

Images in clinical medicine



A rare genetic disorder of osteopoikilosis

 Madhavi Kandarkar,  Deepali Patil

Corresponding author: Madhavi Kandarkar, Ravi Nair Physiotherapy College, Datta Meghe Institute of Medical Sciences, Sawangi Meghe, Wardha, Maharashtra, India. madhavikandarkar2295@gmail.com

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A rare genetic disorder of osteopoikilosis

Madhavi Kandarkar^{1,&}, Deepali Patil²

¹Ravi Nair Physiotherapy College, Datta Meghe Institute of Medical Sciences, Sawangi Meghe, Wardha, Maharashtra, India, ²Department of Musculoskeletal Physiotherapy, Ravi Nair Physiotherapy College, Datta Meghe Institute of Medical Sciences, Sawangi Meghe, Wardha, Maharashtra, India

&Corresponding author

Madhavi Kandarkar, Ravi Nair Physiotherapy College, Datta Meghe Institute of Medical Sciences, Sawangi Meghe, Wardha, Maharashtra, India

Image in medicine

A 30-year-old male patient admitted to a hospital experienced pain in the left knee for 10 years and right knee for 1 month which increased strenuous activity, with a history of COVID-19 1 year back. Multiple areas of sclerosis were seen on epiphysis metaphyseal area in the X-rays of the bilateral knee and the hip joints (A, B). In the X-ray of the vertebra, lipping of vertebrae was seen (C). Sclerotic lesions were seen in (A, B, C, D). The clinical signs and radiological features, sclerotic lesions, and low bone mineral density levels, all led to the diagnosis of a rare genetic case of osteopoikilosis (A, B, C, D). The patient began treatment with regular intake of calcium; 1000 milligrams per day, adequate intake of vitamin D; 800 milligrams per day, and 50 milligrams of intravenous methylprednisolone, with mild to

moderate level of physiotherapy rehabilitation program. It is a rare disease of the bones, affecting 1 in 50,000, equally in both sexes, autosomal dominant, with all patients having LEMD3 gene mutation. It is usually an incidental finding, not

associated with symptoms, but pain, joint stiffness, and joint effusion seen in 20 percent of patients.

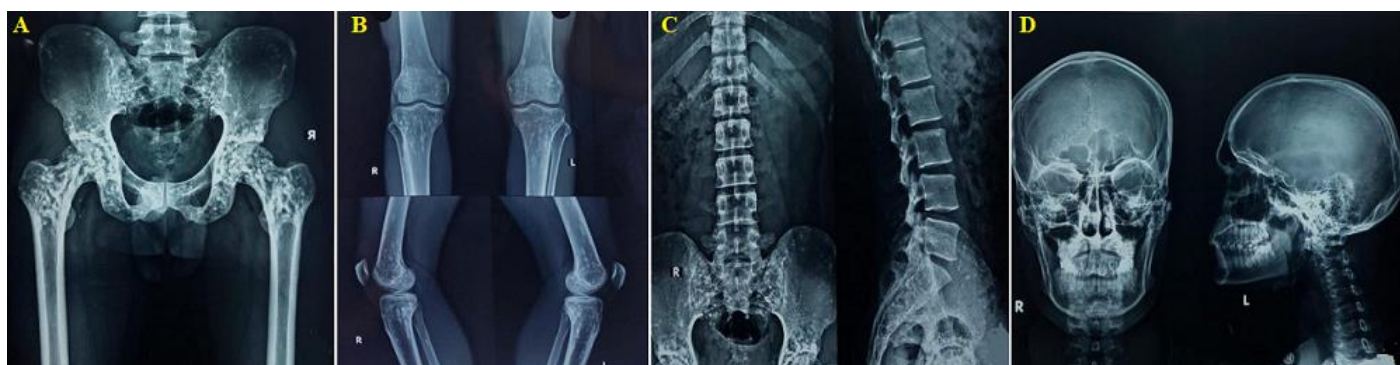


Figure 1: AP and lateral view X-rays; A) X-ray of skull showing sclerotic lesions, erosions and calcific formation; B) X-ray of knee joint showing the sclerotic lesions and lipping of vertebrae; C) X-ray of the lumbar spine demonstrating chondromalacia patella in the knee; D) X-ray of hip showing numerous sclerotic bone islands surrounding the hip joint