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Case Report

Osteopetrosis – a challenge for the orthopaedic surgeon

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Summary

Osteopetrosis (OP) is a rare heterogeneous group of inherited skeletal dysplasias characterised by osteoclast dysfunction, impaired bone resorption and poor bone remodelling. Three groups can be categorised on the basis of clinical findings. These include neurological symptoms, haematological abnormalities and renal tubular acidosis in the first group. Increased bone density, osteomyelitis and frequent fractures are the clinical findings in the second group, and the third group have normal life expectancy but may develop cranial nerve compression and osteomyelitis. Fractures in patients with OP are common and require appropriate pre-, peri- and postoperative management. The long bones are most frequently affected, fractures of the femoral neck and proximal (upper third) shaft being particularly common. This case report proposes possible operative fracture treatment in a patient with OP and highlights the potential perioperative pitfalls in this rare surgical population.

Osteopetrosis (OP) is a rare heterogeneous group of inherited skeletal dysplasias characterised by osteoclast dysfunction, impaired bone resorption and poor bone remodelling. Three groups can be categorised on the basis of clinical findings and by genetic investigations. The autosomal recessive malignant type of OP has an incidence rate of 1/200 000 births and frequently leads to death in early childhood. Patients suffer from neurological symptoms, haematological abnormalities and renal tubular acidosis.[1] The intermediate type has an autosomal recessive mode of inheritance with a less aggressive phenotype. Similar to the 'malignant' type, it is diagnosed in infancy, and the clinical symptoms include increased bone density, osteomyelitis and frequent fractures. It accounts for approximately 15% of cases of OP.[1] The third group, with an incidence rate of 1/20 000 - 500 000 births and autosomal dominant inheritance, is divided into type I and type II OP. These patients have normal life expectancy, but may develop cranial nerve compression and osteomyelitis.

Radiographic examinations often lead to the diagnosis of sclerosis in the cranial vault (type I) or painful 'endobone' formations in the pelvis and vertebral endplates (type II).[2,3] Fractures in OP patients are common and require appropriate pre-, peri- and postoperative management. The long bones are most frequently affected, fractures of the femoral neck and proximal (upper third) shaft being most commonly reported.[4] Several authors have indicated that the proximal tibia is highly susceptible to fracture.[5,6] Furthermore, a tendency to poor healing of bone and soft tissue related to the abnormal bone metabolism has been observed.[4]

This case report proposes possible operative fracture treatment in an OP patient and highlights the potential perioperative pitfalls in this rare surgical population.

Case report

A 37-year-old white man sustained a high-speed head-on motor vehicle accident and remained trapped in his car for 90 minutes before rescue. At the scene he was conscious with a Glasgow Coma Score of 15 and haemodynamically stable. After primary stabilisation he was transferred to a regional level 1 trauma centre and treated according to Advanced Trauma Life Support guidelines. Primary diagnostics revealed comminuted and displaced right-sided fractures of the proximal third femoral shaft (Fig. 1), humeral shaft (Fig. 2) and forearm (first-degree open) (Fig. 3) as well as a chest contusion with lateral rib fractures. Radiographs revealed an increased bone density and an abnormal narrow medullary cavity in all examined bones.

The medical history revealed that the patient had autosomal dominant (type II) OP. The disease had first been recognised in early childhood after a femoral fracture; apart from this the patient had been asymptomatic. At least six family members had been affected by either the recessive or the dominant type of OP within the last three generations. The patient's mother and two of her siblings were diagnosed as inactive carriers of the autosomal dominant

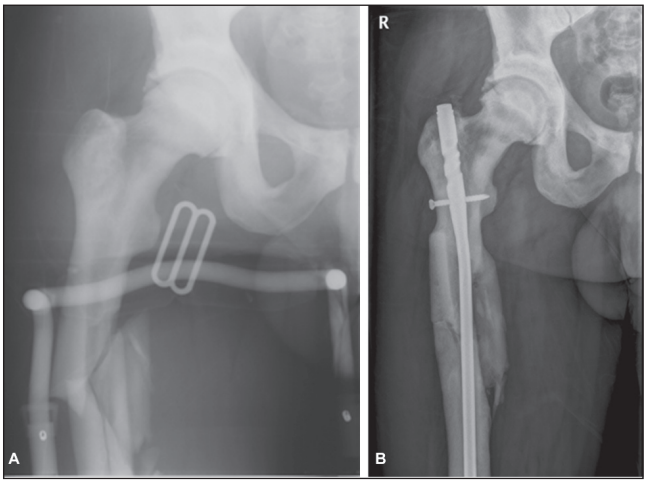

Fig. 1. Right femur: A – pre-operative; B – 6 months postoperative.

