Traumatic Fracture in a patient of Osteopoikilosis with Review of Literature

Rohan Bansal[1], Aditya C Pathak[2], Binoti Sheth[3], Atul K Patil[4]

What to Learn from this Article?

Differential Diagnosis of Osteopoikilosis?

Approach to patient of osteopoikilosis presenting with traumatic fracture?

Abstract

Introduction: Osteopoikilosis or osteopathia condensans disseminata is a rare hereditary autosomal dominant sclerosing bone dysplasia. Patients are usually asymptomatic and the diagnosis is usually made incidentally on radiographs which show presence of symmetric, multiple, well defined, small ovoid areas of increased radiodensity clustered in peri-articular osseous regions with propensity for epiphyseal and metaphyseal involvement. There are no increased risks of pathological fracture in a case of osteopoikilosis and traumatic fracture healing in a case of osteopoikilosis is similar to fracture occurring in other normal patients.

Case Report: A 34 years male, electrician came with history of accidental fall from height while working in office leading to development of pain and swelling over left lower leg and ankle diagnosed with Ruedi-Allgower classification type I pilon fracture(without fibula fracture) no distal neuro-vascular deficit. Patient was offered surgical treatment in form of open reduction and internal fixation of tibial fracture by plate osteosynthesis using antero-medial approach , showed complete union and was followed up for eight months.

Conclusion: Osteopoikilosis has a benign course and it should always be kept as a possible differential diagnosis for osteoblastic metastasis to avoid diagnositic dilemma. Diagnosis can be settled by routine x-rays (for type ,extent and site of lesions, bones affected), clinical features of patient , histopathology and other systemic or pre-existing conditions.

Keywords: Fracture , Osteopoikilosis ,union, Pilon, Osteoblastic metastasis

Introduction

Osteopoikilosis first described by H. Albers-Schonberg in 1915 is a rare autosomal dominant sclerosing dysplasia of unknown etiology with an estimated prevalence of 1:50000 [1].It is seen more commonly in males as compared to females [2,3]. It is usually asymptomatic and is a coincidental finding on radiology. However sometimes it may present with joint pain and swelling(15 % of patients), skin manifestations, rheumatic and skeletal disorders , organ anomalies and endocrine dysfunction [2,4]. Case

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Author's Photo Gallery


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Table 1 – Differential Diagnosis of Osteopoikilosis

Differential	How To Differentiate from Osteopoikilosis
Osteoblastic Bone Metastasis	Age, Bone Scan, Pre-existant Neoplasm, Metastatic disease can affect any bone, but it predominates in the axial skeleton, is rarely seen below the knee or elbow and tends not to follow a periarticular.
Tuberous Sclerosis	Symmetry, metaphyseal and epiphyseal preference, and uniform, well-defined foci are less striking than in osteopoikilosis.It is accompanied by other systemic manifestation seizures, adenoma sebaceaum , Hanartomas and Mental retardation.
Synovial Chondromatosis	Synovial chondromatosis progresses through various stages of activity and primararily affect large joints of body commonly knee. In the acute stage, the entire joint synovium is hypertrophied and hyperemic with numerous foci of cartilage formation. During the intermediate stage, the acute synovial reaction gradually subsides . Endochondral bone formation may occur but requires a blood supply and is confined to loose bodies with a pedicle or to free loose bodies that have regained a synovial attachment. In the late stages of the disease, the generalized synovial reaction reverts to normal. Secondary osteoarthritis results from the presence of multiple loose bodies within the joint.Microscopically they exhibit a periphery of fibrocartilage with underlying cancellous dead bone and a zone of calcification.
Melorheosteosis	Sclerotic lesions of cortical bones, usually in the diaphysis, that resemble “candle-wax-dripping” Cortical hyperostosis with an undulating appearance usually affecting one side of a bone Soft tissue lesions that may calcify -Adjacent to involved bone.May grow to compress nerves .Usually low signal on MRI on Enhance with Gadolinium.Bone scan is markedly positive
Mastocytosis	Symmetry, metaphyseal and epiphyseal preference, and uniform, well-defined foci are less striking than in osteopoikilosis along with systemic manifestation and respiratory and haematological involvement.
Osteopathia Striata	The sclerotic areas within the bone are neither round nor oval. Instead, they are linearly striated and periarticular in distribution. Clinical manifestations of this disorder are subtle or nonexistent.

