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Surgical management of humeral non-union in pediatric osteopetrosis: A case report

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Abstract
Background: Osteopetrosis is a rare genetic bone disorder characterized by impaired osteoclastic bone resorption leading to increased bone density and susceptibility to fractures. Fracture non-union in osteopetrosis patients presents unique therapeutic challenges due to altered bone biology and technical surgical difficulties. While conservative management may be appropriate for certain pediatric cases, surgical intervention with rigid fixation and biological augmentation is often necessary for displaced fractures and established non-unions.
Case Presentation: An 8-year-old boy presented with a 2-year persistent deformity of the right proximal humerus following traumatic injury and initial TENS nail fixation. Implant removal 6 months prior resulted in pseudarthrosis. Physical examination revealed painless abnormal mobility at the non-union site with preserved joint motion. Laboratory investigations were normal, excluding infection and metabolic causes. Skeletal survey revealed characteristic osteopetrosis features: costochondral thickening, rugger jersey spine, Erlenmeyer flask deformity, bone-within-bone appearance, thickened skull cortices, and impaired dentition. Surgical management consisted of extended deltopectoral approach, debridement, corticocancellous bone grafting, and rigid fixation with locking reconstruction plate. Significant technical difficulty was encountered during drilling due to dense cortical bone.
Result: Histopathological examination confirmed osteopetrosis with irregular hyaline cartilage cores within woven and cortical bone. Three-cortex union was achieved at 10 weeks postoperatively. At 1-year follow-up, the surgical site healed completely without complications or deformity, and shoulder range of motion was symmetric with the contralateral side. The patient demonstrated excellent functional recovery and returned to age-appropriate activities.
Conclusion: Humeral non-union in pediatric osteopetrosis can be successfully managed with comprehensive skeletal evaluation to identify the underlying disorder, meticulous surgical technique employing locking plate fixation combined with biological augmentation, and structured postoperative rehabilitation. Recognition of osteopetrosis through characteristic radiographic features and histopathological confirmation enables appropriate surgical planning despite technical challenges inherent to osteopetrotic bone density.

Keywords: Osteopetrosis, humeral fracture, non-union, pediatric, locking plate, bone grafting, surgical fixation

Introduction
Osteopetrosis, also known as marble bone disease, is a rare genetic skeletal disorder characterized by defective osteoclastic bone resorption, resulting in abnormally dense yet brittle bones with increased fracture susceptibility. The disease encompasses a spectrum of clinical presentations, from asymptomatic forms discovered incidentally to severe manifestations with multiple systemic complications [1]. Radiographic features include diffuse osteosclerosis, bone-within-bone appearance (endobones), Erlenmeyer flask deformity of long bones, rugger jersey spine, and loss of corticomedullary differentiation [2, 3].

Fractures in osteopetrosis patients present unique therapeutic challenges due to the altered bone properties, impaired remodeling capacity, and technical difficulties during surgical intervention. The management of non-union in this patient population is particularly complex, requiring meticulous surgical planning, appropriate implant selection, and enhanced biological augmentation. While conservative management may be appropriate for certain pediatric fractures with acceptable alignment, surgical intervention is often necessary for displaced fractures and established non-unions [3].

Humeral fractures in pediatric osteopetrosis patients are uncommon, and literature on the management of humeral non-union in this population remains limited. Most reported cases

involve femoral fractures, with sparse data on upper extremity involvement. Surgical fixation in osteopetrotic bone is technically demanding due to increased bone density, narrow or obliterated medullary canals, and the propensity for drill bit breakage and iatrogenic fractures. Furthermore, fracture healing is significantly delayed compared to normal bone, with higher rates of non-union, infection, and implant failure [4].

This case report describes the comprehensive evaluation, surgical management, and functional outcomes of a complex humeral non-union in an 8-year-old boy with previously undiagnosed osteopetrosis, emphasizing the diagnostic approach, technical considerations, and postoperative rehabilitation strategies.

