



## Dealing with sub-trochanteric fracture in a child with osteopetrosis : A case report

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Osteopetrosis is a rare hereditary condition which may have autosomal recessive or autosomal dominant inheritance. Patients tend to present most commonly with fractures but involvement of cranial nerves and hematopoietic system is not uncommon. Patients with infantile and intermediate type tend to present more often with problems other than orthopaedic problems. While diagnosis can be made on the basis of radiographs, management needs to be customized for every patient. Non operative and operative management both have their advantages and disadvantages. We are here reporting a case of sub-trochanteric fracture in an eight-year-old child which was managed successfully with a dynamic hip screw (DHS). Surgery could be performed successfully by taking precautions during reduction, drilling and screw placement. At the latest follow up, which was after one and half years of surgery, the fracture had united well and the child faced no limitations of activities. Thus, open reduction and fixation with DHS can be considered as an effective management modality for pediatric sub-trochanteric fractures in osteopetrosis.

**Keywords :** Osteopetrosis ; sub-trochanteric fracture ; DHS; osteoclasts.

### INTRODUCTION

Osteopetrosis is a group of dysplastic bone disorder which is characterised by diminished osteo-

clastic function and hence a diminished bone remodelling potential (3). Although this condition was first described in 1904 by Schonberg, advances in the field of genetics have improved understanding of this condition (3,11). That this condition is rare can be gauged by the fact that a little more than 300 cases have been reported till date (3). From a genetic point of view, osteopetrosis results from mutations affecting proton pump, chloride channel or carbonic anhydrase II (9). These mutations lead to a defect in the acidification of bone, which affect the osteoclastic function of bone resorption (11). Inadequate osteoclastic function in presence of normal osteoblastic function results in thick and disorganized bones. Lack of adequate remodelling leads to formation of weak bones, which can fracture easily. Other problems like osteoarthritis, spondylolysis and osteomyelitis are also seen in these patients. Formation of

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*No benefits or funds were received in support of this study. The authors report no conflict of interests.*

Acta Orthopædica Belgica, Vol. 82 - 4 - 2016

excessive bone may obliterate cranial nerve foramina and medullary cavities, which may in turn lead to neurologic and hematologic problems (5). Osteopetrosis has been classified into three types: malignant or infantile, intermediate and benign (9). Infantile or malignant type is frequently associated with death in the first decade. In less severe forms like autosomal dominant osteopetrosis, patients have a normal life expectancy, but the bones are brittle and fracture frequently (1,3,9).

The healing and remodelling of fractures in osteopetrosis is unpredictable. As the healing response is variable, management must be individualised. The decisions to manage operatively or non-operatively, when to mobilise and allow normal day-to-day activities have to be taken on a case-by-case basis, as no fixed guidelines are available for management. An informed decision should be made about management of fracture; the surgeon should always have a backup plan available in case the original option fails.

We are here reporting a sub-trochanteric fracture in an 8 year old child, which was managed with a dynamic hip screw (DHS). It went on to unite uneventfully and at the latest follow up one and half years after the surgery the child had no limitations of activity.

### CASE REPORT

An eight-year-old male child presented to the surgical emergency with a history of fall from bicycle on which he was a pillion rider. The child was having marked pain and swelling on the left proximal thigh and was unable to move the limb. Suspecting a fracture of femur, the lower limb splinted and plain radiographs obtained, which showed that the whole skeleton had an increased density than normal. The bones were chalky white in appearance and medullary canal was not identifiable. There was a sub-trochanteric fracture located just distal to the lesser trochanter. The fracture line was transverse (Fig. 1), which is not a common pattern in sub-trochanteric fractures in individuals with normal bones. The distal femur especially the distal metaphyseal area was found to be broadened (Fig. 2). We performed a skeletal survey and similar dense, chalky white



*Fig. 1.* — Radiograph showing the subtrochanteric fracture with a gross displacement of the fractured fragments.