reports showing co-existence of osteopoikilosis with rheumatoid arthritis and discoid lupus erythematosus have also been published [5]. However pathological fractures in case of osteopoikilosis have not been demonstrated. Fracture healing in a case of osteopoikilosis is similar to fracture healing in a normal patient. Patients with osteopoikilosis usually do not require any specific treatment for the condition.

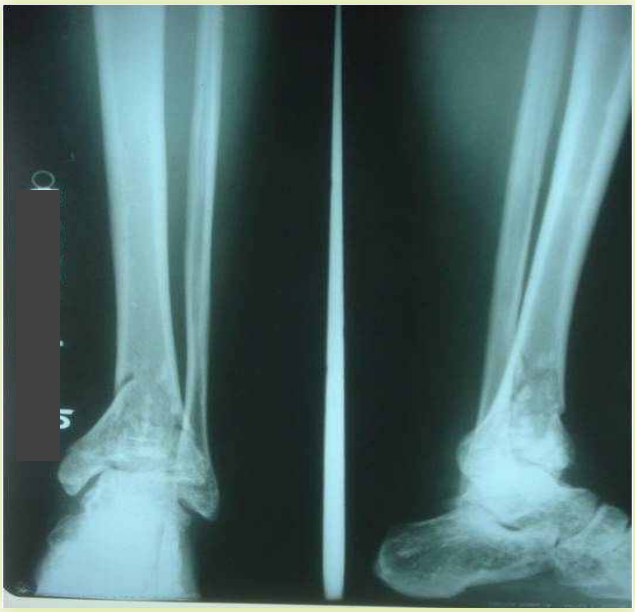

Figure 1 - Radiograph of left Tibia fibula with ankle showing type I pilon fracture. Note the well defined, multiple sclerotic lesions or bony islands in lower end of tibia and tarsal bones

Case Report

A 34 years male, electrician in came with history of accidental fall from height while working in office leading to development of pain and swelling over left lower leg and ankle .Patient was completely asymptomatic prior to the episode. Patient's general examination was normal and no abnormality was detected. On examination, patient had swelling and tenderness over lower one-third left leg with bony crepitus and painful ankle movements of left ankle with no distal neuro-vascular deficit.

Routine full length left tibia-fibula with ankle radiographs were done (Fig. 1) which showed Ruedi-Allgower classification Type I Pilon fracture (without fibula fracture). Incidental finding seen on the radiograph was well defined, multiple sclerotic lesions or bony islands in lower end of tibia and tarsal bones .Radiographs of other parts of body were done which showed presence of similar multifocal bone islands in hand phalanges, metacarpals, carpal bones, metatarsals, tarsals, lower ends of radius, lower ends of ulna and head of femur on both sides (Fig. 2). However ribs , Skull and vertebra were spared (Fig. 3). Lesions were well defined, symmetrical, epiphyseal-metaphyseal involvement of small as well as long bones of body. Patient's serum calcium, phosphates and alkaline

