Case Report
DOI: 10.5152/tftrd.2015.39019
Turk J Phys Med Rehab 2015;61:375-9

Osteopoikilosis-A Case Report

Nurten KÜÇÜKÇAKIR, Lale ALTAN İNCEOĞLU, Serap LATİF RAİF
Department of Physical Medicine and Rehabilitation, Uludağ University Faculty of Medicine, Bursa, Turkey

Abstract

Osteopoikilosis (Spotted Bone, Osteopathia Condensans Disseminata) is a rare autosomal dominant bone disorder with characteristic radiological features. Prevalence has been estimated 1/50.000. The diagnosis usually done according to radiographs which are performed incidentally. Diagnostic lesions of osteopoikilosis are typically diffuse, round or oval, symmetrically shaped sclerotic bone areas. It should be differentiated from osteoblastic metastasis, tuberous sclerosis and mastocytosis due to the similarity of radiological images. We report a case of osteopoikilosis patient presenting with left hip and left groin pain.
Keywords: Osteopoikilosis, osteosclerotic dysplasia, ostheopathia condensans disseminata

Introduction

Osteopoikilosis (spotted bone disease, osteopathia condensans disseminata) is a rare osteosclerotic dysplasia that shows an autosomal dominant transmission and has characteristic radiological features (1,2). Although the exact cause and the pathogenesis are unknown, the inherited deficiency in the normal formation of trabeculation along stress lines of bones is thought to be responsible for the pathogenesis (3). The prevalence was determined as 1/50,000 and the diagnosis can be made at any age range (4). Although lesions are seen in both sexes, they are more common in men than in women (4). It is generally asymptomatic and does not require treatment (5). The diagnosis is often radiological or incidental (1,5,6). Osteopoikilosis is histologically defined as bone islands that form dense core structure and are not associated with bone marrow in trabecular or cancellous bone (7). Radiologically, many sclerotic lesions of symmetric and homogeneous structure that are typically circular or ovoid can be observed; each lesion has a size of a few millimeters (1,6). These lesions are more prominent in long bones and the metaphyseal and epiphyseal pelvis; they are also common in phalanges and in spongy tissues of the tarsal and carpal bones (1). Osteopoikilosis can be confused with osteoblastic metastases, tuberous sclerosis, and bone mastocytosis because of the similarity of radiological images (1). These diseases should be considered in the differential diagnosis.

In this article, a case of patient with osteopoikilosis who presented to our clinic because of pain in the left hip and left groin area is presented.

Case Report

A 26-year-old male patient presented at our clinic with the complaint of left hip and left groin pain. The patient gave a history of increase in pain over the last month. The pain had a mechanical character and increased especially while walking. There was no night pain, pain spread, and morning stiffness. The patient did not describe any numbness, tingling, and weakness in the legs.

Address for Correspondence: Nurten Küçükçakır, MD, E-mail: nurtenkcakir@hotmail.com
Received: October 2013 Accepted: March 2014

Cite this article as:
Küçükçakır N, Altan İnceoğlu L, Latif Raif S. Osteopoikilosis-A Case Report.
Turk J Phys Med Rehab 2015;61:375-9.

