CASE REPORT

OSTEOPOIKILOSIS PUNCTATA – BENIGN SPOTTED DISEASE; A CASE REPORT; A RARE CAUSE OF BONE PAIN AND MIMICKER OF METASTASIS ON RADIOGRAPHS

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ABSTRACT

AIM: To describe Osteopoikilosis Punctata and its associated clinical and radiological findings. **BACKGROUND:** Osteopoikilosis (OPK), also referred to as spotted bone disease, disseminated condensing osteopathy, or osteopecilia, is a benign, autosomal dominant rare boney disorder characterized by sclerosing bony dysplasia and multiple benign enostoses. It usually presents as symmetrically distributed small, numerous, well-defined, homogenous circular radio densities that are clustered in the periarticular regions of the metaphysis and epiphysis of the long bones. However, few rare occurrences have been reported where they diffusely present throughout axial and appendicular skeleton. Radiologically they can be confused with metastasis. There is no gender and age predilection and are usually asymptomatic and found incidentally, however, in 15-20% of the patients, joint effusion is seen on inspection and slight juxta-articular tenderness is felt on palpation. Histological features are similar to bony islands and it may be associated with synovial osteochondromatosis, connective tissue disorders and a rare bone condition called melorheostosis. **CASE DESCRIPTION:** We present a case of OPK Punctata in a 27 years old male patient, presenting with complaints of mild bilateral shoulder pain and referred cervical region pain, which was non radiculopathy in nature. Upon radiological investigations (Plain radiography and CT scans), typical features of OPK punctata were noted but he was referred to our sarcoma service from abroad for expert opinion to exclude metastatic disease. After confirmation of diagnosis on radiology, his shoulder pain was managed along the lines of sub acromial impingement which completely settled within 03 months and he remains symptom free till last follow up. **CONCLUSION:** Osteopoikilosis may be an isolated finding or associated with other pathologies, e.g. skin manifestations, rheumatic and/or skeletal disorders. Due to its mimicking capability of other more severe conditions such as bone metastases and an extremely uncommon cause of bone pain, the main differential diagnosis is osteoblastic metastasis as per clinical correlation of the investigations.

Keywords: Osteopoikilosis, sclerosing bone dysplasia, osteoblastic metastasis, bone pain.

Introduction

Osteopoikilosis (OPK) is a rare bony dysplasia of unknown etiology.[1] It is also called as spotted bone disease or osteopathia condensans disseminata and is characterized by an abnormality in bone maturation process. It is often found incidentally on radiologic examination. It was initially defined and described by Albers-Schonberg in 1915.[2] The occurrence of this rare sclerosing bone dysplasia is autonomous of gender and age. Its overall incidence is stated to be one in every 50,000 subjects.[3] It usually presents as symmetrically distributed small, numerous, well-defined, homogenous ovoid to circular radio densities

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that are clustered in the periarticular region. These radio opaque lesions are commonly found in the metaphyses and epiphyses of long bones, pelvis, tarsi, scapulae and carpi.[4]

It is usually asymptomatic, however, rarely in 15-20% of the patients joint effusion is seen on inspection and slight juxta-articular tenderness is palpated upon examination. In some patients pain is the main presenting symptom of this condition; even though, pain itself is not a prominent feature of OPK punctata. Sometimes this disorder is associated with other rare complications and abnormalities such as spinal canal stenosis, dacryocystitis, dermatofibrosis lenticularis disseminata, scleroderma, discoid lupus erythematosus, keloids, syndactyly, cleft palate, heart or renal malformations, endocrine disorders, and autoimmune disorders.[4,5,6]

There are three main forms of this illness: mixed type, speckled type and striated type.[7] Majority of the cases are autosomal dominant osteosclerotic (ADO) dysplasia characterized by numerous hyperostotic areas that tend to localize in periarticular osseous regions. Sporadic forms are also presented. Heterogeneous mutations in LEMD3 gene are associated with the autosomal dominant variant of OPK.[8] It is classified as a form of bone dysplasia because the primary pathology of this condition involves disruptions in the bone maturation process.[9]