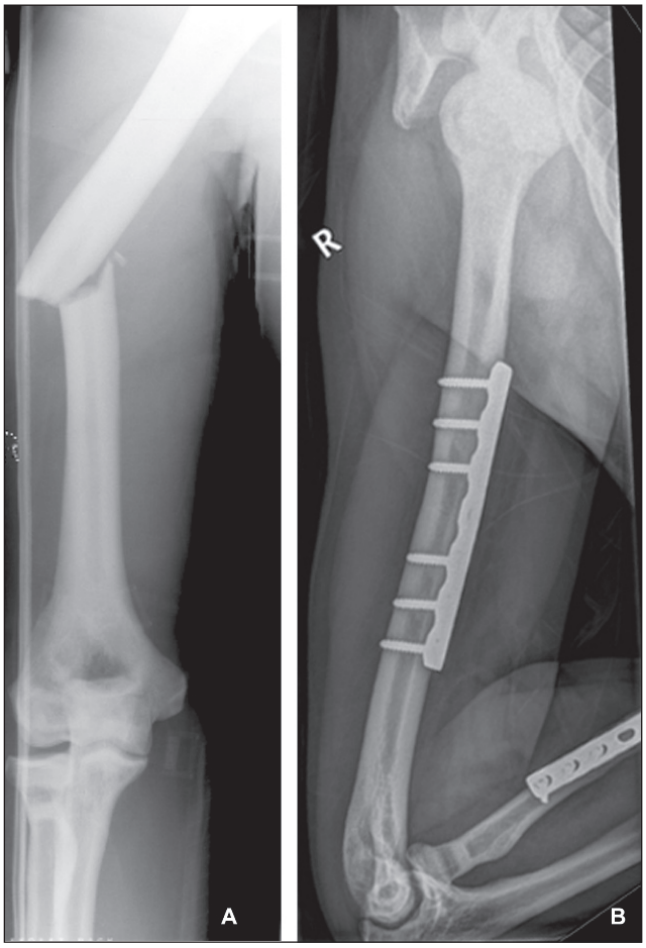

Fig. 2. Right humerus: A – pre-operative; B – 6 months postoperative.

OP. Pre-operatively, severe haematological disturbances and hepatosplenomegaly were excluded. The patient's general condition allowed us to continue with the necessary surgical procedures.

The first-degree open forearm fracture had been washed out and covered with a sterile dressing in the emergency department. Subsequently the comminuted proximal femoral fracture was nailed, which was extremely challenging. The hard osteopetrotic bone caused several drill-bits to wear

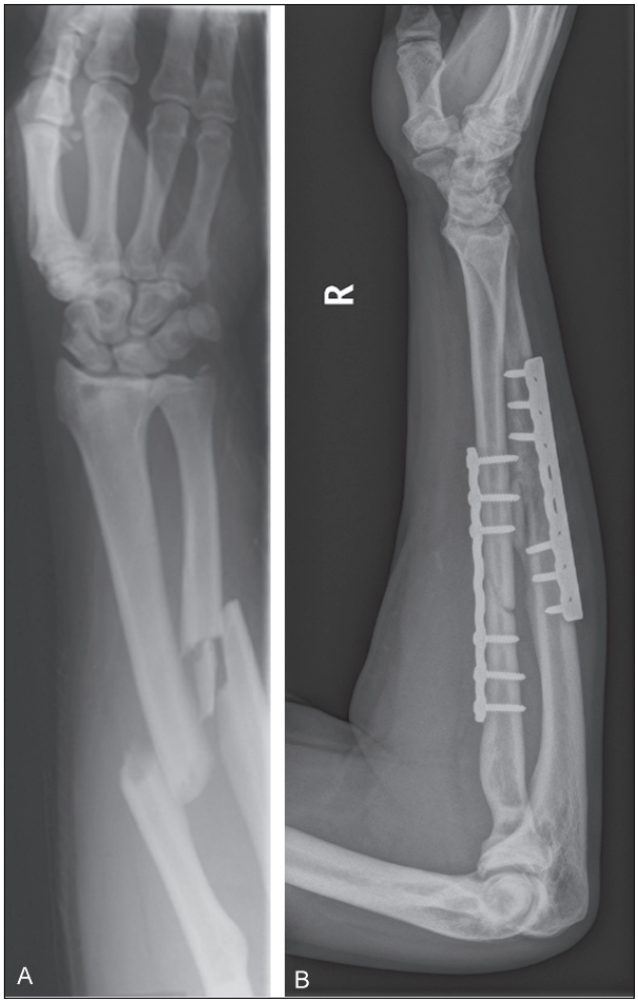

Fig. 3. Right forearm: A – pre-operative; B – 6 months postoperative.

