Management of Pathological Fractures in Hyperostotic Bone Diseases - A review of three cases

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ABSTRACT

Fractures in hyperostotic bone disease are difficult to treat. In conditions like Pycnodysostosis and Osteopetrosis the bone are hyperostotic and are prone for fractures following trivial trauma. Though in Pycnodysostosis, the fracture unite in normal time they are prone for malunion. In case of osteopetrosis the healing time is variable and the orthopedic management is complicated by underlying abnormal cellular metabolism of bones [10]. Though these conditions are rare, their management presents unique and difficult problems to the orthopedic surgeon. In this case series we describe management difficulties faced during management of three cases of hyperostotic bone disease (2 cases of Pycnodysostosis and 1 case of osteopetrosis). Extramedullary fixation was done in two cases of Pycnodysostosis and intramedullary fixation was done in one case of osteopetrosis. In these patients bone union was achieved in normal time.

Keywords: Pycnodysostosis, Osteopetrosis, hyperostotic bone, extramedullary fixation, intramedullary fixation.

INTRODUCTION

Pycnodysostosis is a rare autosomal recessive disorder affecting the bone. Maroteaux and Lamy first reported this syndrome in 1962[7]. The disease is characterised by proportionate dwarfism, well developed secondary sex characteristics, peculiar facies, prominent forehead, beaked nose, and receding jaw, abnormal dentition with a usually normal palate-although sometimes it is cleft-and certain skeletal changes with multiple spontaneous fractures; finally ihere is usually consanguinity in the parents (Maroteaux and Lamy 1966; Dusenberry and Kane 1967) [2,7]. The similarity in appearance of the patients, regardless of sex and race, is striking (Elmore 1967) [3]. Mental retardation does not occur. Blood chemistry is usually normal. Anemia does not occur. Most striking feature- generalised increased density of the skeleton and thickening of the cortices of the long bones, but without obliteration of the medullary canals[2]. Except for the recurrent fractures patient have a good prognosis.

Marble bone disease was first described early in the 20th century (Albers-Schonberg 1904; 1907) although it appears to have occurred even in prehistoric man (RegOly-M# {233}rei 1962). About 200 to 300 cases of this rare condition have been reported (Johnston et al. 1968; Aegerter and Kirkpatrick 1975) [9]. The term osteopetrosis was first used by Karshner in 1926[10]. Osteopetrosis is characterised by increased density and widening of the bones [8]. Essentially, the lesion is one which involves the failure of resorption of calcified fetal chondroid and of primary bone, which prevents replacement by mature lamellar bone [11] .With associated general features like Anemia, thrombocytopenia,and Extramedullary hematopoiesis are manifest clinicallyas hepatosplenomegaly and lymph-node enlargement[12].Some have considered the hematological manifestations to be myelophthisic in nature, due at least in part to the exclusion of the marrow cells by the persistent endochondral tissue [12] . Many authors have reported its clinical features and described its histopathology (Krompecher 1940; Cohen 195 1; Johnston et al. 1968). Shapiro et al. in 1980 confirmed that the cause of the disease is impaired function of osteoclasts [12]. There is no effective treatment, and pathological fractures are an important complication. Their treatment may be difficult because of the abnormal bone structure and an increased susceptibility to bone infection [13]. The patients with these condition present with multiple fractures following trivial trauma. Though these fractures unite they are prone for malunion. So some form of internal fixation with or without bone grafting is essential to get good anatomical union [2, 13].

MATERIAL AND METHODS

We report three cases of hyperostotic bone disease who presented to our institution from the period of May 2007 to Dec 2007. Among them two had pycnodysostosis and one had osteopetrosis. Of them two were male and one female, average age between

Table -1-History, Clinical features and Management of three cases

Sl.no	History	Past/family history	Clinical features	Management
Case1	24 yrs/male presented with c/o pain in the right thigh following trivial trauma.	Had previous H/o fractures of right and left leg 7 and 4 yrs back respectively which was treated conservatively and went for malunion(fig 1).He is 3rd child of a consanginous marriage. His elder sister also has similar features and history of recurrent fractures	He is short statured(145 cm), with features like proptosis, beaking of nose(fig 2),crowding of teeth, maxillary hypoplasia, short stubby fingers(fig 3),pectus carinatum X -ray- shows transverse fracture shaft of femur rt side (fig 4), x ray skull showed wide open sutures(fig 5), x ray hand showed terminal osteolysis(fig 3).	The fracture was fixed with open reduction and internal fixation with broad dynamic compression plate (fig 6).
Case2	24 yrs/ female c/o pain in the right leg following trivial trauma.	There is previous history fracture to the same leg 2 years back which was treated conservatively and went for malunion .patient is the second child of a consanginous marriage.	She is short statured (138 cm), with characteristic facial features like proptosis, beaking of nose (fig7), crowding of teeth, short stubby fingers. Xray shows short oblique fracture of tibia with previous site of malunion proximal to it (fig 8).x ray skull shows wide open sutures, xray hand shows terminal osteolysis.	The fracture was fixed with open reduction and internal fixation with narrow dynamic compression plating (fig9).
Case3	20 yrs male c/o pain in the right thigh following trivial trauma.	Past history revealed subtrochantric fracture of the left femur two years back which was treated conservatively and went for malunion(fig 10). Patient is the second child of a consanginous marriage.	He is below average height (154 cm).he was blind at the time of presentation. Typically he was normal at birth but became blind couple of years back. X ray shows subtrochantric fracture shaft of femur, there is increased density of the cortex with narrowing of the medullary cavity (fig10).spine showed typical 'rugger jersey spine'(fig 11).blood examination showed hypochromic anaemia	The fracture was fixed with open reduction internal fixation with intramedulary nailing with humerus nail(fig 12).