Case Report

A 27-year-old man presented to our sarcoma service from abroad with history of shoulder pain and non radiculopathic cervical region pain. The history was of insidious onset and of about 6 month s duration. Symptoms were mostly exaggerated with overhead activities and internal rotation of shoulder. He was otherwise fit and well with no co morbidities or associated features of any clinical significance. He had plain radiographs from the referring country following which he also underwent CT scans as a concern of metastatic disease was raised and was subsequently referred to our sarcoma service for expert opinion. He underwent various blood tests which were all normal including a complete blood count, Serum Calcium and erythrocyte sedimentation rate. On examination was noted to have pain on forward flexion and abduction beyond 70 degrees and internal rotation up to mid buttock region. Hawkins Kennedy test was positive for impingement. Neck movements were preserved with a negative Spurlings test.

Plain radiographs and computed tomography (CT) scans are of paramount importance in these cases and show multiple small hyperdense (sclerotic), rounded, ovoid or even linear lesions in the skeleton, more so around the physis and measure 2-10 mm in diameter (Fig.1-3). The lesions are seen symmetrically throughout the appendicular skeleton with relative sparing of skull. The imaging differentials of these lesions include sclerotic metastases and tuberous sclerosis. On MRI, the lesions appear hypointense on both T1 and T2 weighted images due to presence of dense cortical bone. The radiological appearances are typical and usually no additional imaging is required unless the patient has a known malignancy and metastasis has to be excluded. To differentiate from osteoblastic metastasis, bone scintigraphy with technetium-99m can be performed, which would show hot spots (increased uptake) in metastases and no uptake in OPK lesions.

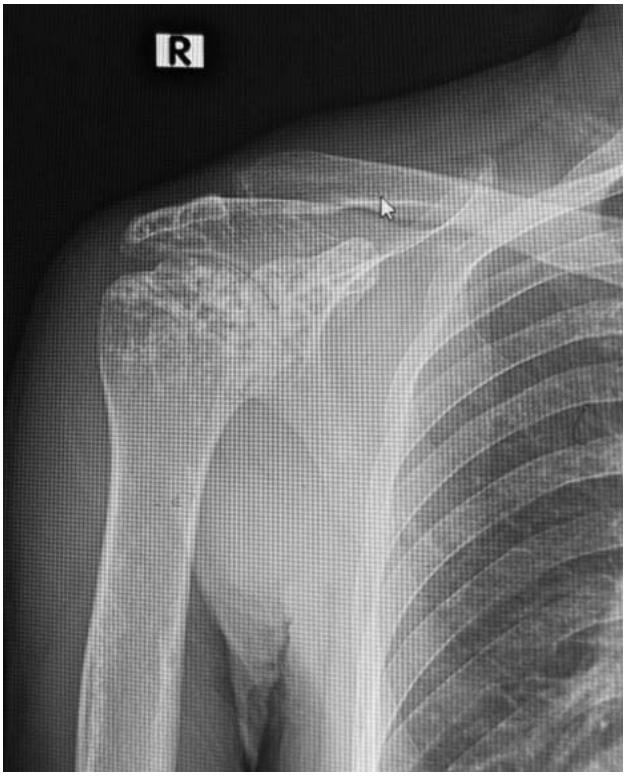

Figure 1: Radiograph of right shoulder showing sclerotic lesions in humerus head and glenoid.

