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Educational Abstract

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Brittle Bone Brothers: Osteogenesis Imperfecta Conventional Imaging Case Series

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Learning objectives

To characterize Osteogenesis Imperfecta (OI) in adults through skeletal conventional imaging

Background

Osteogenesis Imperfecta (OI) or "brittle bone disease", is a genetic disorder characterised by increased bone fragility and decreased bone density due to abnormalities of type I collagen. The diversity of OI clinical expression depends on its classification and age. One OI type may lead to death, alongside multiple bone fractures, osteoporosis, short stature and adult-onset hearing loss. Type I OI were reported to survive up until adulthood. There are many cases found in babies but these 3 brothers' cases were the only OI adult case in our hospital.

Findings & Procedure details

A skeletal conventional imaging was performed to the 3 brothers and all of them has identical results such as bowing deformities of long bones, old union and some non-union fractures with extreme angulation and severe osteoporosis. The 34 year-old presented with bowing of his ribcage and all extremities with some bone destruction of both humeral bones and several old fractures. The 36 year-old presented with bowing deformity of all long bones. The 39 year-old has many current fractures and several old ones which were non-union fractures. There were no cardiopulmonary abnormalities observed in these 3 brothers.

Conclusion

These 3 brittle brothers with the same type of OI can give awareness on how hereditary this disease can be. Conventional x-ray plays a role as the imaging modality needed to diagnose OI.