Table 2: Salient features of pycnodysostosis and osteopetrosis

Features	Pycnodysostosis	Osteopetrosis
Genetic	Recessive	Dominant(mild) Recessive(malignant)
Stature	Short	Normal
Bone	Dense but marrow visible	Dense, no distinction between marrow and cortex possible
Skull	Dense base of skull Open frontanelles Wormian bones Aplastic sinuses	Dense base of skull Sutures and fontanelle unaffected Aplastic sinuses
Mandible	Mandibular angle straightened	Normal
Clavicle	Acromial end dysplastic	Present and normal
Hand	Atrophic terminal phalanges	normal
Pelvis	Coxa plana	Coxa vara
Haematological Findings	Normal	Hypochromic anaemia dominates the picture.

ILLUSTRATIONS

CASE 1

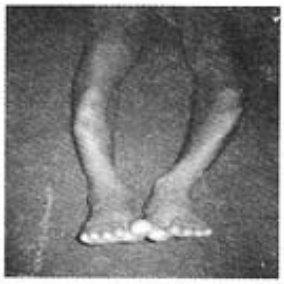

Fig.1 Malunited both bones both legs

Fig.2 Typical facies of pcynodysostosis

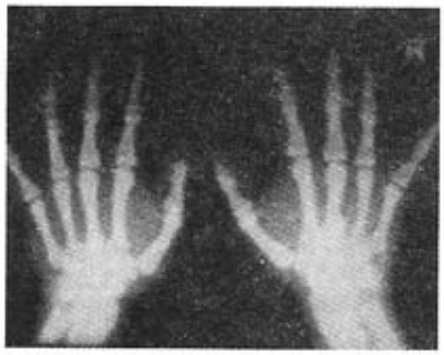

Fig.3 Xray hand showing terminal osteolysis

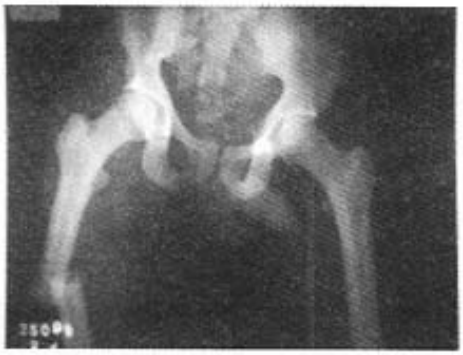

Fig.4 Transverse fracture Rt. shaft of femur

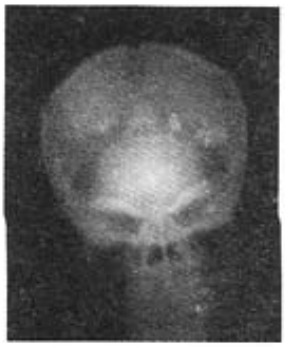

Fig.5 Xray skull showing wide open structure

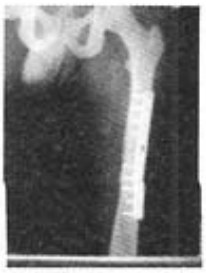

Fig.6 plate osteosynthesis and union after 3 months rt. side

CASE 2

Fig.7. Facial appearance of pycnodysotosis

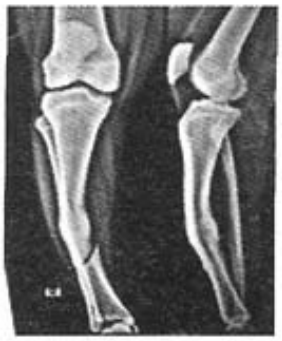

Fig.8. Xray right leg showing malunited fracture above with fresh fracture shaft of tibia

Fig.