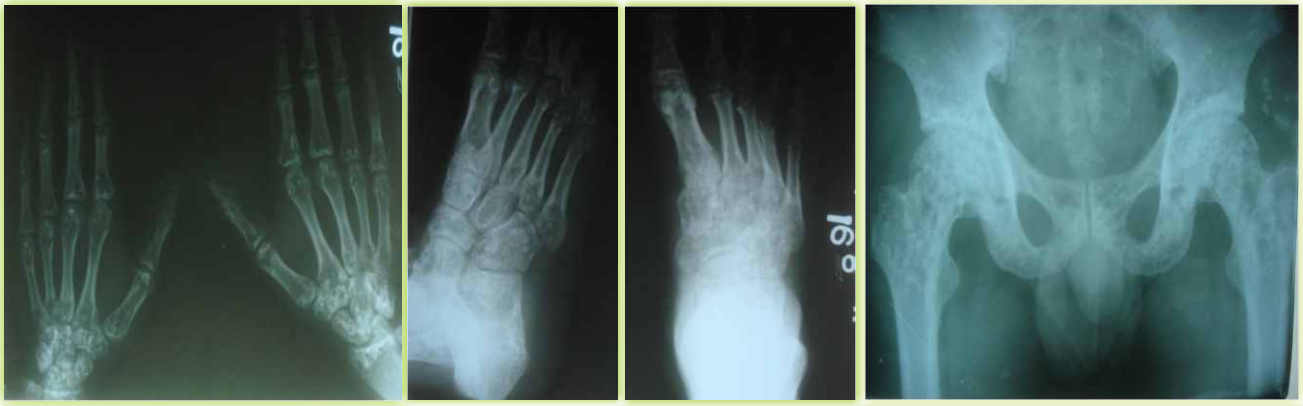

Figure 2 - Radiograph of both hands, feet and Pelvis with Hips showing well defined, multiple sclerotic lesions or bony islands similar to seen in tibia

phosphatase levels were normal. Bone scan was normal. Patient had no evidence of any systemic involvement other than skeletal system.

Patient was offered surgical treatment in form of open reduction and internal fixation of tibial fracture by plate osteosynthesis using antero-medial approach (Fig. 5). Intraoperatively patient's bone had a normal appearance and consistency as compared to a normal bone (Fig. 4). Fracture was treated with locking lower end tibial plate on medial tibial surface with 3.5 mm reconstruction plate anteriorly. Reconstruction plate was used anteriorly for fracture reduction as fracture was very communited anteriorly. Intraoperative biopsy specimen was sent which showed there were focal condensations of compact lamellar bone within the spongiosa confirming the diagnosis of osteopoikilosis.

Post-operatively patient was started ankle and knee mobilisation from third day. Patient was also started nil weight bearing on crutches immediately post operatively. Patient was started partial weight bearing walking after six weeks and full weight bearing after twelve weeks Fracture union was complete after twelve weeks(Fig 6).

Patient was walking full weight bearing after twelve weeks of surgery without any limp or pain and had normal range of motion at knee and ankle. Patient had complaints of prominent plate after eight and implant removal was done after which patient was completely normal and was able to perform all daily routine activities with normal ankle and knee range of motion.

Discussion

Osteopoikilosis (OPK) or osteopathia condensans disseminata can be transmitted as autosomal dominant or can be sporadic [6]. Loss-of-function mutations in *LEMD3* result in osteopoikilosis, Buschke-Ollendorff syndrome and melorheostosis [7].

In osteopoikilosis, osteosclerotic dysplasia of bones develops during childhood and persists throughout life. It occurs as a result of impairment of secondary spongiosa resorption and remodeling. Patients with osteopoikilosis are usually asymptomatic and diagnosis is usually an incidental finding when patient is being investigated for some other reason. However sometimes it may present with joint pain and swelling(15 % of patients), skin manifestations (dermatofibrosis lenticularis disseminata, keloid formation, plantar and palmar keratomas), rheumatic and skeletal disorders (arthritis, exostoses, osteitis condensans ilii, melorheostosis, spinal stenosis,