out before a drill duct for a 9 mm intramedullary nail was achieved (Fig. 1). The humeral fracture was open reduced and fixed with a 4.5 mm plate through an anterior approach (Fig. 2). The mildly comminuted and first-degree open proximal fracture of the radius was treated with an eight-hole 3.5 mm plate after debridement and a second wash-out, and the wound was left partially open to prevent compartment syndrome. Soft-tissue swelling prevented primary treatment of the right ulnar fracture. Open reduction internal fixation with an eight-hole 3.5 mm plate was performed 8 days later, with secondary closure of the forearm wound (Fig. 3). The soft tissue healed without complications and the patient was discharged on postoperative day 17 and underwent 30 days of rehabilitation. Six months postoperatively clinical follow-up revealed good healing, with a full and pain-free range of movement of the elbow joint and symptom-free full weight bearing with appropriate function of the lower limb. Radiological examination of all treated fractures showed good bone alignment and completed bony union.

Discussion

Femoral fractures in young patients are generally the results of high-energy trauma and are frequently associated with multiple injuries. The management of a multiple-trauma patient with several fractures in combination with OP represents a major challenge given the unique bone composi-

tion and the possibility of haematological co-morbidities. Although all patients with OP have dense, fragile bones and obliteration of the marrow cavity, other manifestations of the disease vary. Haematological abnormalities such as anaemia and thrombocytopenia are common in OP and require perioperative correction, and collaboration with a haematologist may be advisable.

Fractures are common in OP and indeed are one of the classic features of the condition; however, there are currently no consensus guidelines for the fracture management in these patients. Open treatment with fixation can be time-consuming and technically extremely demanding, so non-operative treatment should always be considered. In this multiple-trauma patient fracture fixation was advantageous in further stabilising his general condition and facilitating rehabilitation. The comminuted displaced femoral fracture, the open forearm fracture and the short oblique mildly comminuted humeral shaft fracture would have posed a high risk of complications had non-operative treatment been attempted.

Additional considerations in OP are intraoperative failure of instruments and the prevention of iatrogenic fractures, to which these patients are extremely susceptible.

To date, operative treatment options for osteopetrotic fractures have seldom been described and no gold standard procedure is recognised. Chhabra et al. described a case series of nine proximal femoral fractures in 3 patients with autosomal dominant OP.[7] They were treated with an intramedullary nail or with a plate. The intramedullary fixed group had a better outcome than the other group. Plate fixation was very difficult to achieve because of the bone structure and the risk of further fracture. Yang et al. described the management of a subtrochanteric femoral fracture that was initially nailed (with a Jewett nail) but later failed because of the 'marble-like fragile bone'.[8] Gupta and Gupta presented their experience of seven femoral fractures in 5 OP patients treated with either plates or intramedullary nails and 1 conservatively treated patient.[9] The brittleness and hardness of the osteopetrotic bone, the bony obliteration of the marrow cavity and the risk of iatrogenic fracture require patience and caution during drilling and reaming. This in turn leads to prolonged operating time, which can be an issue in multiple-trauma patients.

Further considerations when orthopaedic procedures are done in a patient with OP are the intense heat and thermolability of the hard and soft tissue caused by a compressed-air drill-bit. In our patient an intramedullary device was chosen. The fracture pattern would not have allowed open reduction internal fixation in an anatomical way. Biological bridge plating would have required a long plate and would have caused further considerable additional soft-tissue damage, probably with prolonged non-weight-bearing postoperatively. Defining the nail entry point and opening the almost non-existent intramedullary canal was extremely difficult. It was mainly done using a manual drill bit, which provided better control and reduced the wear-out rate. Several authors describe internal plate fixation as a viable treatment option for femoral shaft fracture in OP. However, it is extremely difficult to achieve a sufficient duct and the drilled holes have a high potential to refracture. There is a paucity of literature on fracture of the humerus, ulna and radius. In our case, both the humeral and forearm fractures were open reduced and fixed with plate osteosynthesis. Nailing of the humeral shaft fracture would have been extremely difficult and time-consuming.

A high degree of caution and patience was exercised during each surgical intervention and maintained for the full duration of the postoperative period because we were aware that our patient might have a tendency to poor healing and an increased risk of refracture, both of which are common in autosomal dominant OP.

Conclusion

Preoperative planning minimises the duration of the operation and the risk of intra-operative complications. The orthopaedic surgeon must consider the brittleness and hardness of the bone during fixation. The longer time to union and the tendency to decreased soft-tissue healing in OP demand intensive postoperative care.

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