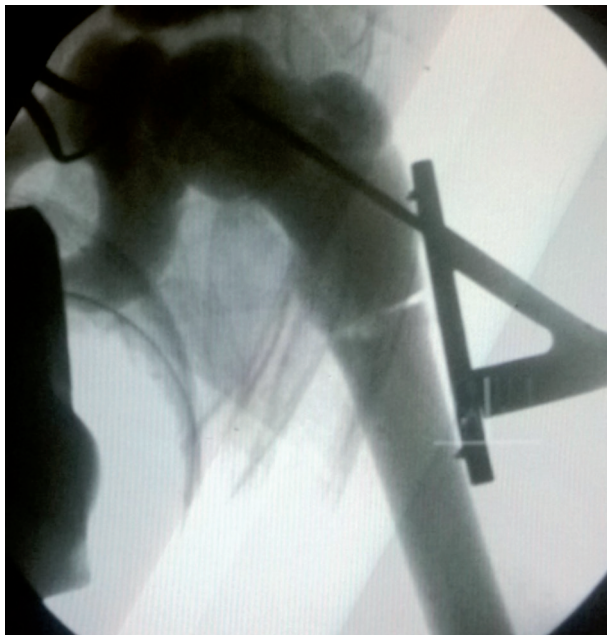


*Fig. 2.* — Radiograph showing the broadened distal femur

bones without any medullary canal were seen in whole of the skeleton. Initial management was by skin traction with a pillow kept underneath the thigh and analgesics to control the pain. Blood parameters were within normal range. A neurological examination of cranial nerves was found to be normal. A detailed family history was obtained from patient's relatives. The patient had a younger sibling with osteopetrosis who had suffered a fracture of olecranon a month back, which went on heal in a cast.

After searching the available literature for best possible treatment option, we weighed our options between non-operative management and operative management. We planned a non-operative management using a hip spica cast but also kept operative management as the backup plan. A pediatric short barrel dynamic hip screw (DHS) was planned in case acceptable reduction was not possible. Considering that drilling of bones in patients of osteopetrosis is difficult, we kept extra drill bits.

Under general anesthesia, the patient was positioned on fracture table and reduction attempted but the reduction was not acceptable and operative management was opted for. The steps followed were of a standard DHS procedure with skin incision made



**Fig. 3.** — Intra operative image intensifier image showing the reduction and the guide wire position

on the lateral aspect. After opening up the fracture site, we had to hold the proximal and distal fragments with bone holding clamps and manipulate the fragments under vision to obtain reduction. As the ideal starting point for the DHS lag screw would have been very near the fracture site, we accepted a relatively proximal starting point to avoid any possibility of the triple reamer entering the fracture site (Fig. 3). After opening the lateral cortex with a sharp drill bit, the guide wire was placed such that the tip did not cross the physis. We completed the process of triple reaming taking frequent breaks and using continuous irrigation with normal saline. Reamer was removed at regular intervals to clean the debris and irrigate the tract. Once the reaming was completed, lag screw of measured length was inserted followed by a 3-hole side plate. The process of drilling holes for insertion of the screws in the side plate was done slowly and with constant irrigation with normal saline and with repeated cleaning of the drill bit. Sharp taps were used to cut threads for the screws. Four drill bits were used for the whole procedure. One drill bit broke during the procedure but was easily retrieved. While inserting the screws, one screw broke off at the shank region. We had good purchase with the other two screws so we carried on and the procedure was completed without much struggle and complication (Fig. 4).



**Fig. 4.** — Radiograph obtained on post-op day one

Sitting up and active assisted exercises were started from post-operative day one under supervision. Sutures were taken off on day 10. After 3 weeks, supervised crutch walking with weight bearing as tolerated was permitted. At 12 weeks' post-surgery, we allowed full, unassisted weight bearing but asked for some restraint on jumping and running activities. After 6 months of surgery, we allowed unrestricted activity. At the last follow up at one and half years after surgery, the child was comfortable, pain free, playing and walking without any limp. He was comfortable in sitting cross-legged and squatting (Fig. 5 and 6). There was complete radiological union at the fracture site (Fig. 7 and 8).