Case Report

Clinical Presentation

An 8-year-old male child presented to the orthopedics outpatient department with a deformity of the right shoulder and upper arm persisting for two years. The patient had sustained a traumatic injury to the right upper arm while playing at school two years prior, for which he underwent titanium elastic nail system (TENS) fixation at an external hospital. The implant was removed 1.5 years after insertion (6 months prior to presentation), following which persistent deformity and functional limitation prompted referral to our institution for evaluation and management of non-union of the right proximal one-third humerus.

Clinical Examination

Physical examination revealed a visible deformity of the right upper arm without evidence of active infection. There were no sinuses, purulent discharge, or local warmth. Painless abnormal mobility in two planes was present at the non-union site. Passive range of motion of the shoulder and elbow joints was preserved and within normal limits. Comprehensive head-to-toe examination revealed no dysmorphic features or syndromic associations. Anthropometric measurements demonstrated age-appropriate height and weight according to Indian Academy of Pediatrics (IAP) growth charts. No family history of bone disorders or rheumatological diseases was elicited. Perinatal history was unremarkable, and there was no history of similar conditions in siblings.

Investigations

Laboratory investigations including complete blood count, erythrocyte sedimentation rate (ESR), C-reactive protein (CRP), alkaline phosphatase (ALP), serum calcium, and serum phosphate were within normal reference ranges, effectively excluding metabolic bone disease, active infection, and rickets.

Radiographic evaluation (figures 1 to 5) of the right humerus confirmed pseudarthrosis at the proximal one-third shaft. No features suggestive of rickets were identified. Given the persistent non-union following adequate initial treatment, a comprehensive skeletal survey was undertaken. Additional radiographs revealed multiple characteristic findings: costochondral thickening, rugger jersey appearance of the spine (sandwich vertebrae with sclerotic endplates), loss of corticomedullary differentiation in the femur, Erlenmeyer flask deformity (metaphyseal widening) of the distal femur, bone-within-bone appearance (endobones) in hand radiographs, thickened skull cortices, and improper dentition on lateral skull radiographs. These constellation of radiographic features raised strong clinical suspicion of osteopetrosis as the underlying predisposing condition for non-union.

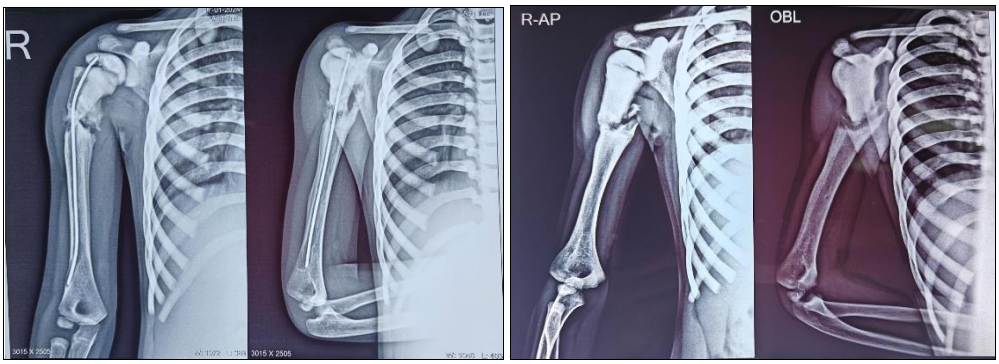

Fig 1: Old xrays- before and after TENS removal

Fig 2: current Xray showing proximal humoral pseudo-arthrosis and costochondral thickening

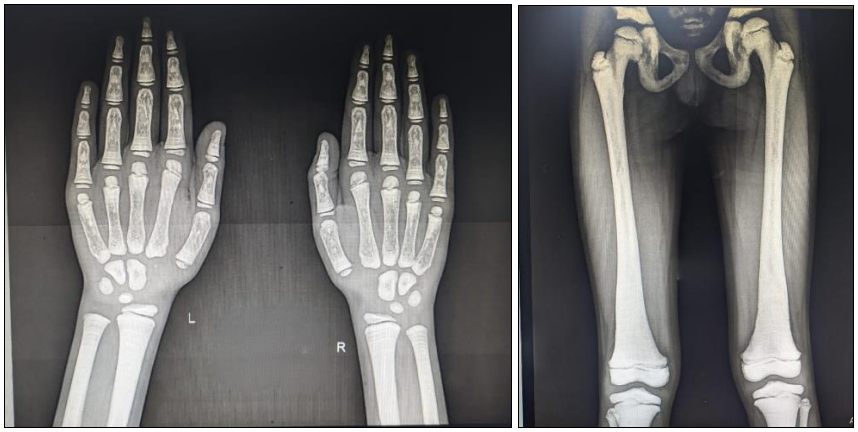

Fig 3: Endobone appearance, Erlenmeyer flask deformity, loss of corticomedullary differentiation

