Clinical case

Osteopoikilosis: Report of a case and review of the current literature.

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Abstract

A 19-year-old-male from Guápiles with history of congenital torticollis and obstructive urolithiasis was seen in the orthopedic clinic because of symetrical hyperostic spots in both hips and sacroilliac joints detected incidentally when an intravenous pyelogram was performed on the patient. The patient manifested pain in both lower limbs of several months' length. Hip and knee radiographs showed symmetrical sclerotic bone lesions suggestive of osteopoikilosis. A bone gammagraphy study showed no pathological findings. The possibility of any other associated medical condition was ruled out. Osteopoikilosis requires no medical treatment.

Keywords: osteopoikilosis, displasia, osteosclerosis

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Osteopoikilosis, or spotted bone disease, belongs to a group of rare clinical entities known as sclerosing bone dysplasias.[1]This entity is acortical bone ectopiainspongy bone. It is characterized by the presence of hyperostosis foci at any point in the skeleton, predominantly in the metaphysis and epiphysis of long bones, as well as in the carpal and tarsal bones.[2]

Although it is a benign bone condition, it has been described as associated with other connective tissue disorders, such as the Buschke-Ollendorf Syndrome, which may cause more serious medical problems.[3]

The description of the first case dates back to 1915. Since then, most world wide reports correspond to isolated cases, probably because family transmitted osteopoikilosis is unusual.[2,4] According to the literature reviewed, this is the first osteopoikilosis case described in Costa Rica, thus, it is a significant contribution to the study of this disorder.

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Case report

A 19-year old male, farmer, from Guápiles, with a history of congenital torticollis diagnosed at 5 years of age and with recurrent obstructive urolithiasis diagnosed five months ago. He is referred to the Orthopedics Department of the Rafael Angel Calderón Guardia Hospital because of the incidental finding in an intravenous pyelogram study of symmetrical hyperostic spots in both hips and sacroiliac joints.The patient complaints of occasional pain in both lower limbs, mild, ill-defined, non-progressive, usually in the morning that has developed for several months and reports no other symptoms of the musculoskeletal system.

There is no history of alcohol consumption, smoking, consumption of other drugs, allergies or family history.

The patient undergoes a series of x-rays, which evidence scleroticlesions; symmetrical, disc shaped, smaller than 5 mm and juxta-articularin knees, hips and sacroiliac joints (Figures 1 and 2). Furthermore, by means of a neck radiograph, the possible presence of a soft tissue mass is documented. Afterwards, a neck ultrasound is performed, which rules out pathologies. A whole body bone scan shows normal skeletal findings.

Discussion

The multifocal sclerotic bone lesions may be the result of multiple etiologic factors, with which a differential diagnosis must be made. Among the factors described are osteoblastic metastases, vascular disorders, congenital disorders, infections, traumas and endocrine disorders.

Usually the patient presents a number of symptoms and some other findings that contribute to the diagnosis.[5]

In the case studied, there are vague symptoms and very rare radiological findings, which suggest a congenital cause, specifically, osteopoikilosis.

This entity has a dominant autosomal transmission and corresponds to an alteration in the maturation of the endochondral bone, which in most cases is detected between