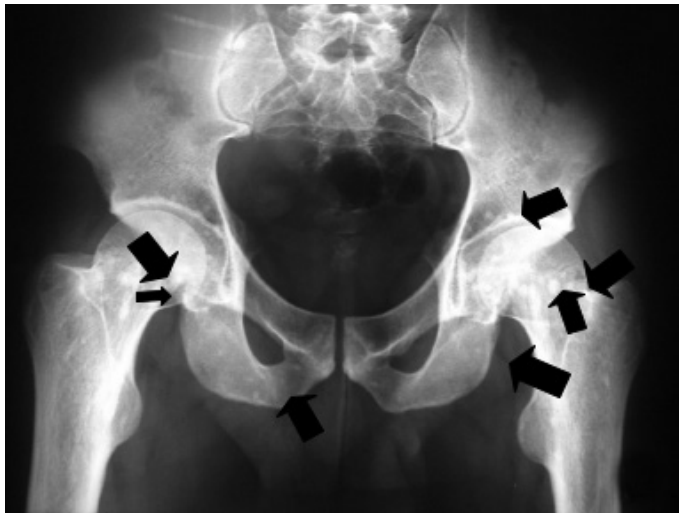

Figure 1. Anteroposterior radiograph of the pelvis. Black arrows show the femoral head and proximal acetabulum; a number of small, well-circumscribed, circular sclerotic bone lesions in the ischium

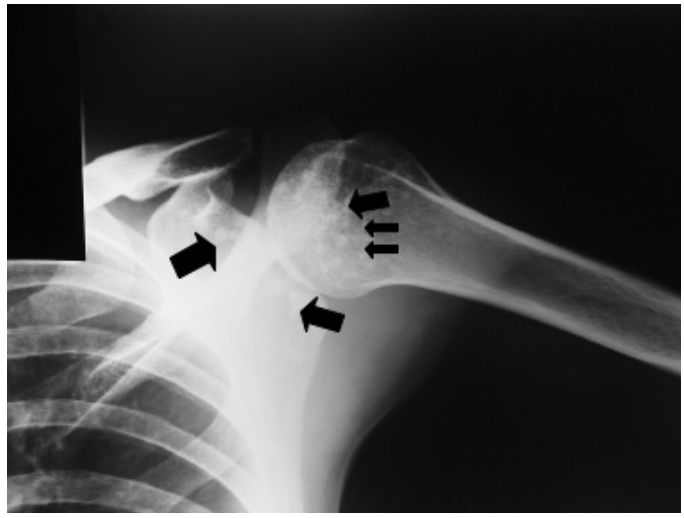

Figure 2. Anteroposterior radiograph of the left shoulder. Black arrows show the humeral head and widely distributed, small, well-circumscribed and circular sclerotic bone lesions around the glenoidal cavity

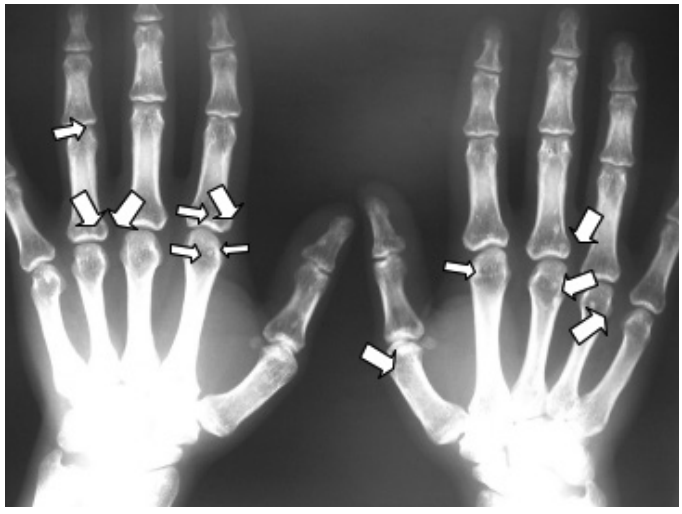

Figure 3. Bilateral anteroposterior radiograph of the hand. White arrows show widely distributed sclerotic lesions

In the examination and inspection of the patient, no skin lesions or rashes, including those in the scalp, were encountered. Other systemic examination findings were normal. In the musculoskeletal examination, tenderness was detected in the following joints: 1st right metacarpophalangeal (MCP) joint; 2nd, 3rd, and 4th MCP, 4th proximal left interphalangeal (IP) joint; right 1st, 2nd, 3rd, 4th, and 5th metatarsophalangeal (MTP) joints; left 2nd and 4th MTP joints, right ankle; and anterior aspect of both shoulder joint capsules. However, no arthritis was detected in any joint. Waist and hip movements were unaffected and painless. Patrick's test was positive on the left at the border. His neurological examination was normal. In laboratory examination, routine biochemistry, erythrocyte sedimentation rate, C-reactive protein, urinalysis, and tumor marker levels were normal. In

