Clin Orthop Relat Res (2008) 466:2002–2008
DOI 10.1007/s11999-008-0256-x

CASE REPORT

Case Reports

Treatment of Subtrochanteric and Ipsilateral Femoral Neck Fractures in an Adult with Osteopetrosis

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Received: 16 November 2007 / Accepted: 1 April 2008 / Published online: 23 April 2008

Abstract We describe a patient with autosomal-dominant osteopetrosis, a subtrochanteric fracture, and an ipsilateral femoral neck fracture treated with a hip spica cast Although the fracture united with coxa vara and external rotation deformities, the patient successfully returned to his normal activities of daily living. Operative fracture treatment in patients with osteopetrosis is difficult, and our patient provides evidence that with nonoperative treatment these patients can return to a functional level when operative treatment is not an option.

Introduction

Albers-Schoenberg first described his eponymal description of marked radiographic density of the bones ("hypersclerotic") [2] in 1904. Approximately 22 years later Karshner referred to the entity as osteopetrosis [26]. Despite the sclerotic radiographic appearance of the thickened cortices and its material hardness, osteopetrotic bone is weak and prone to fracture by minor trauma [5, 13, 29]. Areas of concentrated stress such as the femoral neck and subtrochanteric areas are especially susceptible. Most literature regarding treatment of osteopetrotic fractures concentrates on that of children or on the difficulty of operative intervention in adults [3, 5–7, 12, 13, 16, 17, 21, 22, 27, 29, 31, 35, 38, 43]. We report the case of an adult patient with autosomal-dominant osteopetrosis and a subtrochanteric fracture with an ipsilateral femoral neck fracture treated nonoperatively with a hip spica cast.

Case Report

A 56-year-old man with known autosomal-dominant osteopetrosis sustained a left subtrochanteric fracture of the hip (Fig. 1) after experiencing pain and spontaneously collapsing to the floor while walking. As a young adult, he had worn a cast for a contralateral tibia fracture and had cranial tong traction for a cervical spine fracture. At the time of his hip fracture, he was having dental care for fulminant mandibular osteomyelitis and psychiatric treatment for severe bipolar disorder. The patient was admitted to the hospital for tibial pin traction to await surgery after definitive treatment of his mandibular infection. Traction pin insertion required a power drill because of the hardness of the bone. Good alignment of the fracture was obtained with the traction. A preoperative computed tomographic (CT) scan of the hip was obtained to confirm the presence of an additional nondisplaced femoral neck fracture (Fig. 2) and to assess the femoral canal size.

Despite successful dental treatment, the patient refused surgery for his fracture, including external fixation. After 6 weeks in traction, he agreed to application of a one-and-a-half spica cast. Although radiographs showed minimal callus formation, there was no motion at the fracture site. The patient was discharged to home. With the assistance of home physical therapy, he learned to walk with a walker bearing partial weight on the affected extremity.

Each author certifies that he or she has no commercial associations (eg, consultancies, stock ownership, equity interest, patent/licensing arrangements, etc) that might pose a conflict of interest in connection with the submitted article.

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Fig. 1 An anteroposterior view of the pelvis obtained at the time of injury shows a displaced subtrochanteric fracture and osteosclerotic appearance of the osteopetrotic bone.

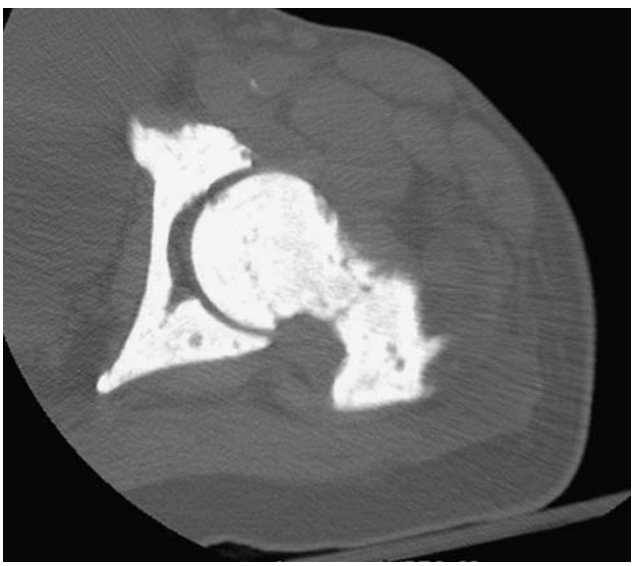

Fig. 2 A CT scan of the left hip and femoral neck shows a nondisplaced fracture of the femoral neck.

