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TRAUMA SURGERY

Recommendations for fracture management in patients with osteopetrosis: case report

Miguel A. Farfán · Carlos M. Olarte · Rodrigo F. Pesantez · Sebastian Suárez · Linda Vallejo

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Abstract

Introduction Osteopetrosis is caused by general increase in bone density and obliteration of the medullary canal. Fractures are a frequent complication and their management is considered a challenge due to increased resistance to reaming and screw positioning; reduction maneuvers have to be done more carefully to avoid intraoperative fractures, and there is an increased risk of drill breakage. There is also a higher risk of infection and malunion, which increases the incidence of surgical revisions in this population.

Case report 55-year-old male with osteopetrosis and a history of two previous proximal femur fractures, who sustained an oblique supracondylar fracture of the left humerus and a simple, intra-articular, rotated fragment with capitelum involvement, as well as a fracture in the base of the coronoid process was admitted in our hospital. We performed an open reduction and internal fixation (ORIF) and 12 months after surgery, the patient's bone has healed and he recovered flexion ($11\mathring{0}$) and extension ($-1\mathring{0}$) without complications.

Discussion During ORIF, two drill bits were broken and screw fixation was challenging due to the strength required. Bone overheating was also present during drilling, evidenced by smoke production and increased temperature of both bone and drill bits. Recommendations to avoid these problems include continuous cold saline irrigation, frequent drill bit changing, and spaced cycles with low-speed drilling. Additionally, high-resistance and high-speed electric drill bits can also be effective. Finally, patients should be closely followed postoperatively due to the high incidence of refracture, infection and malunion.

Conclusions Fracture fixation in patients with osteopetrosis requires strategies to overcome the technical difficulties found during the procedure. Preoperative planning must include the availability of multiple metal drill bits, electric drills, and bone substitutes, having in mind drilling techniques, drilling speed, and temperature control. Patients should be closely followed to evidence any complications such as infections and malunions.

Keywords Osteopetrosis · Supracondylar fracture · Intra-articular fractures · ORIF

Introduction

Osteopetrosis, otherwise known as Albert Schönberg disease, is a genetic disorder mainly characterized by an increase in bone density. This disease also causes bone sclerosis, associated with cranial nerve compression, and increases the risk of having pathologic fractures and osteomyelitis. The increased risk of osteomyelitis is caused by leukocyte malfunction and a decrease in bone blood supply [1]. Additionally, there can be obliteration of the medullary canal. Approximately 50 % of the patients with benign osteopetrosis are asymptomatic, while 40 % can have pathologic fractures, which are usually transverse in nature [2]. Time for consolidation in these types of fractures is usually prolonged, and even though a standard treatment exists, bone remodeling is absent. The diagnostic workup on osteopetrosis is usually started after an incidental

M. A. Farfán (✉) · C. M. Olarte · R. F. Pesantez · S. Suárez
Department of Orthopaedic Surgery, Fundación Santa Fe de Bogotá, Bogotá, Colombia
e-mail: farfan4084@hotmail.com

C. M. Olarte · L. Vallejo
Department of Orthopaedic Surgery, Hospital Infantil de San José, Bogotá, Colombia

finding of hypersclerotic bones on x-ray films, and as of today, there is no definitive treatment for this pathology [3].

In North America, incidence is low: 1/20,000 for the autosomal dominant variant and 1/300,000 for the autosomal recessive variant [4, 5]. Unfortunately, the prevalence or incidence in Colombia is unknown due to the lack of judicious studies and due to its rareness, orthopedic surgeons lack the knowledge to adequately treat these patients.

There are three known fundamental variants according to their genotype: autosomal dominant (mild), intermediate autosomal recessive (moderate) and autosomal recessive (severe). Generalized bone sclerosis results from either a defective bone resorption mechanism or from the overall absence of osteoclasts [3, 6]. Thus, there is an excessive accumulation of bone matrix that produces solid, dense, and rigid bones and a reduction of the medullary canal [1]. As a result, Young's modulus of elasticity is reduced, so bones have a lower resistance to bending forces, and therefore have an increased risk of fracture [5].

Fracture fixation in patients with osteopetrosis is a challenge for the orthopedic surgeon because of abnormal bone strength and fragility, especially when the procedure involves extensive reaming and drilling. Due to bone characteristics, there are limitations such as increased resistance to reaming and screw positioning, reduction maneuvers have to be done more carefully to avoid intra-surgical fractures; there is an increased risk of drill breakage, and overheating of bone and drill bits.