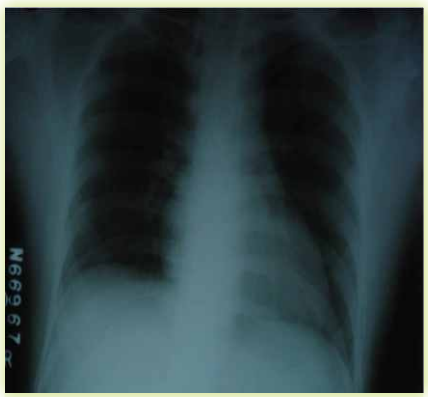

Figure 3 - Chest radiograph with bones devoid of any sclerotic lesion

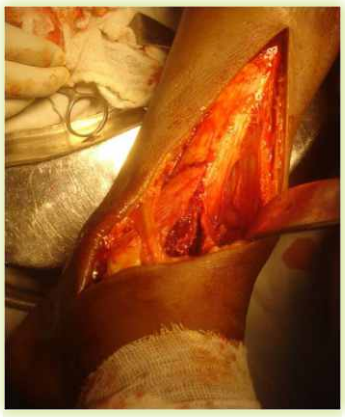

Figure 4 - Intraoperative radiograph of fracture with normal appearing bone

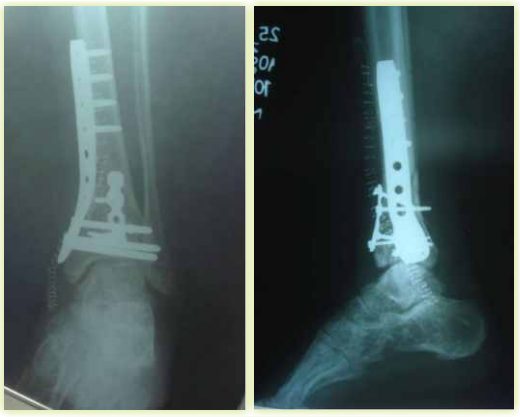

Figure 5 - Post operative Mortise view with internal fixation done

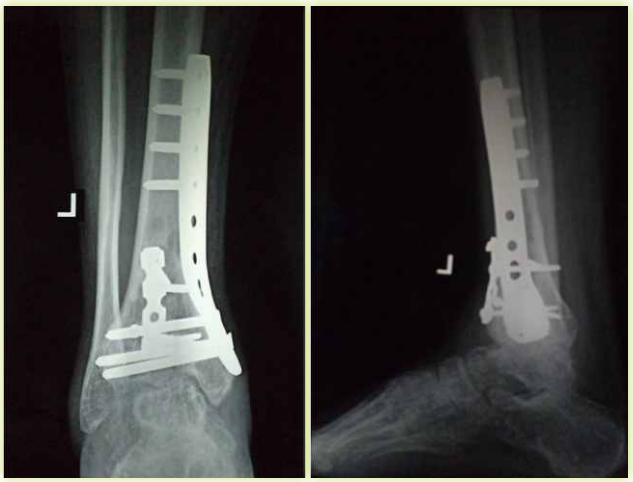

Figure 6a - Antero-posterior view after fracture union
Figure 16b - Lateral view after fracture union

chondrosarcoma, osteosarcoma), organ anomalies (aorta coarctation, double ureter, growth abnormalities, hare lip, dental abnormalities) and endocrine dysfunction (diabetes mellitus) [1,2,3,8] .Case reports demonstrating co-existence of osteopoikilosis with rheumatoid arthritis, discoid lupus erythematosus, reactive arthritis , polyarthralgia and familial Mediterranean fever have also been published. Presence of connective tissue nevi called dermatofibrosis lenticulari disseminata with osteopoikilosis comprise Buschke-Ollendorff syndrome [2,3].