Five months after the fracture, plain radiographs showed moderate callus formation with some translation at the fracture site. A CT scan showed some bridging of the subtrochanteric and femoral neck fractures (Fig. 3). The cast was changed to a long leg cast brace with a waist extension, and the patient was able to bear full weight. Approximately 7 months from the injury, the cast was discontinued. Two and a half years after injury, the subtrochanteric fracture was united and in slight varus and external rotation (Fig. 4). The patient has a mild limp but no pain and is able to walk with only occasional use of a cane for long distances. Evaluation with the Short Form-36 produced the following subscale scores: Physical function 65, Role-physical 75, Bodily pain 78, General health 75, Vitality 85, Social 88, Role-emotional 100, and Mental health 86. The reported health transition score was 100.

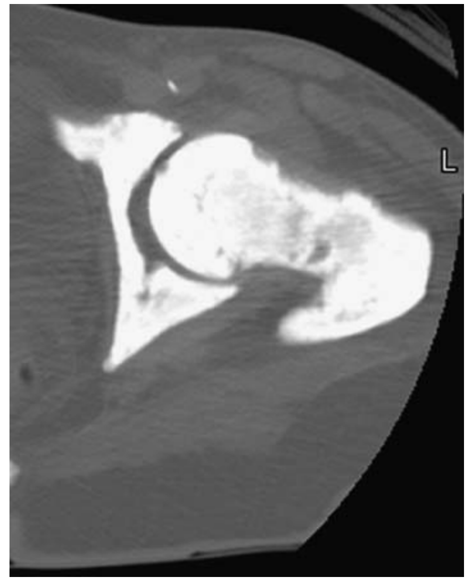

Fig. 3 A CT scan obtained 5 months after spica cast placement shows the healed left femoral neck fracture.

Fig. 4 An anteroposterior view of the pelvis obtained two and one-half years after the initial injury shows a healed left subtrochanteric fracture with coxa vara and external rotation deformities.

Discussion

Osteopetrosis is a disease of osteoclasts that results in failure of bone remodeling [40]. Histologically, mature osteopetrotic fracture callus contains no Haversian organization and has a paucity of osteoclasts [17]. Although the mechanical strength of osteopetrotic bone is decreased, at least one study in rats suggests there is no difference in the elastic modulus or hardness as compared with normal bone [24].

Osteopetrosis has been characterized clinically into three groups: infantile-malignant autosomal-recessive, intermediate autosomal-recessive, and autosomal-dominant [32]. The first form is usually fatal in childhood, and patients with the second form have a moderately decreased life expectancy, whereas those with the autosomal-dominant form generally have a normal lifespan [11]. The autosomal-dominant form is identified radiographically by universal osteosclerosis and has been divided into two subtypes based on the location of this osteosclerosis [10, 11]. Osteosclerosis of the cranial vault is evident with the first type, whereas end-plate thickening of the vertebrae (Rugger-jersey spine) and endobones (bone within bone) in the pelvis are evident with the second type. The second type is associated with increased fracture frequency in the autosomal-dominant population [10, 11]. The diagnosis of osteopetrosis in this group most commonly occurs when patients are being evaluated for a hip fracture [9]. The Type II autosomal-dominant form is that which Albers-Schoenberg first described and is so named after him.

Although the exact etiology is not known, studies suggest osteopetrosis is linked to three different genetic mutations affecting the osteoclast's ability to acidify bone during remodeling: one involving a subunit of the osteoclast vacuolar proton pump, a second involving the chloride channel associated with the proton pump, and a third involving carbonic anhydrase II [14, 18, 19, 28, 33, 34]. The chloride channel mutation appears associated with the autosomal-dominant form [14, 19, 34]. The mutation responsible for autosomal-dominant form Type II has been localized to two possible chromosomal locations, 1p21 and 16p13.3 [8, 41]. Others have suggested the genetic defect in osteopetrosis leads to failure to produce colony-stimulating factor 1, which regulates osteoclast development and differentiation [1].