Bone reduction and fixation also has an increased risk of infection and malunion in patients with osteopetrosis. These can predispose to an increased number of surgical revisions to achieve adequate bone healing, which can be further enhanced through the use of bone grafts or bone substitutes. Therefore, recommendations to avoid these problems include continuous cold saline irrigation, frequent drill bit changing, and spaced cycles with low-speed drilling. Additionally, usage of high-resistance and high-speed electric drill bits are used to avoid those problems. Finally, due to the high incidence of complications, patients must be closely followed up postoperatively at 1 week, 1, 3, 6, 9 and 12 months.

Current management for osteopetrosis is comprehensive, and should include medical and surgical treatments, as well as education for the patient to prevent fractures and other complications in the future.

Clinical case

50-year-old man with a known history of mild osteopetrosis, who sustained a low-energy fall, receiving impact on his left upper extremity was admitted in our hospital. He consults with pain, left upper limb deformity and functional limitation.

He is admitted to the hospital for initial evaluation, and after performing a complete physical examination and ruling out any neurovascular deficit, we diagnosed an oblique supracondylar fracture of the left humerus and a simple, intra-articular, rotated fragment with capitelum involvement in elbow x-rays (Fig. 1a, b). Additionally, there was also evidence of a fracture in the base of the coronoid process, which was later corroborated in a CT scan (Fig. 2). Finally, a generalized increase in bone density was evidenced, with the subsequent stenosis of the medullary canal as part of his osteopetrosis diagnosis.

With this diagnosis, our preoperative planning included: availability of multiple metal drill bits, and power drills for slow-speed cycled drilling. Additionally, frequent follow-up was also discussed with the patient before surgery.

The patient was treated with ORIF with anatomical plates and is positioned in lateral decubitus, with the left arm flexed and supported by a padded post, as shown in Fig. 3. We then performed a paratricipital approach by making a medium-sized, posterior incision. After the exposure and direct visualization of the fracture, we confirmed cortical bone thickening, which increased resistance during drilling, and caused it to break twice during the procedure. Bone overheating was also present during drilling, evidenced by smoke production and increased temperature of both bone and drill bits. After encountering such difficulties, we decided to frequently change drill bits, and used slow-speed cycled drilling.

Fixation was carried out using two 3.5-mm anatomical, locking plates[1] with a variable angle in an orthogonal disposition. The coronoid process was reduced and fixed with two crossed screws from posterior to anterior, as shown in Fig. 4a, b.

After surgery, the patient was placed in a cast for initial stabilization, which was removed 1 week later. Afterwards, he started physical therapy and was seen at our clinic with x-ray films to evaluate clinical and radiographical improvement at 1, 3, 6 weeks, 3, 6, 9 and 12 months after the surgery. This was done to detect any possible complications. Additionally, the patient was referred to a nutritionist, dentist and endocrinologist.

12 months after surgery, the patient's fracture has healed (Fig. 5a, b) with no associated complications, and he also recovered flexion ($11\mathring{0}$) and extension ($-1\mathring{0}$) (Fig. 6a, b). The patient is now fully functional and has returned to his daily activities with no limitations.

[1] Synthes, Inc. (West Chester, PA).