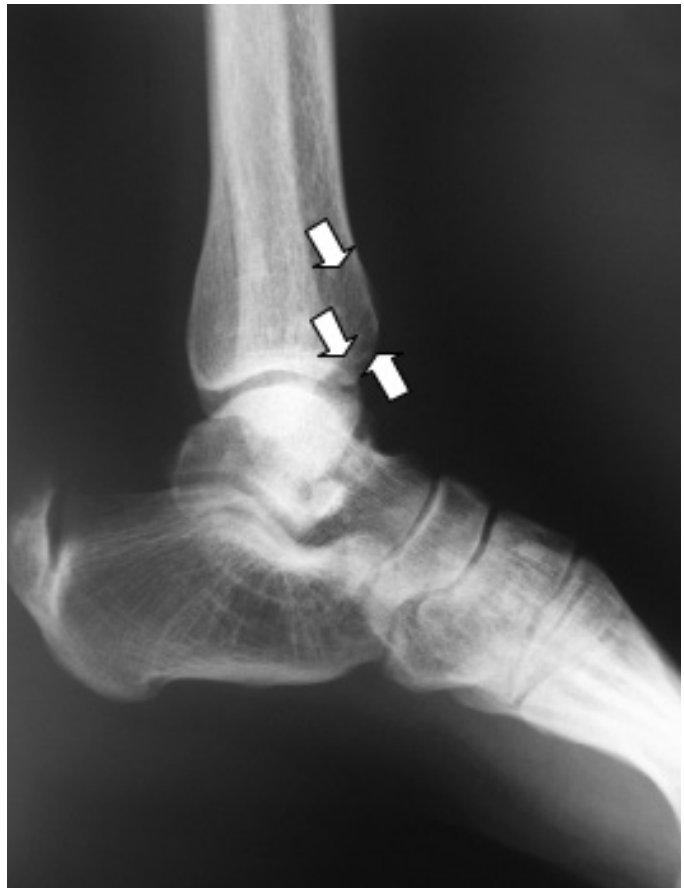

Figure 4. Radiograph of the left ankle. White arrows indicate sclerotic lesions

the radiological assessment, no pathological findings were found in lumbar and chest radiographs. Scattered, small, well-