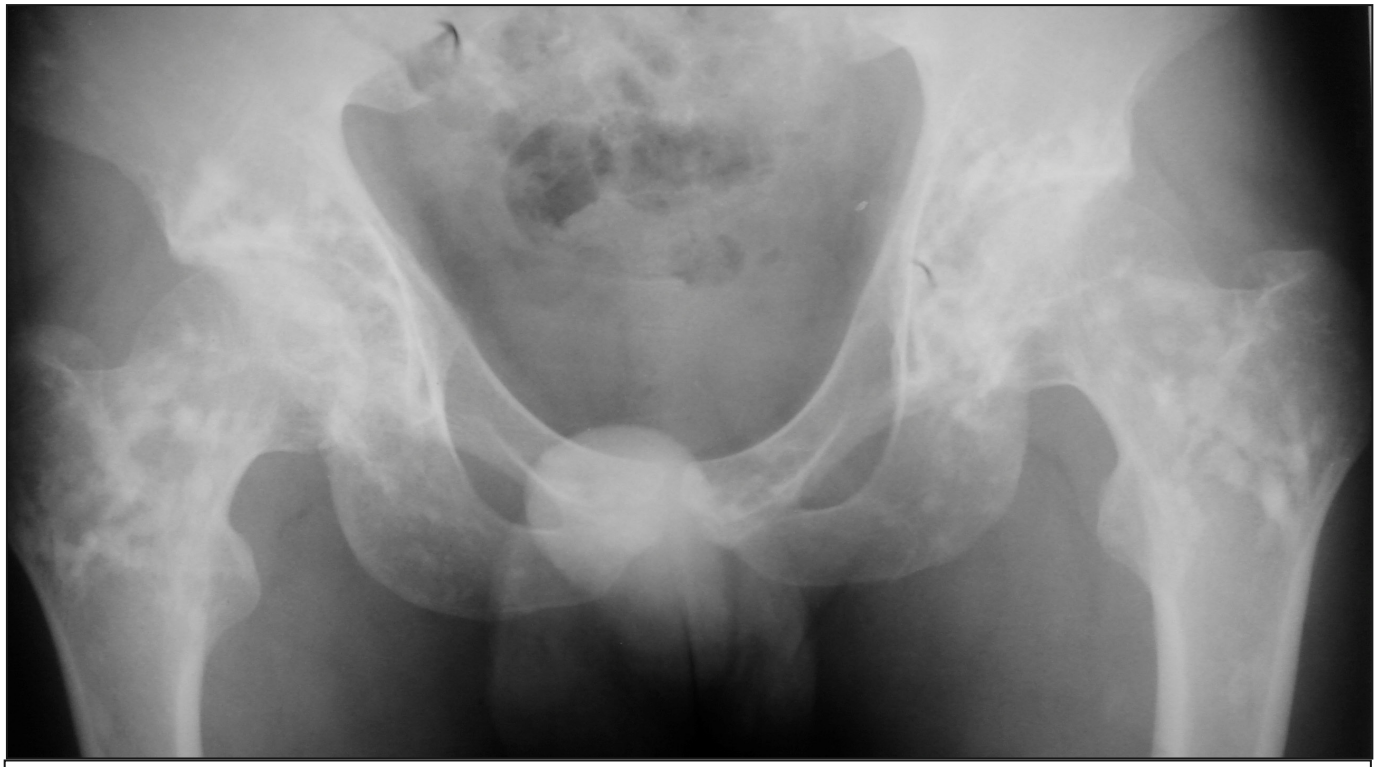

Figure 1. Hip x-ray. The sclerotic lesions can be observed (bone islands or enostosis) symmmetrical, bilateral, well defined borders, of periarticular distribution in both proximal femurs and in acetabular regions of similar sizes.

twenty and thirty years of age.[1-3] As in this case, the detection of osteopoikilosis tends to be accidental, when a radiologic study is requested for some other reason and it evidences the characteristic bone lesions.[5]

Regarding the radiological findings, it is said that the characteristic lesions persist for life. According to a study of four families, among the preferred sites and with periarticular distribution, the following stand out: phalanges (100%), carpal bones (97.4%), metacarpals (92.5%), feet phalanges (87.2%), metatarsals (84.4%), tarsal bones (84.6%), pelvis (74.4%), femur (74.4%), radius (66.7%), ulna (66.7%), sacrum (58.9%), humerus (28.2%), tibia (20.5%) and fibula (2.8%).[6]