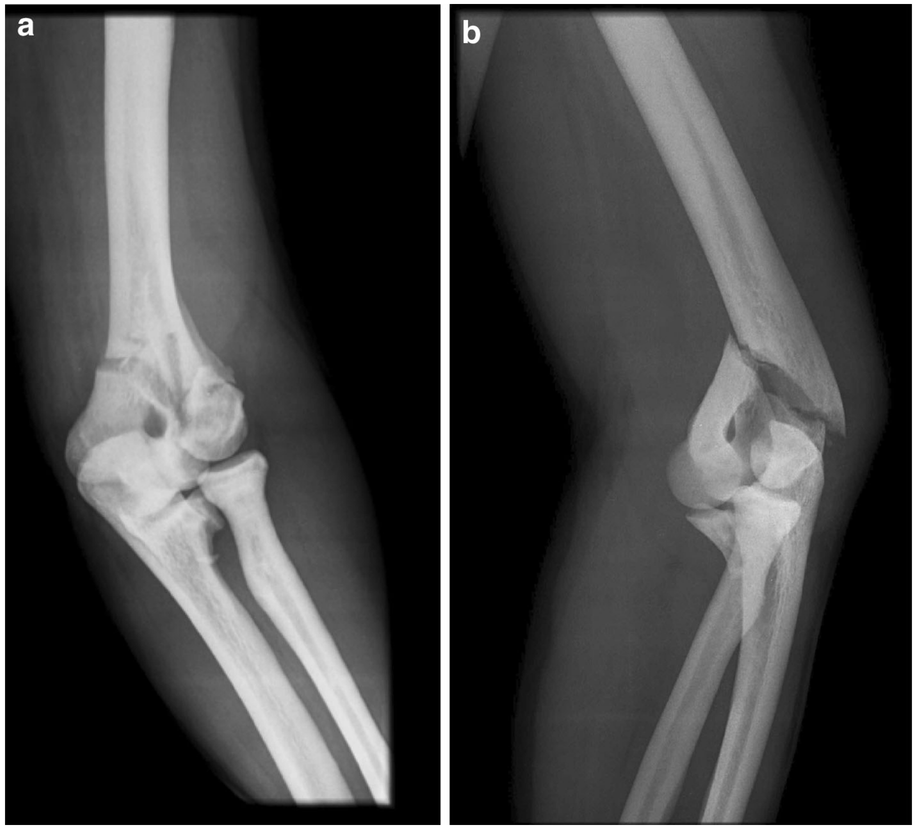

Fig. 1 **a**, **b** X-ray film showing the non-displaced oblique supracondylar fracture of the left elbow

Fig. 2 Left elbow CT scan showing supracondylar fracture of the left humerus, complete fracture of the coronoid apophysis, and posterior dislocation of the elbow

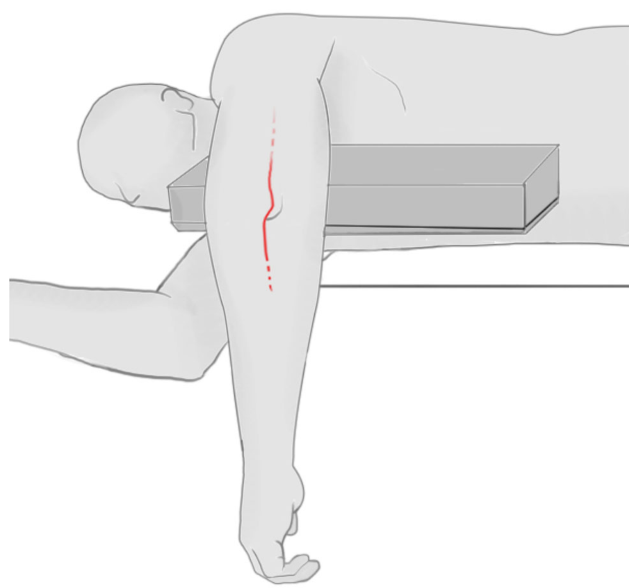

Fig. 3 Positioning of patient in lateral decubitus with the upper arm supported by a padded post. Posterior incision is shown

Discussion

It's important to highlight that this patient's fracture is pathological in nature as a result of a low-energy trauma in a middle-aged patient. This can be inferred by the fracture pattern, since it has linear tracks, bone sclerosis and medullary canal obliteration.

Various authors have given recommendations on the treatment for fractures in patients with osteopetrosis.

