

An adolescent girl with infantile osteopetrosis presenting with a proximal femur fracture

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Infantile osteopetrosis is a rare autosomal recessive genetic disorder that is characterised by abnormal osteoclast activity leading to increased bone density [1]. It usually presents during infancy and the survival beyond the first decade of life is rare [2]. A 15-year-old girl fell out of bed and presented to the emergency department with pain and deformity of right thigh. She was a child of consanguineous parents and was diagnosed with osteopetrosis during infancy based on the classical radiological changes (diffuse sclerotic bone changes) and clinical features [3]. She had multiple complications such as severe global developmental delay, growth retardation (less than 3rd percentile), seizure disorder, bilateral pseudoproptosis with visual impairment, bilateral hearing impairment, obstructive sleep apnoea, hepatosplenomegaly, anaemia and thrombocytopenia. She did not have previous fractures. She was haemodynamically stable on admission. X-rays showed a diffuse dense skeleton (Figure 1A) with a rugger-jersey spine (Figure 1C) and a right proximal femur fracture (Figure 1B-Arrow). The distal pulses were felt with an oxygen saturation of 98%. She was managed non-operatively with skin traction and analgesics and was later lost to follow up. Although this condition is well described, it is most unusual for infantile-onset osteopetrosis of such severe degree to survive up to adolescence and such patients invariably have multiple complications. We opted to manage the fracture non-operatively as internal fixation is extremely challenging in such patients.

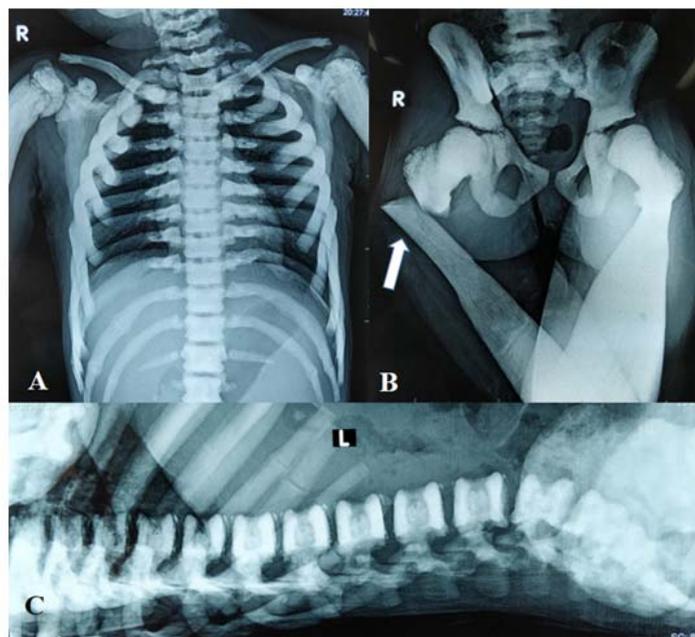


Figure 1. X-rays showing a diffuse dense skeleton (Figure 1A) with a rugger-jersey spine (Figure 1C) and a right proximal femur fracture (Figure 1B-Arrow).

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