Fig 4: Thick skull cortex, poor dentition. Rugger jersey spine

Fig 5: post op radiograph

Surgical Management

Following comprehensive counseling of the parents regarding the diagnosis, prognosis, and potential surgical complications, informed consent was obtained. The patient underwent surgery under general anesthesia in the supine position. An extended deltopectoral approach was utilized to access the non-union site. Thorough debridement of the non-union site was performed, and tissue samples were sent for microbiological culture and histopathological examination. No purulent material was encountered intraoperatively. The medullary cavities of both proximal and distal fragments were opened to enhance biological potential.

Cortico-cancellous autograft was harvested and applied at the non-union site to improve the biological environment and promote osteogenesis. Rigid internal fixation was achieved using a locking reconstruction plate to provide mechanical stability. Physis was carefully spared. As anticipated from the preoperative diagnosis of osteopetrosis, significant technical difficulty was encountered during drilling for screw insertion due to the extremely dense cortical bone. Slow-speed, high-torque drilling with frequent cooling using normal saline was employed to prevent thermal necrosis and drill bit breakage. Following fluoroscopic confirmation of satisfactory reduction and fixation, the wound was closed in layers over a suction drain. Postoperatively, the limb was immobilized in a U-slab for 8 weeks to augment mechanical stability and protect the fixation construct during early healing.