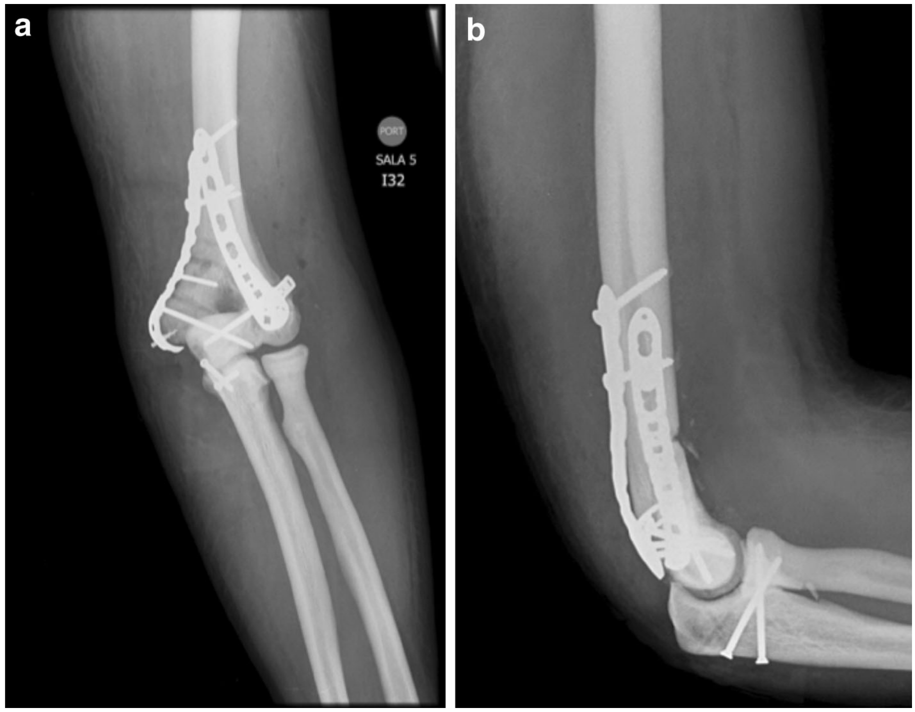

Fig. 4 **a**, **b** Immediate ORIF results

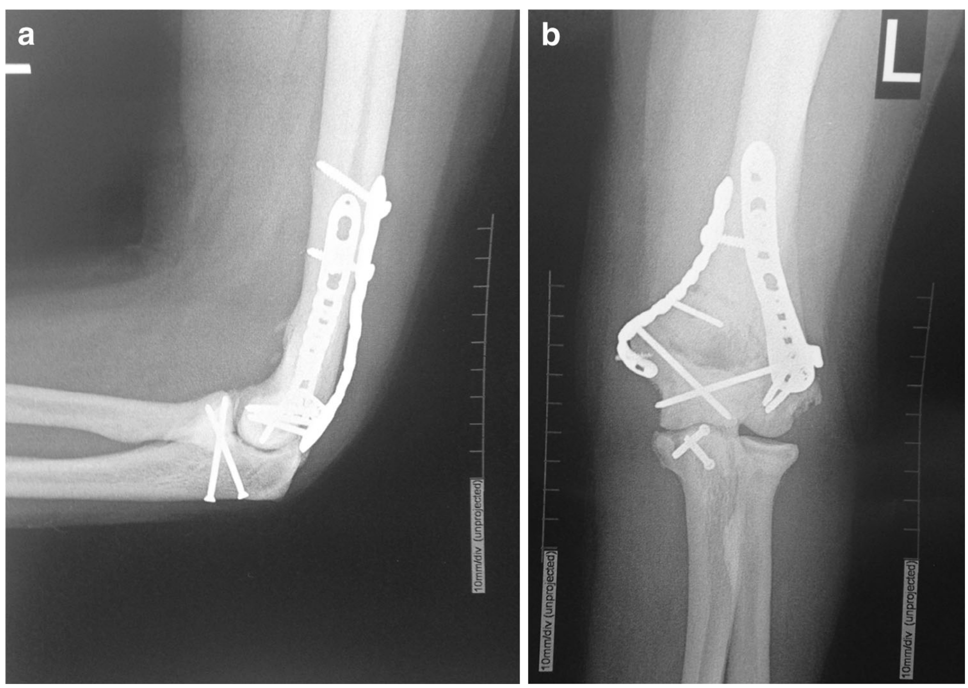

Fig. 5 **a**, **b** 1-year follow-up ORIF results

Historically, shaft fractures in long bones were treated orthopedically, especially in children [7]. However, intertrocantheric and subtrocantheric femur fractures are preferably managed with internal fixation due to the risk of delay in consolidation and nonunion [5].

There is also a set of recommendations for fractures that require ORIF to overcome the bone's abnormal strength and fragility. Ramesh et al. [8] described how bone perforation is the technical aspect with the highest complexity when dealing with these types of fractures due to its increased strength. They suggest using a metal-cutting drill bit to make the holes, followed by a regular, 3.2-mm drill bit to facilitate the surgical procedure and decrease the risks of thermal necrosis. In addition, they also recommend

