Osteopoikilosis: a case report of a symptomatic patient

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ABSTRACT

Osteopoikilosis (OP) is a very rare benign sclerosing bony dysplasia with an autosomal dominant inheritance. We describe the morphology of an osteopoikilosis male patient, associated with severe pain on wrist and hand joints, report on the relative literature and focus on clinical significance, due to mimicking capability of other more severe conditions such as bone metastases.

CASE REPORT

INTRODUCTION

Osteopoikilosis (OP) (synonyms: osteopathia condensans, asymptomatic bone dysplasia, spotted bone disease) is an uncommon sclerosing bone dysplasia. First detailed description of that rare pathological condition was made by Albers-Schönberg in 1915 (1). It appears with variable onset in both sexes regardless the age but preferably adult males. Incidence is not known since the disease is of little clinical importance most of the time and constitutes an incidental radiological finding. Until 1971, Szabo was able to find only 300 cases in the literature and claimed that the incidence of osteopoikilosis is 1 in every 50,000 subjects (2).

It is featured by multiple, discrete round or ovoid lumps in cancellous bones (3), particularly in epiphyses and metaphyses of long bones, scapulae, pelvis, carpi and tarsi (4).

OP is usually an asymptomatic condition (5), founded randomly in routine radiographic imaging examinations. Occasionally, OP can be painful without causing any deformity or dysfunction at the location site (6). The disease in inherited by the autosomal dominant character (7). Co-existence of OP and severe pain on the adjacent joints is very rarely reported according to our literature review.

CASE REPORT

A 50-year-old man was admitted to our Hospital, reporting the presence of diffuse severe pain, located at the right wrist and hand joints. The pain was intermittent and worsened mostly during flexion, extension, abduction and adduction of wrist and hand joints. The study of the right hand X-ray revealed the existence of multiple radio-opaque circular or ovoid spots with define linear sclerotic margins, approximately 2-4 mm in diameter each, located in cancellous bone of the distal epiphysis of radius and ulna, carpal bones, proximal and distal epiphyses of metacarpals and phalanges (Figures 1, 2). All bones were free of any cortical erosion or periosteal reaction. No other signs, such as rubor or edema were noticed. Range of movements of carpal, carpometacarpal, midcarpal, metacarpophalangeal, carpophalangeal joints was not affected. Morphology of the spots noticed on X-rays was identical to those seen in OP. Moreover, the relative clinical and laboratory tests such as routine blood count, ESR, 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.

DISCUSSION

OP is featured by numerous 2-10 mm round or oval shape densities that are symmetrically distributed within the epiphyses and metaphyses of long bones, which appear as dense radio-opaque spots in cancellous bone tissue (3, 5, 7, 8). Histologically, the foci in OP are formed by dense trabeculae of cancellous bone tissue, forming a nidus without communication with bone marrow (5, 9).

According to an epidemiologic study on 53 patients with OP, members of four families, Benli et al (7), noticed that the most frequent sites for OP appearance were the phalanges (100%), carpal bones (97.4%), metacarpals (92.3%), phalanges of the foot (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 (12.8%). It's generally accepted in the literature that OP is more frequently located in long bones and pelvis (3, 4). Male to female ratio is 3:2. Furthermore, OP appears in childhood and persists through life (7).

OP is more frequent in self-contained communities were consanguineous marriages are frequent (7). Familial clustering suggests a dominant inheritance, associated with LEMD3 gene, responsible for the disease, featured by abnormality in enchondral bone maturation process. The above, leads to bone sclerosis that appears as dense spots in radiographic imaging (2, 3, 5).

Typical cases are usually easily diagnosed with simple X-ray imaging. Physicians and radiologists must be aware in order to distinguish whether the anomalies noticed on an X-ray screening are caused by OP or other pathologies such as bone metastases. On more complicated cases, MRI may aid in differential diagnosis, showing multiple circular or ovoid hypointense lesions located in proximal or distal epiphyses, but there are very few reports of OP MRI finding in the literature (10, 11). In addition, radionuclide bone scan (scintigraphy) can also help distinguishing OP from osteoblastic bone metastases, but abnormal bone scan does not exclude OP (12, 13). Bone scintigraphy demonstrates usually absence of radiotracer uptake in OP patients.