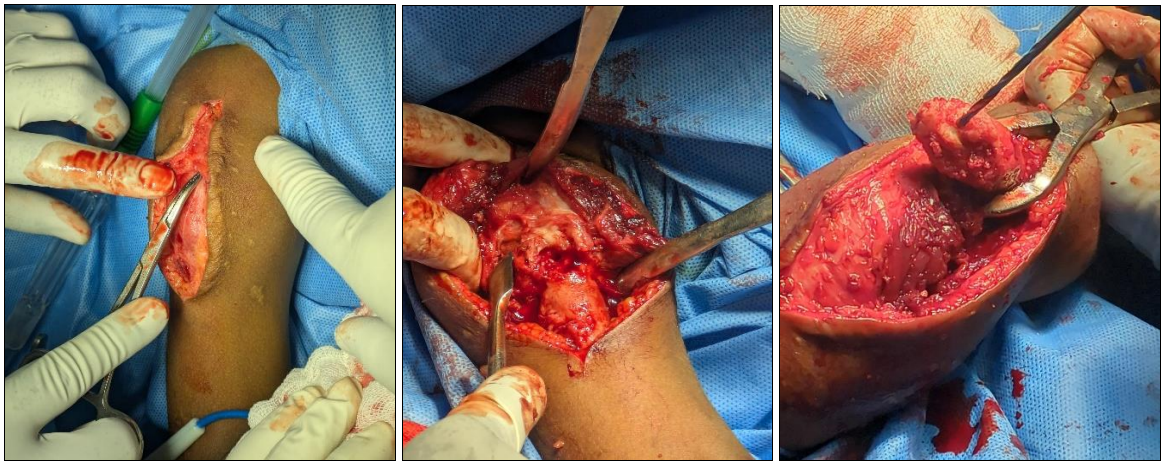

Fig 6: Incision, exposure, debridement and medullar canal opening

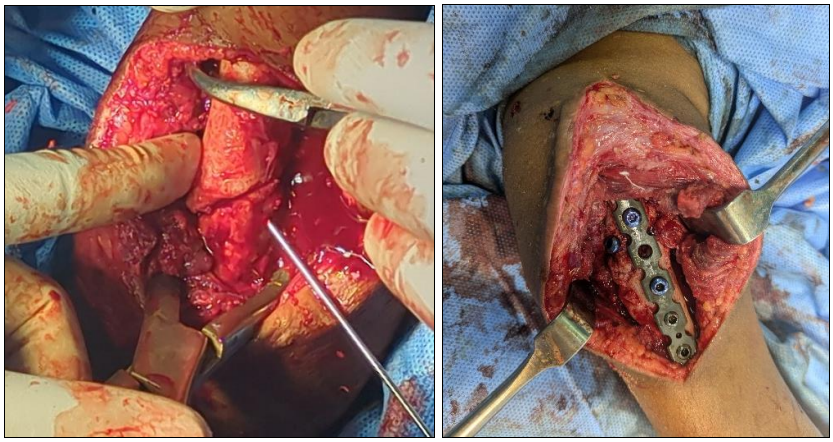

Fig 7: Reduction and fixation

Histopathological Findings

Histopathological examination of tissue obtained from the non-union site revealed irregular cores of hyaline cartilage embedded within woven and cortical bone, a finding strongly consistent with osteopetrosis. This confirmed the clinical and radiological suspicion and established the definitive diagnosis.

Rehabilitation and Follow-Up

After 8 weeks of immobilization, a cautious, graded rehabilitation protocol was initiated. Passive range of motion exercises were commenced, followed by assisted active movements and eventually active range of motion exercises, progressing over an 8-week period. Union rate was significantly slower compared to similar fractures in age-matched children with normal bone quality. Three-cortex union was achieved at 10 weeks postoperatively, as confirmed by serial radiographic assessment.

At one-year follow-up, the surgical site had healed satisfactorily without evidence of infection, implant failure, or recurrent deformity. Range of motion of the shoulder joint was symmetric and comparable to the contralateral side. The patient demonstrated excellent functional recovery with no activity restrictions and returned to age-appropriate activities including sports participation.

Discussion

This case exemplifies the complex interplay between an underlying bone disorder and fracture healing complications, highlighting the importance of comprehensive skeletal evaluation in cases of delayed union or non-union following adequate initial treatment. Osteopetrosis, though rare, should be considered in the differential diagnosis of persistent non-union, particularly when radiographic features suggest increased bone density [5].

The autosomal dominant form of osteopetrosis (osteopetrosis tarda or Albers-Schönberg disease) typically presents in late childhood or adolescence with pathological fractures following minor trauma. Approximately half of affected individuals are asymptomatic and diagnosed incidentally or following fracture. The characteristic radiographic features diffuse osteosclerosis, bone-within-bone appearance, Erlenmeyer flask deformity, rugger jersey spine, and thickened skull base are pathognomonic and sufficient for diagnosis [1, 2]. In our patient, these classic features were present on skeletal survey, and the diagnosis was confirmed histopathologically.

Fracture management in osteopetrosis presents significant challenges. Conservative treatment may be appropriate for minimally displaced fractures in children, given the remodeling potential of the pediatric skeleton [6, 7]. However, Erkus *et al*. reported successful conservative management of a humeral fracture in a 9-year-old girl with osteopetrosis tarda, demonstrating complete remodeling over 7 years [4]. Nevertheless, surgical intervention is often necessary for displaced fractures, polytrauma, floating elbow, or established non-union, as in our case.

The initial management with TENS nailing in our patient was appropriate for the acute fracture presentation.

However, TENS fixation may provide inadequate stability in osteopetrotic bone due to the narrow medullary canal and altered biomechanical properties. The subsequent development of non-union likely resulted from inadequate biological environment and mechanical instability, compounded by the impaired bone remodeling inherent to osteopetrosis.

Surgical fixation in osteopetrotic bone demands meticulous technique and anticipation of technical difficulties. The extremely dense cortical bone makes drilling arduous and time-consuming, with increased risk of drill bit breakage, thermal necrosis, and iatrogenic fracture. Aslan *et al*. emphasized the importance of slow-speed, high-torque drilling, frequent cooling with saline, and availability of multiple drill bits [5]. In our case, these precautions were taken, and despite anticipated difficulties, satisfactory fixation was achieved without drill bit breakage or iatrogenic complications.