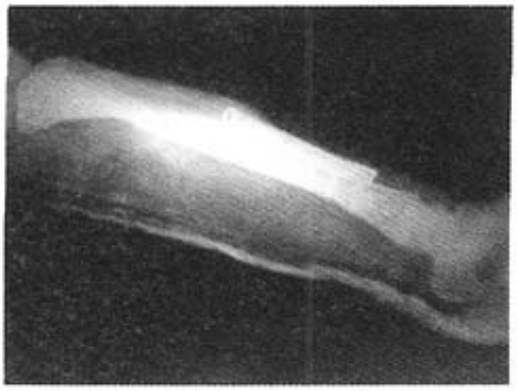

Fig.9. Internal fixation with narrow DCP

CASE 3

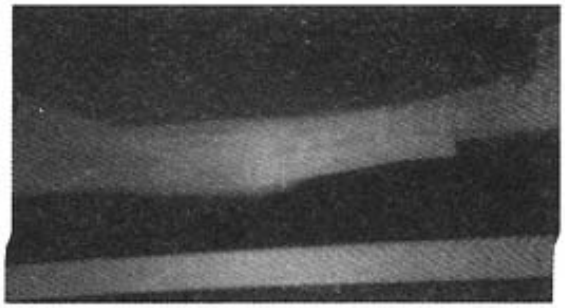

Fig.9. Internal fixation with narrow DCP

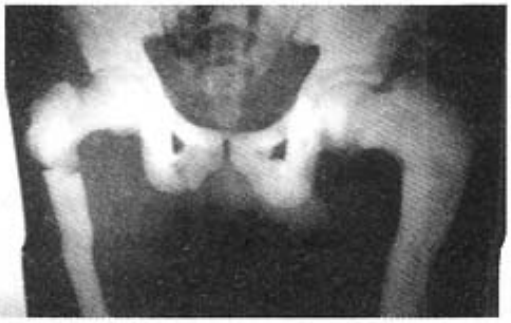

Fig.10. Pathological fracture rt. femur in osteopetrosis, narrow medullary canal

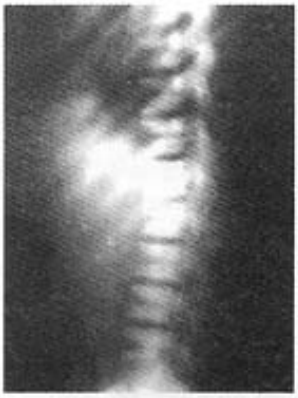

Fig.11. Osteoporosis xray dorsolumbar spine lateral view shows "Rugger Jersy Spine"

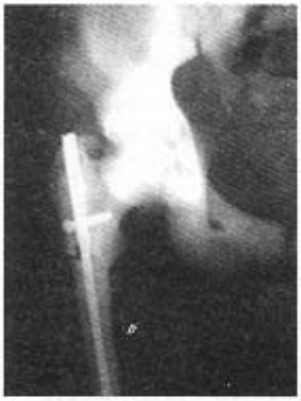

Fig.12. Open reduction and internal fixation with humerus nail. Post operative xray

20to30 years. All these patients presented with pathological fracture of the long bones. The clinical details of the patients are documented in table 1. Extramedullary fixation with dynamic compression plating was done in two cases of pycnodysostosis. Intramedullary fixation done with humerus nail was done in one patient with osteopetrosis.

RESULTS

All patients were followed in monthly intervals. In case one, patient was allowed non weight bearing for the first two months, then partial weight bearing for next month. The patients were allowed to weight bear fully after 3 months .The fracture united in three months (fig 7).

In case two, the x-ray showed signs of callus at 2 Months. She is now on partial weight bearing crutch walking till our last followup (fig 11).

In case three, after one month of follow up there is no signs of union yet. Patient has been advised to continue non weightt bearing crutch walking.

DISCUSSION

Hyperostotic bone diseases are rare in occurrence. Though in terms of fracture management there is not much difference, it is important to differentiate between pycnodyostosis and osteopetrosis. The salient and differentiating features are given in the following table 2.

Fractures in this patient usually unite in normal time.But they are prone for malunion leading to deformity.As this further leads to refracture,it almost always needs some form of internal fixation.

Now the question is:

Whether to go for intramedullary fixation or extrameullary fixation?

In the first two cases exramedullary fixation was done, during the surgery the bone was found to be very hard. Medullary canal was found to be very narrow.

We found it difficult even to insert an artery forceps into it. Drilling process was quiet tedious even with a power drill and new drill bits.

In the third case intramedullary fixation was planned. During the procedure the reaming process was found to be quiet cumbersome. Serial reaming was done with drill bit on power drill. While reaming false passage was found to be created in distal fragment, so rereaming was done. There is increased chance of splintering, false passage formation during the procedure .Humerus nail was inserted and proximal locking was done.

CONCLUSION

The fracture in hyperostotic bone is unique in their own respect. Fracture fixation presents various problems to orthopaedic surgeon. Though the bones are brittle enough to undergo pathological fracture, their fixation is difficult due to sclerotic cortex.

Whether to go for extramedullary or intramedullary fixation is a subject of controversy. It is clear that extramedullary fixation is technically easy compared to intramedullary fixation. The decision between them can be taken according to the surgeons' choice.

The essence is essential internal fixation, adequate preoperative planning and surgical skill to tackle the per-operative difficulties for successful management of the pathological fracture in hyperostotic bones.

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