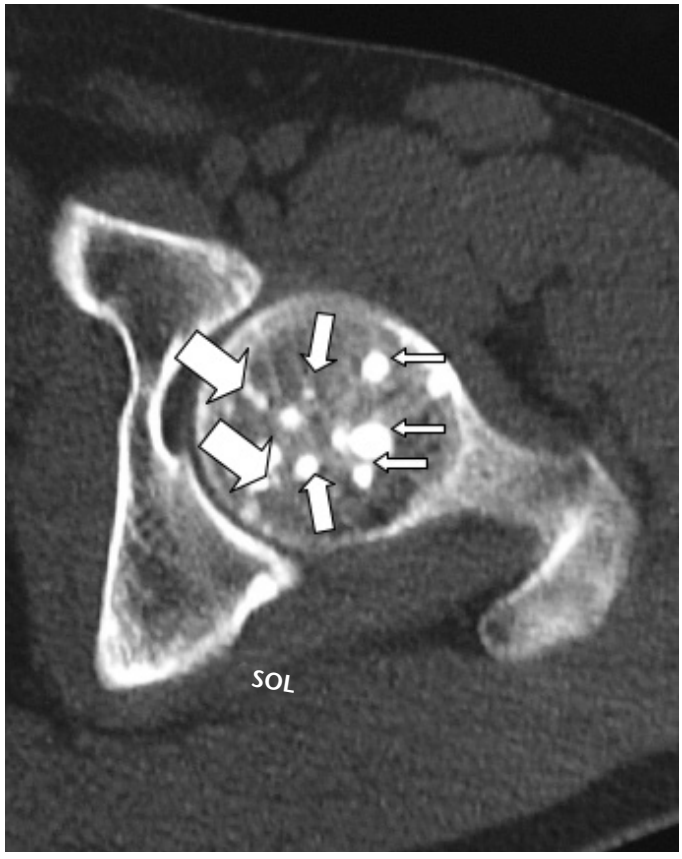

Figure 5. The left hip computed tomography, transverse cross-section passing through the femoral head. White arrows show numerous hyperdense lesions that conform with the osteopoikilosis in the femoral head; lesions are well-circumscribed and have size in millimeters

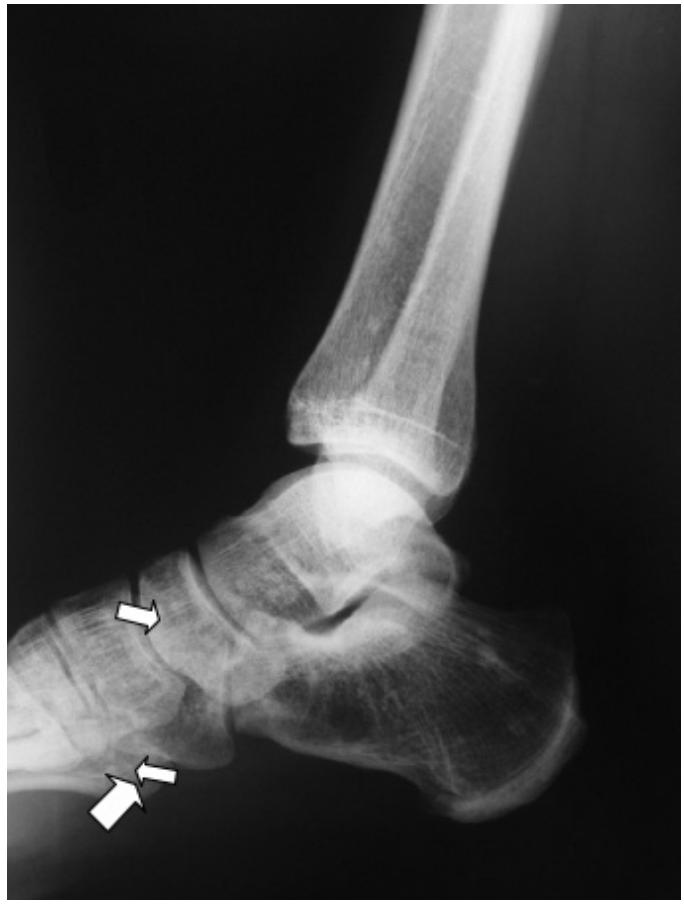

Figure 6. Radiograph of the left side of the ankle of the mother of 26-year-old patient. The white arrows show well-circumscribed sclerotic lesions

circumscribed, and circular sclerotic bone lesions were detected in the anteroposterior radiographs of the pelvis (Figure 1) and the shoulder (Figure 2). Similar lesions were also detected in the anteroposterior radiographs of the hands (Figure 3) and the left ankle (Figure 4). Computed tomography (CT) of the left hip joint and whole-body bone scintigraphy were then performed. CT of the left hip joint showed multiple scattered and hyperdense areas of size in millimeters (Figure 5) located in the left part of the femoral head and neck and in the acetabulum. This view suggested a diagnosis of osteopoikilosis. No significant increase in the activity was detected in Technetium-99m (Tc-99m) whole body bone scintigraphy.

The patient was diagnosed with characteristic radiological findings and osteopoikilosis by excluding other differential diagnoses. The patient was informed about the disease and was followed up. Similar lesions were observed in the evaluation of patient's mother among the other family members at our polyclinic; the mother had prominent lesions in her left ankle (Figure 6). The family was directed to the department of genetics for genetic examination. Paracetamol 1500 mg/day was started as treatment for the patient's hip pain. A decrease in hip pain was detected in the follow up a month later. A written informed consent was obtained from the patient for the publication of this report.

