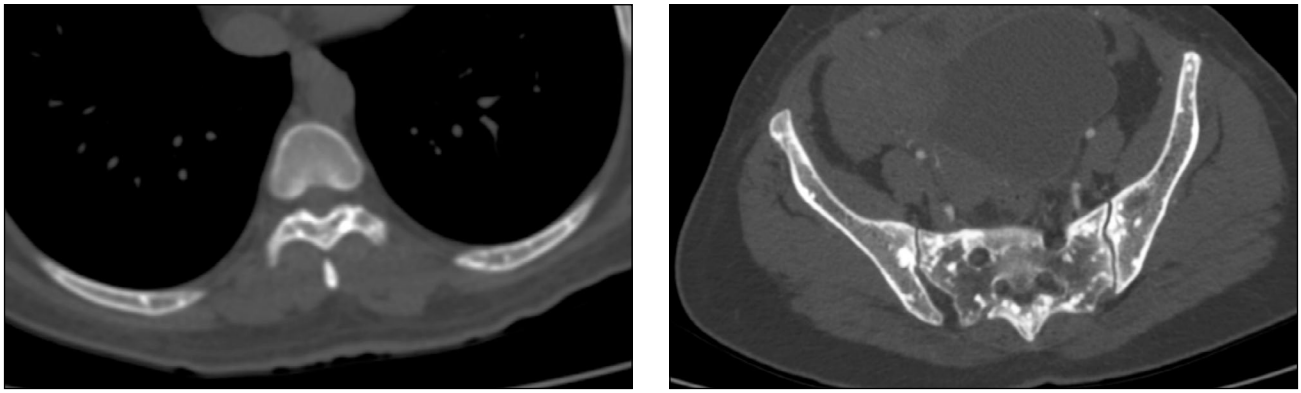

图2穿过第七根肋骨的横截面计算机断层扫描。显示大量高密度病变，边缘清晰。大小以毫米为单位。

图3穿过骶髂关节的横截面计算机断层扫描。显示沿骶骨、髋骨和骶髂关节对称分布的小而硬化的圆形阴影。

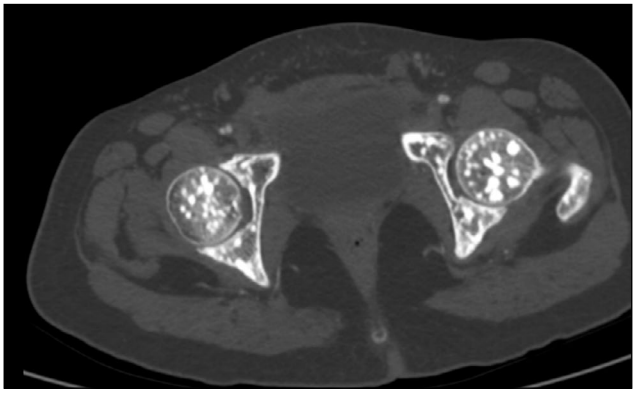

图4穿过股骨头的横截面计算机断层扫描。显示大量高密度病变，符合股骨头脆性骨硬化，病变边缘清晰。大小以毫米为单位。

克—奥伦多夫（Buschke－Ollendorff）综合征，该综合征包括与播散性结缔组织和皮肤黄色痣相关的脆性骨硬化[7,8]。脆性骨硬化病变通常是在对不相关的疾病进行影像学研究时偶然发现的[3]。

脆性骨硬化的放射学病变很典型：其特征是大量对称、均匀、边界清楚、尺寸较小（直径1-10mm）的圆形或椭圆形硬化病变。最常见的受累部位是短管骨的骨骺和长骨的干骺端。此外，据报道，腕骨和跗骨、肩胛骨、骨盆和骶骨常受到影响[9,10]。肋骨、锁骨、脊柱和颅骨的受累并不常见[11]。

由于它们的相似性，脆性骨硬化的影像学表现可能与成骨性骨转移相混淆，但它们存在显著差异，使我们能够做出鉴别诊断。与骨转移相反，脆性骨硬化中的硬化病变是对称、大小一致的，不会引起皮质侵蚀。

因此，骨显像在最终诊断中起着重要作用。事实上，正常放射性核素骨扫描通常可排除成骨性骨转移的可能性。然而，文献中报道了几例脆性骨硬化伴异常骨扫描的病例[12,13]。

结论

虽然脆性骨硬化是一种罕见疾病，但通过典型的放射学表现可以很容易地作出诊断。

临床医生必须了解并识别这种影像模式，以便做出准确诊断，防止进一步检查和积极治疗。

附加信息

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Paparella M.T.和 Gangai I.——对与主题和手稿写作相关的研究工作做出了同等贡献；Porro Ch.Eusebi L.和 Silveri F.——文献研究和数据采集；Cammarota A.和 Guglielmi G.——手稿的批判性修订。

出版同意：已获得患者的书面同意，以公布相关医疗信息和手稿中的所有附带图像。

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AUTHORS' INFO

*** Guglielmi Giuseppe,** MD, Professor;
address: Viale L. Pinto 1, 71121 Foggia, Italy;
ORCID: http://orcid.org/0000-0002-4325-8330;
e-mail: giuseppe.guglielmi@unifg.it

Paparella Maria Teresa, MD;
ORCID: http://orcid.org/0000-0003-2573-9509;
e-mail: mt.paparella@gmail.com

Gangai Ilaria, MD;
ORCID: http://orcid.org/0000-0001-9594-4810;
e-mail: hilary_ps@libero.it

Porro Chiara, MD;
ORCID: http://orcid.org/0000-0002-7526-6968;
e-mail: chiara.porro@unifg.it

Eusebi Laura, MD;
ORCID: http://orcid.org/0000-0002-4172-5126;
e-mail: lauraeu@virgilio.it

Silveri Ferdinando, MD;
ORCID: http://orcid.org/0000-0002-7847-245X;
e-mail: fsilveri@libero.it

Cammarota Aldo, MD;
ORCID: http://orcid.org/0000-0003-4211-5140;
e-mail: aldo.cammarota@crob.it

ОБ АВТОРАХ

*** Guglielmi Giuseppe,** MD, Professor;
address: Viale L. Pinto 1, 71121 Foggia, Italy;
ORCID: http://orcid.org/0000-0002-4325-8330;
e-mail: giuseppe.guglielmi@unifg.it

Paparella Maria Teresa, MD;
ORCID: http://orcid.org/0000-0003-2573-9509;
e-mail: mt.paparella@gmail.com

Gangai Ilaria, MD;
ORCID: http://orcid.org/0000-0001-9594-4810;
e-mail: hilary_ps@libero.it

Porro Chiara, MD;
ORCID: http://orcid.org/0000-0002-7526-6968;
e-mail: chiara.porro@unifg.it

Eusebi Laura, MD;
ORCID: http://orcid.org/0000-0002-4172-5126;
e-mail: lauraeu@virgilio.it

Silveri Ferdinando, MD;
ORCID: http://orcid.org/0000-0002-7847-245X;
e-mail: fsilveri@libero.it

Cammarota Aldo, MD;
ORCID: http://orcid.org/0000-0003-4211-5140;
e-mail: aldo.cammarota@crob.it

* Corresponding author / Автор, ответственный за переписку