Although, OP is a rather rare condition, there are several diseases that should be taken under consideration for its differential diagnosis, such as osteoblastic metastasis, tuberous sclerosis, Paget's disease, mastocytosis, osteopathia striata, melorheostosis, synovial chondromatosis, sesamoid ossicles and Ollier's disease (3, 7, 14). Among these conditions the most critical to differential diagnose when thinking about OP is bone metastases that represent the most common malignant bone tumors (15). This fact is supported by Carpintero et al (16), who reported that 5 out of 10 patients with OP admitted with findings similar to osteoblastic metastasis. The sclerotic lesions in OP are symmetrical, bone epiphyses and metaphyses are mainly involved, foci are uniform in size and never induce cortical erosion. On the other hand, osteoblastic metastases may cause subcortical destruction. OP diagnosis can be supported when blood levels of alkaline and acid phosphatase are within normal range and bone scan is of normal appearance (3, 5, 12, 17).

It has been reported that in 15-20% of patients, slight joint pain and effusion could be noticed (3). However, pain can be emerged without any deformity or dysfunction at the location site (6). The mechanisms of joint pain are of unknown origin and only hypotheses can be made. The produced joint pain presumably is an incidental finding in the course of such disease. OP bony lesions are focal condensations of cancellous tissue subject to the same metabolic changes as the surrounding bone (6). Due to the fact that OP lesions in adults can occasionally disappear and then reappear in denser pattern (18), someone could consider that an increase of these bony condensations, under unknown circumstances, might induce pain due to increased localized bone metabolism. The joint pain is possibly caused by irritation of the joint capsule attachment by sclerotic areas that appear during instant increased metabolic activity. Furthermore, an increased intraosseous pressure due to venous stasis at the area of the lesion could evoke pain at the region. Sometimes OP is associated with dermatological conditions just as those in Buschke-Ollendorff syndrome. In such cases, OP co-exists with disseminated white skin lesions (dermatofibrosis lenticularis) (4, 19). Another bone condition that can coexist with OP is melorheostosis, a genetically based disease (4). Synovial osteochondromatosis, spinal stenosis, keloid formation, scleroderma-like lesions, dystocia, dwarfism, dacryocystitis, rheumatoid arthritis complicated with dry eyes and ankylosing spondylitis with familiar Mediterranean fever are also associated conditions with OP (3, 20, 21, 22, 23).

Due to the benign nature of OP complications are very rare (7). Possible complications described in the literature are osteosarcoma (24), giant cell tumor (25) and chondrosarcoma (26). The three above mentioned complications are very rare and unique to best of our knowledge.

TEACHING POINT

Despite the fact that osteopoikilosis is a very rare asymptomatic condition that most physicians are not familiar with, it is valuable to take it into consideration, particularly when diagnostic issues on bone radiography occur and severe pain at the adjacent joints co-exists.

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FIGURES

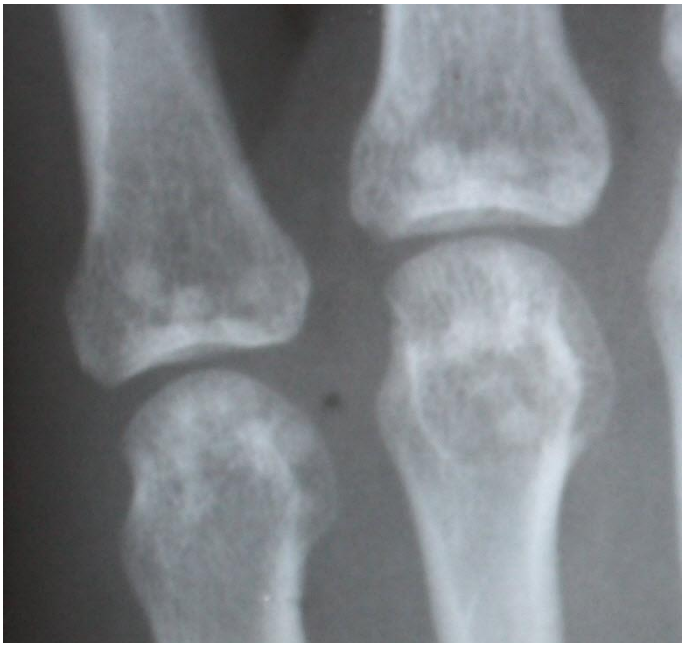

Figure 1 (magnified view – complete hand radiograph on next page): 50-year-old man with osteopoikilosis. Anteroposterior, right hand radiograph. Multiple radio-opaque circular or ovoid dense spots with define linear sclerotic circumference, located at the proximal and distal epiphyses of (here 4th and 5th) phalanges and metacarpals are noticed.