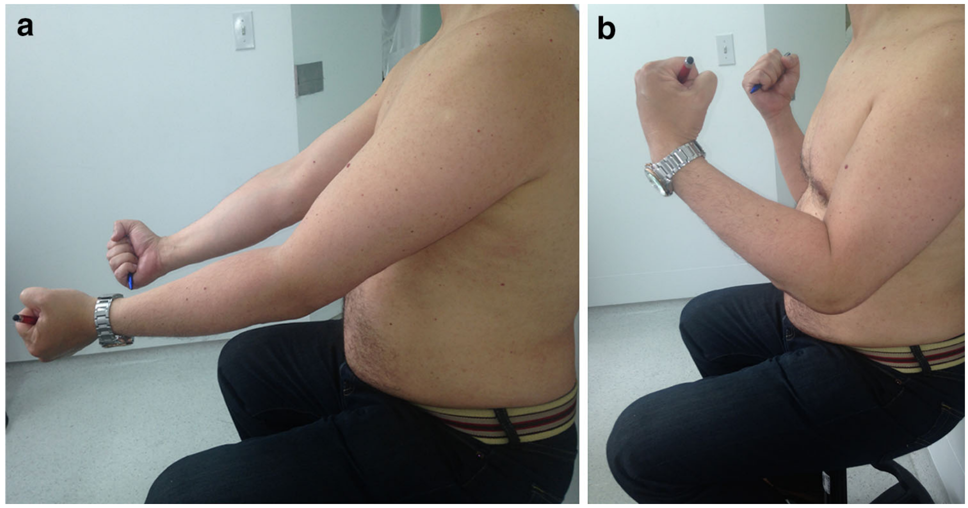

Fig. 6 **a**, **b** 1-year follow-up elbow range of motion

Table 1 Recommendations for fracture management in patients with osteopetrosis

Preoperative planning	When suspecting osteopetrosis, have in mind typical X-ray findings: increased bone density, obliteration of the medullary canal, cortical thickness, fracture in a low-energy trauma
	Availability of multiple metal drill bits, electric drills, high-resistance drill bits, high-speed electric drill, and bone substitutes
	Analyze type of fracture, previous fractures, comorbidities and rehabilitation possibilities
During surgery	Use continuous cold saline irrigation during drilling
	Change drill bits frequently
	Drill at low speed and in spaced cycles
	Avoid torsional and bending forces during surgery to prevent intrasurgical fractures
	Avoid mallet usage
Postoperative follow-up	Consider close follow-up (1 week, 1, 3, 6, 9 and 12 months)
	Consider evaluation by other specialists (nutritionist, dentist, endocrinologist)
	Be aware of higher risk of infection and malunion

the use of continuous irrigation with normal saline during the drilling process to avoid heating and rupture of the drill bit. High-speed, electrical motors are the best option in these patients. Therefore, Srivastay et al. [5] suggest bringing multiple, spare drill bits to the operating room in case one of them breaks during the procedure. They also recommend using selftapping or selfdrilling screws.

Moreover, Bhargava et al. [8] suggest frequent changing of drill bits during perforation, progressive cooling of bone during the procedure, and continuous normal saline irrigation. Constant cooling and changing of material will avoid bone necrosis, loosening of implants, and infection. Bone must be gradually perforated with as little depth as possible in each try. Also, considering the fragility of bone and its increased risk of fracture, the use of the mallet should be avoided [1]. It is important to remember that the use of cancellous bone autografts is not possible in these patients due to the inherent characteristics of bone.

Orthopedic surgeons should suspect this rare entity before the procedure to adequately plan the surgical approach and consider the possible complications that can occur during fixation [9]. One should suspect this disease in two scenarios: first, patients presenting with major fractures that were caused by inconsequent impacts and, second, x-ray films with universal bone sclerosis and medullary stenosis, particularly in the axial skeleton.

After treating the patient and reviewing the recommendations in the literature, we adopted some of them to improve patient care and avoid difficulties during surgery and postoperative complications. Some of the recommendations we used were drill bit changing, and slow-speed cycled drilling. However, we did not cool down bone and drill bits during the procedure, which led to overheating and breakage of two drill bits. We highly encourage orthopedic surgeons to follow the recommendations presented in this paper to avoid difficulties in surgery and in postoperative care (Table 1).

Finally, patients should also be closely followed to evidence any complications such as infection and malunions. We propose patients should be followed at 1 week, 1, 3, 6, 9 and 12 months. By doing so, the orthopedic surgeon may detect complications as soon as they appear and can therefore adjust treatment as needed.

Conclusions

The approach on patients with osteopetrosis frequently starts with the orthopedic surgeon's initial assessment because most cases are first manifested as a fracture. Recommendations for fixation include: an adequate operative planning, request of appropriate materials to facilitate drill insertion, decrease drill speed, availability of bone substitutes, and techniques to decrease temperature and secondary intraoperative fractures. The patient should also have a close follow-up with additional evaluation by other specialists. After treating and having reviewed the current literature, we conclude that treatment in osteopetrosis is challenging and the recommendations to enhance results are imperative to obtain successful results.

Conflict of interest The authors report no conflicts of interest.

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