In the Costarican case, the patient presents small sclerosis foci of various sizes, disc shaped, symmetrical and well defined; that do not alter the cortical or bone shape. Usually, to differentiate between a metastasis and an osteopoikilosis-type bone dysplasia of periarticular distribution, a whole body bone scan study is performed. In this case, it ruled out a possible metastasis. It should be noted that the differentiation through a whole body bone scan is not absolute. There are case reports of

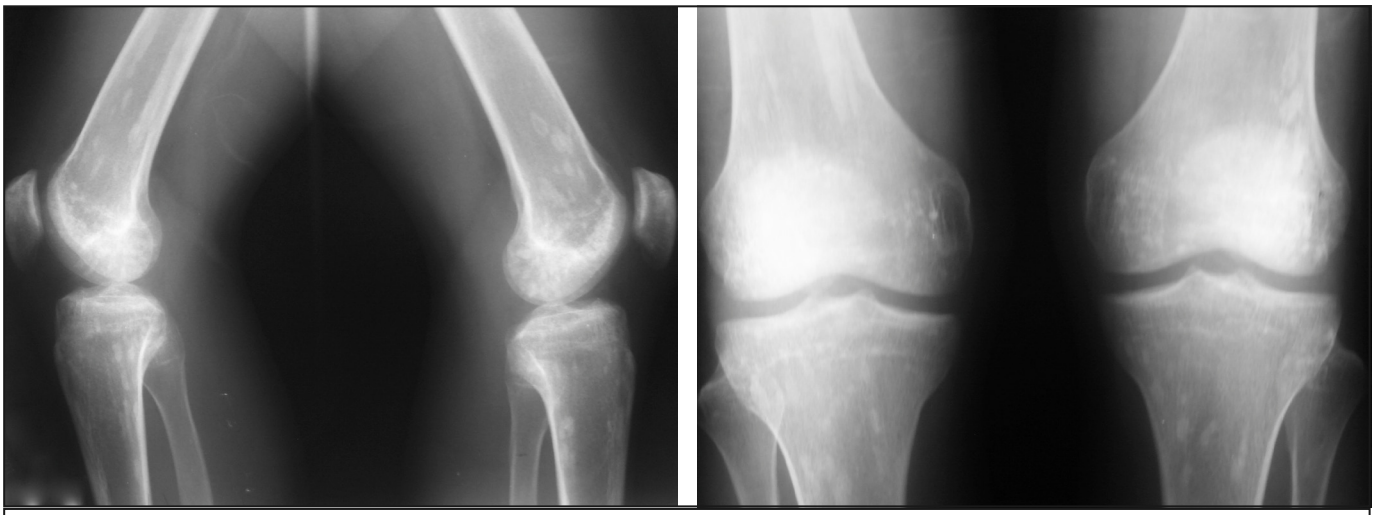

Figure 2. Knee x-ray, lateral (A) and anteroposterior (B). The same kind of lesion as the one in the one in the hip x-ray can be observed, predominantly in the meta-epiphyseal region.

osteopoikilosis that describe the detection of pathological bone findings by means of a whole body bone scan.[7]

Osteopoikilosis is considered a benign bone condition that does not require any treatment or intervention. However, the association of the osteopoikilosis family with other disorders that may require medical attention has been described; such as scleroderma, endocrine diseases, rheumatoid arthritis, syndactyly and cleft palate. Osteopoikilosis has also been associated with the Buscke-Ollendorff syndrome, as already mentioned, and with the Klippel-Feilsyndrome.[2,3]

In this case, the mentioned conditions have been ruled out. However, the concomitant presence of congenital torticollis and recurrent obstructive urolithiasis draws our attention, since the literature reviewed does not report any case that associates either of these two entities with the osteopoikilosis.

It is important to emphasize that in this case, the patient denies family history of osteopoikilosis. A history of hereditary bone disease in his family is not known either. This is expected; since it is mostly a subclinical disorder, if a radiological study in family members does not exist, its identification in other subjects is difficult.

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Conflicts of interest: Authors report no conflict of interest.

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