Discussion

Osteopoikilosis is a rare bone disease first described in the beginning of 20th century by Albert–Schönberg (8). The incidence of the disease was reported as 1/50,000 (9). Although it was reported that this disease can be observed in both sexes and at any age (10,11) and usually shows autosomal dominant transmission, sporadic cases are also reported (5,10,12). In the examination of the first-degree relatives of our patient, typical osteopoikilosis lesions were also observed in the patient's mother.

Although the disease usually remains asymptomatic, joint effusion occurs in 15%–20% patients with moderate joint pain (1,6). Because the disease does not have typical clinical findings, diagnosis is often radiological or incidental. Hip pain and tenderness in the finger, leg and shoulder joints suggest that he is in the symptomatic group.

Radiological lesions of osteopoikilosis are typical. They are characterized by numerous symmetrical, homogeneous, well-circumscribed, small (1–10 mm in diameter), round or oval-sha-

ped sclerotic lesions. The most commonly affected areas are the epiphyses of short tubular bones and the metaphyses of long bones. In addition, carpal bones, tarsal bones, scapula, pelvis, and sacrum are reported to be frequently affected (13,14). In our case; small, well-circumscribed, circular sclerotic bone lesions were detected in the pelvis, shoulder, hand, and foot radiographs in accordance with the literature.

The radiological lesions of osteopoikilosis may frequently be confused with osteoblastic bone metastases because of their similarity. For this reason, the accurate diagnosis of this benign disease is important (1,15,16). Osteoblastic metastases are the lesions that are more asymmetrical than the osteopoikilosis lesions, and they vary in size and occur in more locations. Axial skeleton involvement, positive scintigraphy findings, and bone destruction at the forefront suggest metastases. Bone scintigraphy, especially, has a crucial role in definitive diagnosis. Although bone scintigraphy is usually normal (1,17) in osteopoikilosis, unusual involvements may be seen, especially in younger patients (18-20). Osteopoikilosis diagnosis is not excluded with abnormal involvement (8,10). Increased involvement is thought to be an indicator of active bone remodeling in osteopoikilosis lesions (10,21). Four different cases at 29–35 years of age having abnormal scintigraphic involvements were reported in the literature (10,20,22,23). If the cellular activity in the lesions of osteopoikilosis continues, it should not be neglected that malignant transformation may develop in these areas (24). With regard to the differential diagnosis, apart from osteoblastic bone tumors, mastocytosis, tuberous sclerosis, osteopathy striata, melorheostosis, and osteoma should be considered; in addition, it should be remembered that they may occur concurrently (5). Melorheostosis presents with a more painful clinical picture because of the associated bone deformities, ossification, and nerve compressions in the soft tissues around the joint (25). It can concurrently present with enchondroma (26). Compared with other concomitant conditions, the association of osteopathia striata type 1 and melorheostosis, which has a more severe clinical picture with ectopic bone formation, with osteopoikilosis has been reported to be more frequent in endosteal and periosteal areas of long bones (13). In mastocytosis and tuberous sclerosis; symmetricity, metaphyseal and epiphyseal involvement, uniform and well-circumscribed focii are rarely seen in lesions (1).

Although osteopoikilosis is generally accepted as benign, clinical situations that may accompany and complications must be considered. Although isolated osteopoikilosis does not mandate treatment, the therapeutic necessity arises in the presence of accompanying pathological and clinical findings (1,5).