Fractures occur frequently in patients with osteopetrosis, particularly in patients with the autosomal-dominant form [10]. The treatment of these fractures has been described in the literature mostly in the form of case reports and small case series (Tables 1, 2). This body of evidence repeatedly reports the difficulties associated with operative treatment, including the extreme hardness of the bone, which impedes drilling and cutting, hardware failure, periprosthetic fractures, coxa vara deformity, delayed union, pseudarthrosis, refracture, and periprosthetic infection [3, 5, 6, 12, 13, 17, 21, 27, 29, 35, 38, 43]. When comparing all forms of operative treatment in studies that report time to union, the average time to union is approximately 26 weeks. Conversely, of the five studies that describe nonoperative treatment of 35 osteopetrotic femur fractures, pseudarthrosis developed in one patient, another patient underwent placement of several pins to treat a nonunion, and three patients had coxa vara develop; the remaining 30 patients achieved union with no reported delays or complications [3, 5, 16, 21, 22].

One of the earliest reports of nonoperative osteopetrotic fracture treatment was by Alexander in 1923 [3]. He described treatment using a plaster cast for a peritrochanteric fracture in a 43-year-old woman; pseudarthrosis developed in this patient. Hasenhuttl reported treating a 27-year-old man with a peritrochanteric fracture with Russell traction in which the fracture united at 10 weeks [22]. Dahl et al. treated two patients, between 18 and 22 years of age, with peritrochanteric fractures with plaster cast immobilization in which the fractures united [16]. Armstrong et al. described nonoperative treatment of four peritrochanteric fractures in patients between the ages of 8 and 30 years [5]. Two patients were treated nonoperatively, without specific description of the method, and their fractures united. Another patient, described as a teenager, was treated with a hip spica cast, and the fracture healed without complication. The final patient, a 14-year-old girl, was treated with traction for 10 weeks and the fracture united by 16 weeks [5]. Gupta and Gupta treated a peritrochanteric fracture in a 31-year-old man with traction, and the fracture united by 16 weeks [21].

Surgery for osteopetrotic peritrochanteric fractures, however, is associated with considerable difficulty and complications [3, 5–7, 12, 13, 16, 17, 21, 22, 27, 29, 31, 35, 38, 43]. Kleinberg described the treatment of a peritrochanteric fracture with a plate, screw, and cortical strut allograft [27]. The plate broke and the fracture site became angulated but the fracture united [27]. Yang et al. attempted to treat a peritrochanteric fracture in a 21-year-old woman with a Jewett nail, but placement failed [43]. The fracture then was fixed with three screws and a hip spica, which she wore for 4 months. Her course was complicated by hip contractures, which were released. Initial callus formation appeared at 6 months and the fracture united by 12 months [43]. Milgram and Jasty [29]. described treating a peritrochanteric fracture in a 52-year-old woman with a Holt nail plate. The nail and screws were difficult to place, but the fracture united after 2 years [29]. Ashby described surgical treatment of three patients with peritrochanteric fractures [6]. The first, a 49-year-old woman, was treated with a Zickel nail. This procedure was described as difficult taking 6 hours to complete, with extremely hard bone that was difficult to drill; also, there was no identifiable medullary canal. The distal segment was fragmented on nail insertion, and cerclage wires were placed. The patient achieved mobility but was lost to followup for 3 years. A radiograph at that time showed displacement and comminution of the fracture about the nail; it was removed and a THA was performed. Three years later, an incomplete periprosthetic fracture was discovered at the tip of the femoral stem, which was treated with partial weightbearing for 2 months; this healed by 10 months. The second patient, a 61-year-old woman, fractured her left hip and was treated with a nail-plate device. Union was achieved. Eight years later, she fractured her right hip, which also was treated with a nail-plate device. A deep infection developed requiring removal of the hardware. She then underwent a THA 8 months later but rehabilitation failed and she did not regain walking ability [6]. de Palma et al. treated a peritrochanteric fracture, in a 27-year-old man, with a Jewett plate [17]. The patient was bearing full weight by 2 months and the fracture united. At 1 year, the plate was removed and 2 months later, the femur fractured at the level of the distal hole. This was treated with a dynamic compression screw and the fracture healed [17]. Armstrong et al. treated five peritrochanteric fractures operatively, and all were treated with either a screw-plate or a nail-plate [5]. Placement of fixation devices was described as difficult, and in one patient, the reamer for the screw-plate was destroyed and was converted to a nail-plate. Nonunion occurred in one of these five patients [5]. Gupta and Gupta treated a peritrochanteric fracture in a 31-year-old man with a blade plate that united by 16 weeks [21]. Chhabra et al. described operative treatment of six peritrochanteric fractures in adults between 22 and 45 years of age [13]. Treatments included two with dynamic hip screws, three with Küntscher nails, and an intramedullary nail. Both dynamic hip screws failed, one became infected and was associated with a nonunion, and the other pulled out and was associated with a nonunion. Two of the fractures treated with Küntscher nails united, but in the third patient, the nail migrated, after which exchange nailing was performed and the fracture healed. The fracture treated with the intramedullary nail united [13].