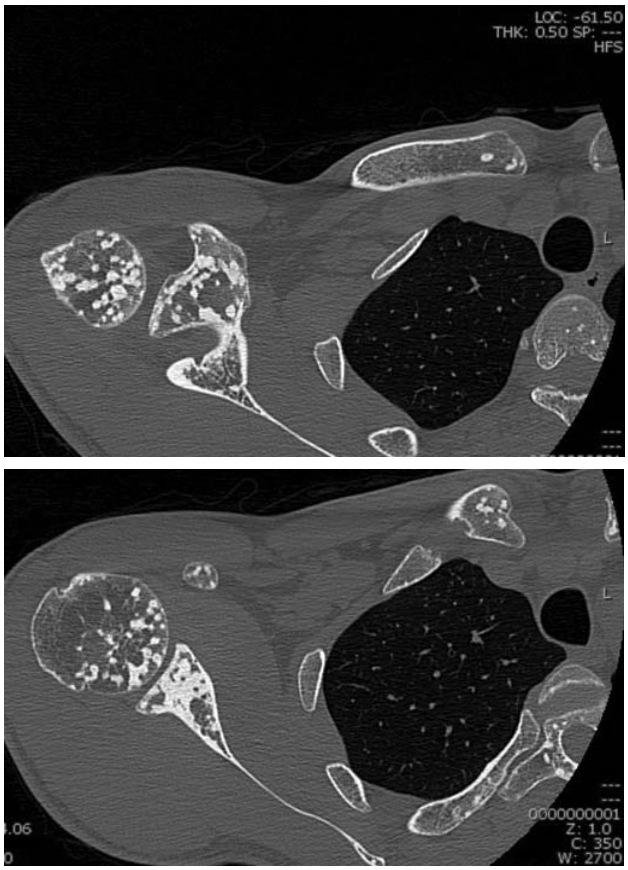

Figure 2: Axial CT scan images in bone window showing numerous sclerotic lesions in humerus, scapula, clavicle, vertebral elements and rib.

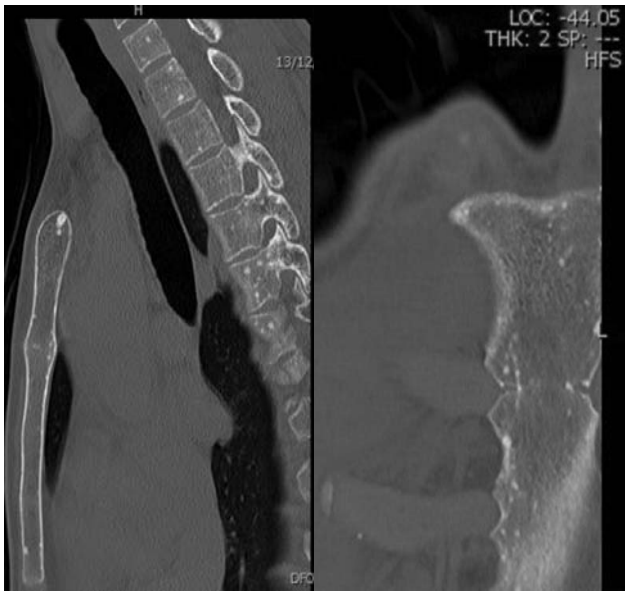

Figure 3: Reformatted CT images showing numerous sclerotic lesions in vertebral column and sternum with random distribution of the lesions and no associated periosteal reaction or extra osseous component.

Discussion

OPK Punctata is caused due to the failure of secondary spongious bone resorption. It is a rare occurrence of inherited sclerosing bone dysplasia. It consists of multiple benign bone islands that are scattered throughout the appendicular and the axial skeleton of the subject affected by this condition. Classically the bone islands arebunched around the larger joints.[10,11]

OPK Punktata starts in childhood and persists throughout life with appearance of numerous oval shaped densities of size 2 to 10 millimeters that are distributed within the epiphysis and the metaphysis of the long bones. The lesions are distinguished in a fairly symmetrical distribution on a radiograph in a wide variety of bones.These involve the cortex as well as the medullary canal. They are hyperdense on CT images and radiographs. On MRI, due to their sclerotic nature, they remain hypointense on all sequences. No associated soft tissue component seen. There is no predilection for fracture or malignancy. The bone islands are less commonly found in the skull, ribs, mandible and vertebral bodies.[12,13] Histology of these lesions reveal dense trabeculae of spongious bone, occasionally forming a nidus without communication with the bone marrow.[12,11]

Treatment is usually symptomatic depending on the presentation as these lesions inherently do not cause any symptoms, as was the case in our patient. Rarely the active lesions have been treated with bisphosphonate therapy; however, the results are debatable.

Conclusion

OPK is a radiological diagnosis and usually incidental, however, they may be associated with other associated pathologies. The main differential is still osteoblastic metastasis and a multidisciplinary team approach will ward off unnecessary investigations including a biopsy.

Conflict of Interest: No financial or institutional conflict.

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