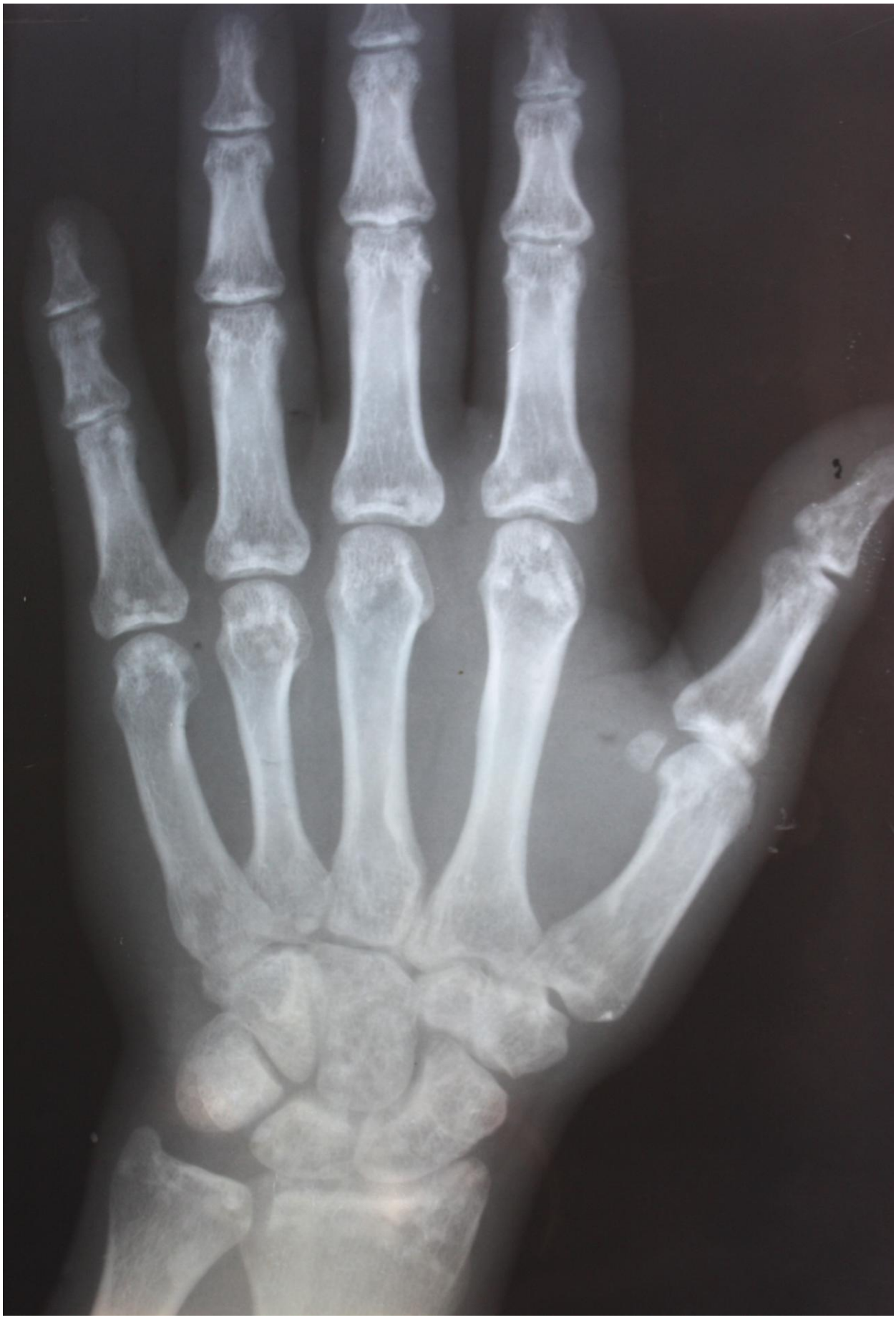

Figure 1: 50-year-old man with osteopoikilosis. Anteroposterior, right hand radiograph. Digits are in extension, with respect to the longitudinal axis of the 3rd digit. Multiple radio-opaque circular or ovoid dense spots with define linear sclerotic circumference, located at the proximal and distal epiphyses of phalanges and metacarpals, carpal bones and distal epiphyses of radius and ulna are noticed. All bones were free of any cortical erosion or periosteal reaction.