Cutaneous lesions with a number of different features may be encountered in 25% of these patients. Discoid lupus erythematosus, keloids (1,27), and dermatofibroma are often concomitant skin lesions; these lesions associated with osteopoikilosis are related to metabolically active fibroblast proliferation, similar to the pathogenesis osteopoikilosis lesions (28). The association of dermatofibroma with osteopoikilosis was described as Buschke–Ollendorff syndrome (dermatofibrosis lenticular disseminada) (29). In addition, the association of osteopoikilosis with musculoskeletal disorders, organ abnormalities (coarctation of the aorta, double ureter, precocious puberty, urogenital defects) (4), endocrine dysfunction (diabetes mellitus), dental, facial anomalies (30), dacryocystitis (31), scleroderma (31) fibromyalgia syndrome, spinal canal stenosis, and myelopathy (30,32) was previously described. Our patient dies not have skin lesions, developmental anomalies, or rheumatic disease.

Because of the possibility of osteosarcoma development to be associated with active osteogenesis in osteopoikilosis (15), regular follow-up of patients with osteopoikilosis is also important.

Conclusion

Although osteopoikilosis is a rare condition especially in young patients, it is diagnosed through typical radiological findings. In the literature, an algorithm regarding the diagnosis and treatment is not yet available in patients with osteopoikilosis. Therefore, radiological diagnosis of these rare patients is important to avoid unnecessary invasive methods of diagnosis and aggressive treatments.

Informed Consent: Informed consent was obtained from patient who participated in this case.

Peer-review: Externally peer-reviewed.

Author Contributions: Concept - N.K.; Design - N.K., S.L.R.; Supervision - L.A.İ.; Resources - L.A.İ.; Materials - N.K.; Data Collection and/or Processing - N.K.; Analysis and/or Interpretation - N.K., S.L.R.; Literature Search - N.K.; Writing Manuscript - N.K.; Critical Review - L.A.İ.; Other - S.L.R.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study has received no financial support.

References

1. Resnick D, Niwayama G. Enostosis, hyperostosis and periostitis. In:Resnick D, editor. Diagnosis of Bone and Joint Disorders. Philadelphia:W.B. Saunders Company; 1995;6:4404-11.
2. Benli IT, Akalın S, Boysan E, Mumcu EF, Kiş M, Türkoğlu D. Epidemiological, clinical and radiological aspects of osteopoikilosis. J Bone Joint Surg 1992;74:504-6.
3. Lagier R, Mbakop A, Bigler A. Osteopoikilosis: a radiological and pathological study. Skeletal Radiol 1984;11:161-8. [CrossRef]
4. Günal I, Seber S, Başaran N, Artan S, Günal K, Göktürk E. Dacryocystitis associated with osteopoikilosis. Clin Genet 1993;44:211-3. [CrossRef]
5. Sarralde A, Garcia-Cruz D, Nazara Z, Sanchez-Corona J. Osteopoikilosis: Report of a familial case. Genet Counsel 1994;5:373-5.
6. Yochum TR, Rowe LJ. In: Yochum TR, Rowe LJ, editors. Essentials of skeletal radiology 3rd edn. Lippincott Williams&Wilkins, Baltimore, Philadelphia 2005.
7. Walker GF. Mixed sclerosing bone dystrophies. Two case reports. J Bone Joint Surg 1964;46:552-654.
8. Wadhwa SS, Mansberg R. Abnormal bone scan in osteopoikilosis. Clin Nucl Med 1999;24:71-2. [CrossRef]
9. Allen DC, Grenon H. The association of Buschke-Ollendorf syndrome and nail-patella syndrome. J Am Acad Dermatol 2002;46:621-5. [CrossRef]
10. Mungovan JA, Tung GA, Lambiase RE, Noto RB, Davis RP. Tc-99m

