A challenging paediatric pathological femur fracture in pyknodysostosis (osteopetrosis acro-osteolytica): lessons learnt

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DESCRIPTION

Pyknodysostosis, also known as *osteopetrosis acro-osteolytica*, is a rare autosomal-recessive bone dysplasia characterised by osteosclerosis and short stature. It involves mutations in the gene that encodes cathepsin K, a lysosomal metalloproteinase highly expressed in osteoclasts and important for bone matrix degradation.[1] Clinical features also include kyphoscoliosis and deformities of the chest; high-arched palate leading to severe upper airway obstruction;[2] and dysmorphic facial features. Radiographs demonstrate a generalised increase in bone density. Histological evaluation shows attenuated Haversian canal systems with decreased osteoblastic and osteoclastic activities. Serum chemistries are normal.[3]

We present a case of a 12-year-old girl who presented with a diaphyseal femur fracture following a fall off a trampoline. She is known to have *pyknodysostosis* (figure 1) with severe obstructive sleep apnoea with overnight continuous positive airway pressure support. This was an isolated closed injury with no neurovascular deficit. Non-operative management is seldom appropriate for such injuries as it involves an unacceptably high rate of complications due to long periods of immobilisation. While surgical fixation offers early mobilisation and weight-bearing, it proves challenging in conditions associated with forms of osteosclerosis due to dense brittle bone.[3]

We performed closed reduction and internal fixation using a submuscular plating technique (precontoured 16-hole 3.5 mm locking-plate) with uneventful recovery. Drilling for the screws proved most challenging; we did, however, overcome this by using one drill-bit per hole and constant irrigation with cold saline to reduce the effects of heat generated. Despite these precautions, iatrogenic breakage of drilling instruments is a likely complication (figure 2).

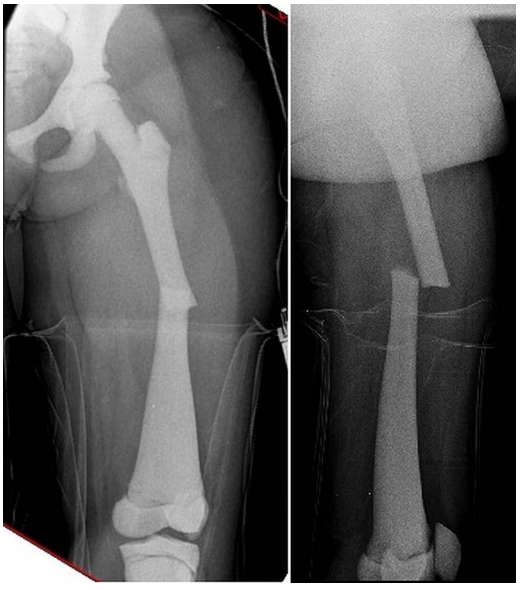

Figure 1 Anteroposterior and lateral radiographs of left femur with a disphyseal transverse fracture.

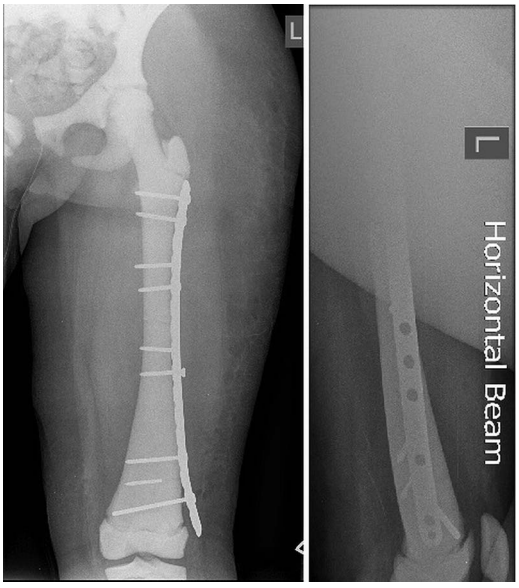

Figure 2 Anteroposterior and lateral radiographs of left femur following submuscular plate fixation.

Learning points

- Formal preoperative anaesthetic assessment is essential as patients with *pyknodysostosis* often have an obtuse angle of the mandible, high-arched palate and upper airway obstruction.
- Preoperative surgical planning should include ensuring the availability of multiple drill-bits and being prepared for iatrogenic metalwork breakage.
- Intraoperatively, using one drill-bit per hole with constant cold saline irrigation is a useful technique.

Competing interests None.

Patient consent Obtained.

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REFERENCES

1 Arman A, Bereket A, Coker A, *et al*. Cathepsin K analysis in a pycnodysostosis cohort: demographic, genotypic and phenotypic features. *Orphanet J Rare Dis* 2014;26:60.
2 Kiran S, Goel M, Singhal P, *et al*. Pyknodysostosis: anaesthetic considerations—a case report. *Internet J Anesthesiol* 2009;21:13.
3 Arkader A, Dormans JP. Pathological fractures associated with tumours and unique conditions of the musculoskeletal system. In: Beaty JH, Kasser JK, eds. *Rockwood and Wilkins' fractures in children*. 7th edn. Philadelphia, PA: Lippincott, Williams & Wilkins, 2010:120–91.