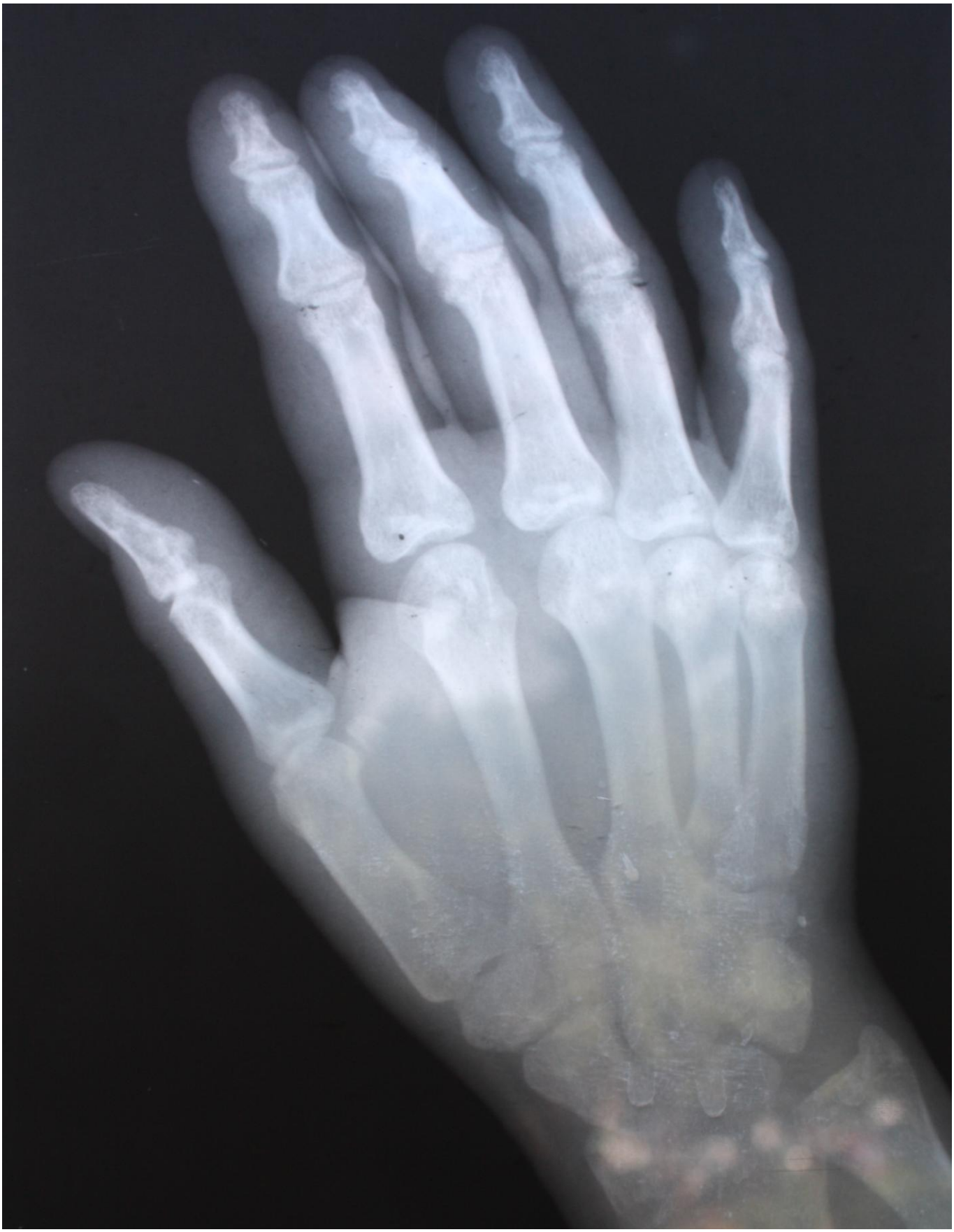

Figure 2: 50-year-old man with osteopoikilosis. Oblique, right hand X-ray image of a 50 year old male. The rim is placed 10o degrees oblique in relation to sagittal plane. Digits of carpometacarpal and phalangophalangeal joints are relaxed.
Multiple radio-opaque circular or ovoid dense spots with define linear sclerotic circumference, located at the proximal and distal epiphyses of phalanges and metacarpals, carpal bones and distal epiphyses of radius and ulna are noticed. All bones were free of any cortical erosion or periosteal reaction.

ABBREVIATIONS

ANA = anti-nuclear antibodies
anti-DS-DNA = anti-double stranded DNA antibodies
ESR = erythrocyte sedimentation rate
mm = millimeters
MRI = magnetic resonance imaging
OP = osteopoikilosis

KEYWORDS

Osteopoikilosis, severe pain, differential diagnosis