## DISCUSSION

Infantile or malignant variety of osteoporosis is often diagnosed in infancy but the autosomal dominant form may not be diagnosed till late in life (1). Autosomal dominant type is considered compatible with normal life and often the patients are diagnosed for the first time when they present with a fracture and the radiographs obtained at that time reveal the presence of bones of different density (1). The radiographic picture of increased density of the whole skeleton along with an absence of medullary canal is hard to miss by someone who has some knowledge of this condition. Bone mineral density (BMD) and mechanical strength of bone are believed to have a positive correlation. In osteopetrosis though, the situation is different. However, in osteopetrosis even with an increased skeletal density radiographically there is an increased risk of fracture. Tuukkanen et al (12) evaluated the mechanical strength and mineral density in three osteopetrotic mutations in rats. They reported that in osteopetrosis the increased BMD and bone mass do not strengthen bone but lead to increased bone fragility when tested with bending forces and that the cross-sectional structure of long bone shafts was markedly different in osteopetrotic mutants, having a thin cortex and a medullary area filled with primary trabecular bone. They hypothesized that the osteopetrotic bone is weaker because the lack of bone remodeling permits the accumulation



*Fig. 5.* — Clinical picture at one and half years. Patient is sitting cross legged comfortably



*Fig. 6.* — Comfortable squats at one and half year follow up

of primary bone, which has inherently less strength than secondary bone.

The available literature on osteopetrosis spans all the fields of the medical science but less than one-tenth deal with orthopedic manifestations. Most of them are case reports and case series; showing that although rare, this condition is often diagnosed successfully. A definitive information on management options for such fracture is still lacking. An analysis of available literature showed that while conservative management could be successful (1,3,9), they were associated with their own problems like nonunion, deformities, prolonged traction and immobilization. A number of available reports favor



**Fig. 7.** — Antero posterior radiograph at one and half years follow up showing union of the fracture



**Fig. 8.** — Lateral radiograph at one and half year follow up, showing complete union at the fracture site

surgery to help obtain a better alignment, to aid in early mobilization and achieve a more reliable final outcome (2,6).

Birmingham et al (4) have provided a considerably detailed review of the cases managed non-operatively and operatively before the year 2008. Aslan et al (2) have provided a thorough review of cases reported between 2008 and 2013. On a detailed study of these reviews, one can conclude that femur is the most commonly fractured bone with the proximal part being the commonest site. Peritrochanteric fractures were seen in children in only two cases, rest all were in adults. These two cases had been managed non operatively (1,4). Georgiev and Alexiev reported good outcome of an intertrochanteric fracture after surgical management with a proximal femur LCP (7). Of the various implants used dynamic hip screw (DHS) was used in 3, dynamic condylar screw (DCS) in 2, intramedullary nails in 7 and locked compression plate (LCP) in 8 peri-trochanteric fractures. In 8 fractures older nail-plate combination devices were used. While

the devices used for the management have been different, all the authors have reported difficulty in performing the surgery because of high density brittle bones, which may sustain iatrogenic fractures if adequate precautions are not followed. Sen et al (10) have described the use of metal cutting drill bits.

In the present case, the child was otherwise normal, there were no cranial nerve involvement and the blood parameters were within range; thus, the child most likely had the autosomal dominant type of osteopetrosis. The fracture was completely displaced (Fig. 1) and the proximal fragment was in varus. Closed reduction and hip spica application under general anaesthesia was planned with a back-up operative plan in case the reduction was not acceptable. Implant selection was debated upon and considering the absence of a medullary canal and unavailability of age suitable implant, intramedullary nail was not further considered. We did not have LCP suitable for an eight-year-old child available but DHS in pediatric sizes was available and hence was opted as the implant of choice. The pro-

cedure was completed successfully with the minor complication of one screw breaking at the shank. The patient went on to have a complete union of the fracture with no deformity and no complications.

The case we have presented is of a rare condition, which has now been diagnosed, managed and published frequently in the literature. Still we believe that in absence of a consensus management option for the such fractures, every case of osteopetrosis managed by a different method is worth reporting. We were successful in managing a sub-trochanteric fracture in an eight-year-old child with DHS, which can be a good alternative in young children. While one may consider non-operative management, with good surgical technique one can avoid most of the complications of surgery and hence unsatisfactory reduction should not be accepted. That being said principles like low speed-high torque drilling, constant irrigation with cold saline, use of sharp drill bits/metal cutting drill bits and cleaning of drilled debris need to be followed religiously in these cases.

To summarize, an orthopaedic surgeon should have awareness of osteopetrosis and the expected difficulties in its management. The aim of fracture management should be to achieve sound union with the available treatment methods, be it operative or non-operative. The present case report thus adds to the knowledge base of osteopetrotic sub-trochanteric fractures in children and its management.

#### Acknowledgement

The authors want to thank the patient and his parents for providing us with consent for publication of the patient details and photographs.

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