Radiographs show presence of multiple, small, well-defined, variably shaped and widely symmetrically distributed sclerotic areas all over the skeleton with propensity for epiphyseal and metaphyseal involvement and predilection for phalanges(100%), carpal bones(97.4%), metacarpals (92.5%), foot phalanges(87.2%), tarsal bones(84.6%), metatarsals(84.2%), 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%) according to literature with characteristic sparing of skull, ribs and vertebrae [1]. Patients have normal hematological investigations. Patients have a normal bone scan with normal serum calcium, phosphates and alkaline phosphatase levels [8]. Histologically, bone islands consisting of mature bone with thickened trabeculae seen within spongy bone. At the periphery of the lesion, the lesional trabeculae merge with the normal bone and there is no sclerotic rim. Occasionally, woven bone is a minor part of the lesion The condensations of cancellous bone consist of a peripheral area of trabeculae in which osteocytes are scant, and there are no osteoblasts or osteoclasts, together with a central core of irregular trabeculae in which both osteoblasts and osteoclasts are present. The lesions appear to be metabolically active; in the immature skeleton they become denser with growth, but later they may change their sue or even disappear altogether. The precise origin of these abnormal areas remain debatable, but they appear to represent foci of deranged differentiation in cancellous bone [1].

Differential diagnosis includes osteoblastic bone metastasis, melorheostosis, osteopathia striata, mastocytosis, tuberous sclerosis and synovial chondromatosis (Table 1). However characteristic radiological appearance of bony lesions with a normal bone scan clinches diagnosis of osteopoikilosis. Typically patients are asymptomatic, although as many as 20% may have mild articular pain and effusion. Clinically osteopoikilosis must be distinguished from more severe dysplasias, such as tuberous sclerosis, mastocytosis, and, most importantly, osteoblastic metastatic lesions. Typically, findings of bone scans are normal in osteopoikilosis, and this feature has sometimes been used to differentiate it from metastatic bone disease. A radionuclide bone scan is essential in distinguishing OPK from primary bone tumors or osteoblastic bone metastases. Bone scan findings are usually normal in patients with OPK, but reveal slightly increased activity similar to the bone island or enostosis that reflects active osseous remodeling that may be detected, especially in young patients with classic radiographic findings that are consistent with OPK. An abnormal scan finding in an older patient should, however, be thoroughly investigated. A malignant transformation is also conceivable if the cellular activity exists in the foci of OPK [9,10]. A symmetric distribution of the lesions has been observed in osteopoikilosis, although this has not been statistically analyzed. The radiopaque areas can increase or decrease in size and number, or even disappear [11]. Such a feature is not seen in osteoblastic metastasis.

Patients with osteopoikilosis do not have an increased incidence of pathological fractures [12] and fracture healing in a patient of osteopoikilosis is similar to fracture healing in a normal patient. Time taken for fracture union in a case of osteopoikilosis is also comparable to that taken for fracture union in a normal patient [12]. Patients with osteopoikilosis lead a normal asymptomatic life and can perform all daily routine activities. No specific treatment for osteopoikilosis is required. In conclusion although osteopoikilosis has a benign course, it should always be kept as a possible differential diagnosis for osteoblastic metastasis to avoid misdiagnosis and avoid unnecessary

costly investigations.

Conclusion

Patients with osteopoikilosis lead a normal asymptomatic life and can perform all daily routine activities. No specific treatment for osteopoikilosis is required. In conclusion although osteopoikilosis has a benign course, it should always be kept as a possible differential diagnosis for osteoblastic metastasis and diagnosis dilemma can be settled by routine x-rays (for type ,extent and site of lesions, bones affected) Clinical features of patient, Histopathology and other systemic or pre-existing conditions.

Clinical Message

Although the natural course of this condition is benign and requires no treatment, the complications and coexisting pathologic conditions require medical attention. Therefore, it is important that an accurate diagnosis be made.

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Author's Corner

I and Dr Aditya were in Final year of MS orthopaedics and had never seen such lesions in our previous orthopaedic career. We were surprised to see the xrays, but as we showed the xray to our seniors Dr Binoti Mam and Dr Atul sir, they then guided us to diagnosis of Osteopoikilosis. It was an intresting case and we had followed it up with great curiosity and developed our naïve learning curve in orthopaedic - Dr Kunal Bansal

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