MDP Uptake in osteopoikilosis. Clin Nuclear Med 1994;19:6-8. [CrossRef]
11. Busch KFB. Familiar disseminated osteosclerosis. Acta Radiol 1937;18:693. [CrossRef]
12. Berlin R, Hadensiö B, Lilja B, Linder L. Osteopoikilosis:a clinical and genetic study. Acta Med Scand 1967;18:305-14.
13. Vanhoenacker EM, De Beuckeleer LH, Wan Hul W, Balemans W, Tan GJ, Hill SC, et al. Sclerosing bone dysplasias: genetic and radioclinical features. Eur Radiol 2000;10:1423-33. [CrossRef]
14. Amezcua-Guerra LM, Mansilla Lory J, Fernandez Tapia S, Pijoan C, Leboreiro I, Martínez-Lavín M, et al. Osteopoikilosis in an ancient skeleton: more than a medical curiosity. Clin Rheumatol 2005;24:502-6. [CrossRef]
15. Mindell ER, Northup CS, Douglass HO Jr. Osteosarcoma associated with osteopoikilosis. J Bone Joint Surg Am 1978;60:406-8.
16. Mesci E. Coexistence of osteopoikilosis with reactive arthritis: a case report. Rheumatol Int 2006;26:672-5. [CrossRef]
17. Whyte MP, Murphy WA, Siegel BA. 99mTc-pyrophosphate bone imaging in osteopoikilosis, osteopathia striata, and melorheostosis. Radiology 1978;127:439-43. [CrossRef]
18. An YS, Yoon JK, Lee MH, Joh CW, Yoon SN. Abnormal bone scan in an adult with osteopoikilosis. Clin Nucl Med 2004;29:856-8. [CrossRef]
19. Taşkaynatan MA, Hazneci B, Yaşar E, Özgül A, Kalyon TA. Osteopoikiloz: iki olgu sunumu. Romatizma 2005;1:49-54.
20. Bal S, Turan Y, Deniz G, Gürgan A. Anormal Kemik Sintigrafisi Olan Osteopoikilozlu Bir Olgu. Türk Fiz Tıp Rehab Derg 2008;54:69-72.
21. Carpintero P, Abad JA, Serrano P, Serrano JA, Rodríguez P, Castro L. Clinical features of ten cases of osteopoikilosis. Clin Rheumatol 2004;23:505-8. [CrossRef]
22. Tunç E, Savaş S. Osteopoikilosis: Report of a familial case. TJMS 1999;29:701-4.
23. Albers-Schönberg HEL. Eine seltene, bisher nicht bekannte Struktur-anomalie des skelettes. Fortschr Röntgenstr 1916;23:174-7.
24. Havitcioğlu H, Günal I, Göçen S. Synovial chondromatosis associated with osteopoikilosis; a case report. Acta Orthop Scand 1998;69:649-50. [CrossRef]
25. Debeer P, Pykels E, Lammens J, Devriendt K, Fryns JP. Melorheostosis in a family with autosomal dominant osteopoikilosis: report of a third family. Am J Med Genet A 2003;119A:188-93. [CrossRef]
26. Bovee JV. Multiple osteochondromas. Orphanet J Rare Dis 2008;3:3. [CrossRef]
27. Bicer A, Tursen U, Ozer C, Kaya TI, Dusmez D, Ikizoglu G. Coexistence of osteopoikilosis and discoid lupus erythematosus: a case report. Clin Rheumatol 2002;21:405-7. [CrossRef]
28. Verbov J. Buschke-Ollendorf syndrome. (Disseminated dermatofibrosis with osteopoikilosis). Br J Dermatol 1977;96:87-90. [CrossRef]
29. Roberts NM, Langtry JA, Branfoot AC. Case report: Osteopoikilosis and the Buschke-Ollendorff syndrome. Br J Radiol 1993;66:468-70. [CrossRef]
30. Borman P, Özoran K, Aydofi, Coflkun S. Osteopoikilosis: report of a clinical case and review of the literature. Joint Bone Spine 2002;69:230-3. [CrossRef]
31. Ostrowski DM, Gilula LA. Mixed sclerosing bone dystrophy presenting with upper extremity deformities. A case report and review of the literature. J Hand Surg 1992;17:108-12. [CrossRef]
32. Weisz GM. Lumbar spinal canal stenosis in osteopoikilosis. Clin Orthop 1982;166:89-92. [CrossRef]