The choice of locking reconstruction plate for definitive fixation was based on several considerations. Locking plates provide angular stability and fixed-angle constructs that do not rely on friction between the plate and bone, thereby preserving periosteal blood supply. This is particularly advantageous in osteopetrosis, where endosteal blood supply is compromised by narrow medullary canals and impaired vascularization. Studies have demonstrated superior outcomes with locking plate fixation compared to conventional plates in osteopetrotic fractures. The rigid fixation achieved with locking screws provides absolute stability conducive to primary bone healing, which may be more favorable than secondary healing in osteopetrotic bone [6, 7].

Biological augmentation with corticocancellous bone grafting was performed to enhance the healing environment. While some authors advocate for bone morphogenetic protein (BMP) supplementation to promote osteogenesis in osteopetrotic non-union, autogenous bone grafting remains the gold standard for biological stimulation. The combination of mechanical stability through rigid internal fixation and biological enhancement through bone grafting addresses both arms of the "diamond concept" of fracture healing, which emphasizes mechanical environment, biological environment, osteogenic cells, and growth factors [7].

Prolonged immobilization for 8 weeks postoperatively was employed to protect the fixation construct during early healing, given the known delayed consolidation in osteopetrotic bone. Aslan *et al*. reported that fracture healing in osteopetrosis is slower than normal, often requiring extended time to achieve radiographic union [5]. Our patient achieved three-cortex union at 10 weeks, which, while delayed compared to normal bone, represents a favorable outcome for osteopetrotic non-union.

The cautious, graded rehabilitation protocol was designed to balance early mobilization to prevent joint stiffness with protection of the healing bone. Studies have demonstrated that functional outcomes in pediatric humeral fractures are excellent with appropriate rehabilitation [4]. Our patient achieved full range of motion comparable to the contralateral side at one-year follow-up, with no functional limitations.

This case underscores several important principles in the management of fractures and non-union in osteopetrosis. First, comprehensive skeletal evaluation is warranted in cases of persistent non-union following adequate initial treatment to identify underlying bone disorders. Second, surgical fixation with locking plates combined with biological augmentation represents an effective treatment strategy for osteopetrotic non-union. Third, anticipation of technical difficulties, meticulous surgical technique, and availability of appropriate equipment are essential for successful outcomes. Fourth, prolonged immobilization and cautious rehabilitation are necessary given the delayed healing characteristics of osteopetrotic bone. Fifth, despite the challenges, excellent functional outcomes can be achieved with appropriate management.

Potential complications that warrant vigilance during follow-up include infection, implant failure, recurrent fracture, and delayed union or non-union. The infection rate in osteopetrotic fractures has been reported as 12-29% in some series, related to prolonged operative time, impaired vascularity, and potential marrow involvement.[4] Our patient experienced no infectious complications, likely attributable to meticulous sterile technique, prophylactic antibiotics, and thorough debridement. Long-term follow-up is essential to monitor for refracture, a recognized complication given the inherent fragility of osteopetrotic bone.

Conclusion

This case demonstrates that humeral non-union in pediatric osteopetrosis can be successfully managed with comprehensive preoperative evaluation, meticulous surgical technique employing locking plate fixation with biological augmentation, and structured postoperative rehabilitation. Early recognition of osteopetrosis through characteristic radiographic features is crucial for appropriate treatment planning and counseling. Surgeons should anticipate technical challenges related to bone density and be prepared with appropriate equipment and techniques. Despite inherent difficulties, excellent functional outcomes are achievable with combined mechanical stability and biological enhancement. This report adds to the limited literature on upper extremity fracture management in pediatric osteopetrosis and emphasizes the importance of multidisciplinary approach in managing complex skeletal disorders.

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Conflict of Interest

The authors declared no potential conflicts of interest with regard to the submitted article. Each author certifies that he or she has no commercial associations that might pose a conflict of interest in connection with the submitted article.

Ethical standards

Institutional ethics committee approved the study.

Informed consent was obtained from the participant (legal guardian) included in the study

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