There are fewer reports describing treatment of femoral neck fractures in adults. Armstrong et al. treated seven patients nonoperatively with nonweightbearing and all had coxa vara develop that subsequently was treated with a valgus osteotomy. In three patients, the fractures were treated with either pins or a compression screw and all united. One patient was treated nonoperatively with nonweightbearing but a nonunion developed that was treated with pins at 6 months, after which the fracture united [5]. Rolauffs et al. reported a 39 year-old man with a femoral neck fracture treated with multiple parallel screws [31]. Three of the four screws broke, the fracture settled into varus, and osteomyelitis of the femur subsequently developed. A resection arthroplasty was performed, and at the 6-year followup, the patient was full weightbearing and pain-free with a limp [31].

Operative treatment of osteopetrotic fractures typically is reported as difficult and associated with many complications [3, 5–7, 12, 13, 16, 17, 21, 22, 27, 29, 31, 35, 38, 43]. Twenty-five cases of open operative treatment of osteopetrotic fractures have been reported (four femoral neck fractures and 21 peritrochanteric fractures). In this cohort there is a 12% nonunion rate and a 12% infection rate. All infected fractures achieved union. In the peritrochanteric group, the rate of hardware failure is 29%, the rate of reoperation is 29%, and the incidence of periprosthetic fracture is 14% (Table 1). There is evidence that in this population the outcomes for operative treatment of femoral neck fractures [5, 31] have been moderately better than for peritrochanteric fractures [5, 6, 13, 17, 27, 35, 43]. Only one report describes use of an external fixator for an osteopetrotic fracture, but the good result suggests this may be a reasonable option [7]. Although fewer cases have been published, nonoperative treatment of these fractures is associated with fewer complications by direct comparison [3, 5, 16, 21, 22]. The most common complication of nonoperative treatment is coxa vara deformity (31% of cases), the majority of which are with femoral neck fractures. There is only one report of a nonunion in peritrochanteric fractures treated nonoperatively (Table 2). Coxa vara malalignment was not a clinically important complication in our patient, as he returned to a high level of functioning. The physical function subscale score (SF-36) is a reliable, valid, and responsive functional measure for patients with a hip fracture but would not likely distinguish between varus and nonunion although the two would likely have differing implications for subsequent treatment [23]. Our patient's physical function score was 65 at 2.5 years. The average physical function score for men aged 55–64 years is 80, which shows a steady decline with aging [39]. Although our patients' score is slightly below the average for his age group, it is substantially higher than the average physical function score of 24 for patients younger than 60 years with end-stage hip arthrosis [20].

Some published basic science research suggests 1,25 dihydroxyvitamin D (calcitriol) and parathyroid hormone can increase the number and function of osteoclasts in osteopetrotic mice [30]. There also is clinical evidence high-dose calcitriol may decrease the symptoms of osteopetrosis [32]. In addition, clinical data exist supporting use of recombinant parathyroid hormone to enhance fracture healing in general [4]. However in our patient, treatment of these fractures was not augmented by medical management.

Although our patient may have been treated operatively had he not refused surgery, the good functional outcome suggests nonoperative treatment should be considered as an option. The reported incidence of perioperative infection is

Table 1. Published studies of operative osteopetrotic femoral fractures in adults

