A case report of a symptomatic osteopoikilosis patient caused by novel mutation in LEMD3

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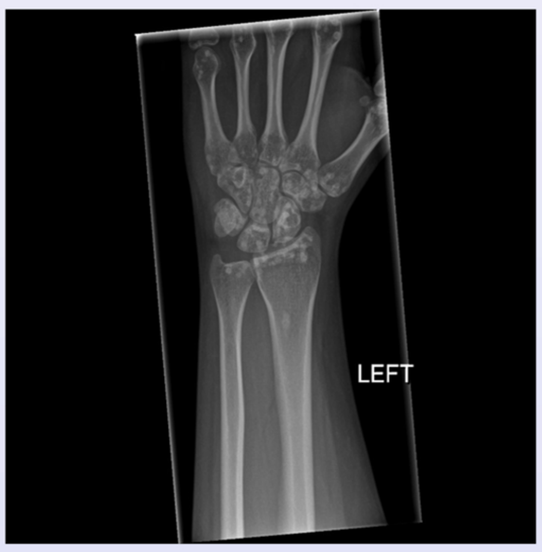
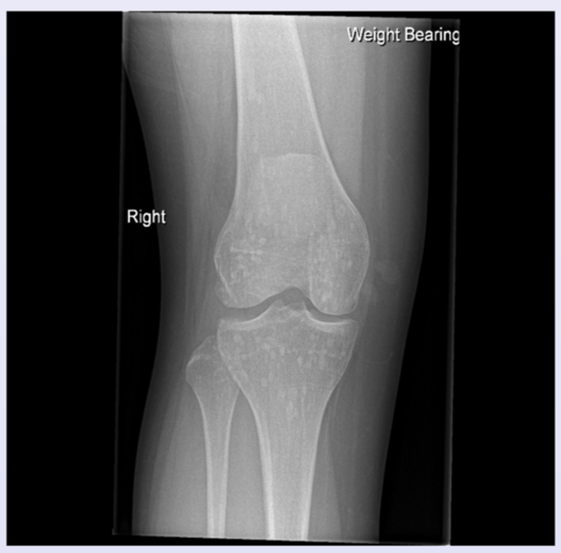
Case History

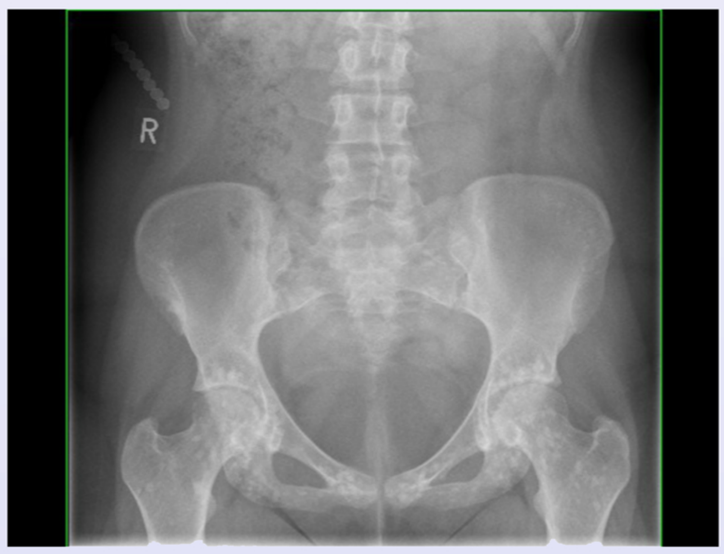
- 17 years lady presented with a fall onto her left knee, following which she continued to have severe persistent pain and occasionally her knee gave away with intermittent "locking".
- A referral to metabolic bone disease clinic was made as the left knee X-ray showed discreet spherical areas of increased radiological density with normal CT and MRI scans of the left knee.
- Physical examination was unremarkable.
- Her brother was under investigation for lumps under skin.
- Morphology of the spots noticed on X-ray was identical to those seen in osteopoikilosis.

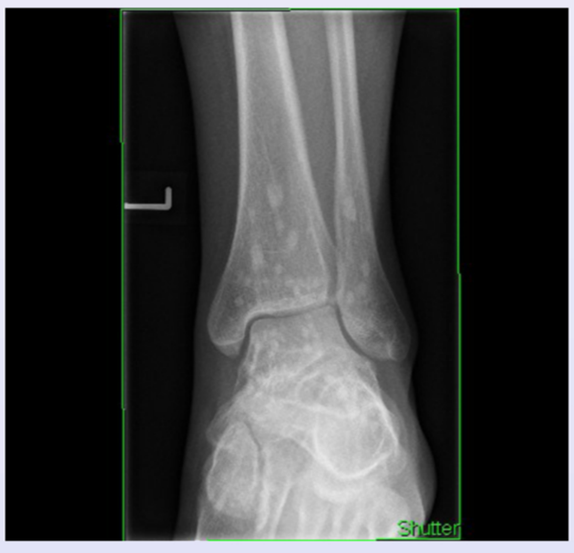
Further Management

- The relative clinical and laboratory tests were negative for any type of arthritis, infection or osteoblastic bone metastases which were in the differential diagnosis.
- Bone densitometry was normal.
- Skeletal survey revealed extensive changes representing osteopoikilosis throughout the skeleton involving the hands, feet, humerus, ulna and radius, femur, tibia and fibula, pelvis and sacrum.
- LEMD3 heterozygous gene mutation was positive.
- Subsequently she developed a lump over the lateral aspect of right scapula which increased in size and causing discomfort particularly at night.
- MRI and CT scans of the right scapula were unremarkable apart from a bony mass.
- Surgical removal of the bone lump (Although the risk of malignant transformation of osteopoikilosis is rare) confirmed to be osteochondroma.
- Her brother has been diagnosed to be suffering from Buschke Ollendorff syndrome, which can be associated with both osteopoikilosis and melorheostosis.









Osteopoikilosis

- Spotted bone disease.
- First described by Albers-Schonberg in 1915,the estimated incidence is 1 in 50,000.
- Benign, rare, autosomal dominant sclerosing dysplasia of bone characterized by the presence of numerous white densities bone islands in the skeleton.
- Associated with a heterozygous mutation in the LEMD3 gene.
- Differential considerations for this condition include osteoblastic metastases, tuberous sclerosis,mastocytosis and synovial chondromatosis.
- Pain is not a prominent feature of OPK, but in some patients (15-20%), pain could be a presenting symptom of the disorder.

Conclusion

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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