Study	Age (years)	Gender	Fracture location	Treatment	Complications	Status at last followup
Kleinberg [27]	35	M	L, P	Plate and screw, cortical allograft	Plate breakage and angulation	Union
Yang et al. [43]	21	F	L, P	Jewett nail	Failed placement of nail with placement three interfragmentary screws and hip spica cast	Union at 12 months
	52	F	P	Holt nail plate		Unions at 2 years
Belz et al. [7]	46	F	L, S	Uniplanar external fixator 13 weeks		Complete union with removal at 13 weeks
Ashby [6]	49	F	L, P	Zickel nail	Fragmentation of distal fragment with nail placement, placed cerclage wires	Union
	52	F	L, P (3 years later)[†]	THA after nail removal		Full weightbearing
	55	F	R, P (3 years later)[†]	THA	Periprosthetic fracture 3 years later	Union at 10 months
	61	F	L, P (8 years prior)	Nail plate device		Union
	69	F	R, P	Nail plate device	Deep infection with hardware removal	Nonunion.
	70	F	R, P	THA after 8 months	Failed rehabilitation	Nonambulatory
de Palma et al. [17]	27	M	R, P	Jewett plate	Union, removed plate at 1 year, refractured, dynamic compression plate	Union
Armstrong et al. [5]	6–16 (N = 3)		FN	Pins/compression screw		Union
	6–16		FN	Nonweightbearing/pins	Nonunion treated with pins at 6 months	Union
	Adult (N = 3)		P	Nail plate/compression screw plate		Union
	8–30		P	ORIF		Nonunion
	8–30		P	ORIF		Union at 6 months
	31	M	P	Blade plate		Union at 12 to 16 weeks
Rolauffs et al. [31]	39	M	R, FN	Parallel screws	Hardware failure, osteomyelitis, Girdlestone	Girdlestone
Su et al. [35]	29	M	L, P	ORIF		Union
Chhabra et al. [13]	22	F	L, P	DHS revision after prior Jewett nail	Infection	Infection, nonunion
	22	F	R, P	DHS	Hardware failure	Pullout, nonunion
	41	F	L, P	Kuntscher nail	Rod migration with exchange rodding	Union 2 months, rod removed at 6 months
	42	F	L, P[†]	Kuntscher nail		Union
	45	F	R, P (3 years later)	Kuntscher nail		Union
	42	M	R, P	Proximally locked intramedullary nail		Union at 2 months

[†] Refracture; F = female; M = male; R = right; P = peritrochanteric; L = left; S = femoral shaft; FN = femoral neck; ORIF = open reduction internal fixation; DHS = dynamic hip screw.

Table 2. Published studies of nonoperative osteopetrotic femoral fractures in adults

Study	Age (years)	Gender	Fracture location	Treatment	Complications	Status at last followup
Alexander [3]	43	F	R, P	Nonoperative: plaster cast	Pseudarthrosis	Nonambulatory
Hasenhuttl [22]	27	M	P	Russell traction		Union at 10 weeks, ambulatory
Dahl et al. [16]*	18–22 (N = 2)	F	P	Plaster splint/cast		Union, ambulatory
Armstrong et al. [5]	6–16 (N = 3)		FN	Nonweightbearing	Coxa vara deformity treated with a valgus osteotomy	Union, ambulatory
	8–30 (N = 2)		P	Nonoperative		Union, ambulatory
	Teenager	M	P	Hip spica		Union, ambulatory
	14	F	P	Traction 10 weeks		Union at 16 weeks, ambulatory
Gupta and Gupta [21]	31	M	P	Traction		Union at 12 to 16 weeks, ambulatory
Current study	56	M	L, P and FN	Hip spica cast	Coxa vara deformity	Union at 7 months, ambulatory

*Patients with malignant autosomal-recessive osteopetrosis; F = female; M = male; R = right; P = peritrochanteric; L = left; FN = femoral neck.

12% and led to nonunion in all cases [5–7, 13, 17, 27, 31, 35, 43]. Patients with a known history of osteomyelitis, a common ailment in patients with osteopetrosis, would likely be at higher risk for infection [25, 42]. In addition, the operative complications of periprosthetic fracture and hardware failure carry substantial indirect potential morbidity including additional soft tissue injury and reoperation. Open reduction and internal fixation of a peritrochanteric fracture puts the nondisplaced femoral neck fracture at risk for displacement and vascular compromise, unless it also is stabilized internally [36, 37]. Placing screws across the femoral neck introduces an additional potential site for a perioperative complication [5]. Clinkscales and Peterson suggested the overall monetary cost of treating patients with casting was less than the cost for external fixation or intramedullary nailing of femoral fractures, after an average followup of 4 years [15].

In complicated osteopetrotic ipsilateral femoral neck and proximal femur fractures in which internal fixation would be difficult or not an option, closed treatment initially with traction and then with a hip spica cast may lead to a good functional outcome without the risks of infection, hardware failure, periprosthetic fracture, and the potential for additional fracture displacement.

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