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Original Research

Management and Outcomes of Femur Fractures in Patients with Duchenne Muscular Dystrophy

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Abstract

Background: Duchenne muscular dystrophy (DMD) is a severe, progressive X-linked recessive neuromuscular disorder characterized by muscle weakness and atrophy. Additionally, patients with DMD have significant reductions in bone mineral density compared to age-matched controls, which is exacerbated by concomitant steroid use. These findings dramatically increase fracture risk, which may irreparably decrease functional status. The aim of this case series is to examine outcomes of operative versus nonoperative management of femur fractures in this patient population.

Methods: An IRB-approved retrospective chart review was completed for patients with DMD treated at a single institution for a femur fracture between 2013-2022. Patients were excluded for incomplete documentation, treatment initiation at an outside hospital, or diagnosis of a different muscular dystrophy. Demographic variables, treatment information, functional status, and adverse events were collected for each patient. Descriptive statistics were used to summarize demographic and outcome variables.

Results: A total of 10 patients with 11 femur fractures were included for analysis. All patients were male with an average age of 12.7 years and clinical follow-up of 286 days. Five fractures in five patients underwent operative fixation (**Group A**), and six fractures in five patients underwent nonoperative management (**Group B**). In Group A, three patients were short-distance ambulators prior to injury, and all patients regained a similar functional status postoperatively. All three patients were treated with a locked intramedullary nail. One patient in Group B was a short-distance ambulator prior to injury; the remainder were nonambulatory. All patients in Group B were primary wheelchair users at final follow-up. There were no adverse events as a result of treatment in either group.

Conclusion: Nonoperative management with cast immobilization remains an acceptable option for nonambulatory patients and those with minimally displaced fractures not amenable to surgical intervention. Surgical intervention is recommended for higher-functioning patients with the goal of restoring ambulatory status. Regardless of treatment modality, patients should receive aggressive physical therapy directed at early weight-bearing, range of motion, and mobilization to preserve strength, muscle mass, and mobility.

Level of Evidence: Level IV case series

Key Concepts

- Management of femur fractures in Duchenne muscular dystrophy should account for fracture morphology as well as patient functional status.
- Surgical management is safe and effective in promoting fracture union and preserving functional status.
- Nonoperative management is preferred in nonambulatory patients and for minimally displaced fractures.

Introduction

Duchenne muscular dystrophy (DMD) is a severe, progressive neuromuscular disorder inherited in an X-linked recessive pattern. It is characterized by muscle weakness and atrophy that affects an estimated 1/3600 male births.[1] The progressive muscle wasting is due to a mutation in the dystrophin gene, which normally functions as a bridging protein between the plasma membrane of myofibrils and the extracellular glycoprotein matrix to stabilize the muscle during contraction.[2,3] The phenotypical traits are not present at birth, and the disease is typically diagnosed at age four.[4] As the disease progresses, untreated individuals typically become nonambulatory in their early teens and have significant decline in cardiopulmonary function by their late teens.[5]

In addition to cardiopulmonary complications, patients with DMD have a significant reduction in bone mineral density compared to age-matched controls given their relatively limited weight-bearing and the decrease in forces acting across the long bones. Moreover, as glucocorticoids (GC) become a mainstay of treatment to improve motor strength and slow disease progression, affected individuals are at even higher risk for osteopenia and osteoporosis given GC upregulation of osteoblast apoptosis, osteoclastogenesis, and increased bone turnover.[6-8] This constellation of findings places these patients at a significantly increased fracture risk–specifically in the lower extremity.[6] Fractures of the lower extremity pose a therapeutic challenge as prolonged immobilization and/or restriction of activities may irreparably decrease a patient's function and accelerate a decline in ambulatory status.

Nonoperative treatment with casting is the historical treatment modality of choice for long bone fractures in this patient population, but casting requires a prolonged duration of immobilization and poses high risk for malunion and cast-associated pressure ulcers compared to operative treatment.[9] Operative intervention with intramedullary nailing (IMN) has gained popularity among surgeons caring for patients with neuromuscular disorders, credited for decreasing immobilization time, malunion rates, and permitting early mobilization.[10,11] Flexible intramedullary nailing (FIN), commonly employed with titanium or stainless-steel intramedullary nails, necessitates a length-stable fracture pattern and is relatively contraindicated in patients weighing more than 100 lbs.[12] While rigid trochanteric nails do not have such restrictions, they are associated with the rare risk of

femoral head avascular necrosis, and smaller size options appropriate for this patient population are not always readily available.[13]

Due to limited high-quality data and combined reporting of all muscular dystrophies, controversy remains over the optimal treatment for patients with DMD presenting with a femur fracture. The aim of this case series is to examine outcomes of surgical intervention versus nonoperative management of femur fractures in patients with DMD specifically.

Materials and Methods

An IRB-approved retrospective review of patients with DMD seen for femur fractures between 2013-2022 was conducted. Patients were excluded for incomplete documentation, treatment initiation at an outside hospital, or diagnosis of a different muscular dystrophy. All records were independently reviewed by two authors for all data collection.

Variables for analysis included patient demographics (including age at injury), steroid use, ambulatory status, mechanism of injury, and fracture laterality. Radiographs were reviewed by the lead author, and fractures were classified as femoral neck, intertrochanteric, subtrochanteric, shaft, supracondylar, or distal femur.

Patients were subdivided into those whose fractures were managed with surgery (**Group A**) and those who were managed nonoperatively (**Group B**). Relevant evaluated treatment-based information included treatment modality, weight-bearing status, and duration of immobilization. Pre- and post-treatment functional status was captured for each patient. Complications were defined as malunion, nonunion, intraoperative fracture, femoral head avascular necrosis, and failure to wean mechanical ventilation within 24 hours of surgery.

Descriptive statistics were used to summarize demographic and outcome variables in each group. Fisher's Exact Test was used to compare observed frequencies between the two groups. Statistical significance was defined as $p<0.05$.

Results

A query of the medical record yielded 23 patients. Ten of these patients were excluded for no clear diagnosis of DMD, one patient was excluded for diagnosis of spinal muscular atrophy, one patient was excluded for a diagnosis of a Collagen VI-related dystrophy and one patient was excluded for undergoing diagnosis and treatment at an OSH. After application of exclusion criteria, a total of 10 patients with 11 femur fractures were included for this series. All management decisions were left to the discretion of the treating orthopaedic surgeon. Of this cohort, five fractures in five patients underwent operative fixation (**Group A**), and six fractures in five patients were treated nonoperatively (**Group B**).

In Group A, all patients were male. The mean age at fracture was 11.8 years old (SD: 1.5 years). Mechanism of injury varied in this group and are listed in Table 1 along with a summary of fracture morphology and perioperative functional restrictions. Four (80%) of these patients were on chronic systemic steroids at baseline. Prior to injury, three (60%) of these patients were short-distance ambulators. All patients in Group A returned to their baseline ambulatory status postoperatively. Of the three patients that received treatment with a locked IMN, all were done with a lateral-entry nail to preserve blood supply to the femoral head. There were no adverse events as a result of treatment noted, and all fractures went on to radiographic union at final follow-up.

In Group B, all patients were male. The mean age of injury was 13.6 years old (SD: 2.2 years). All injuries were the result of a low-energy fall from sitting and were managed with non-weight-bearing long leg casts or knee immobilizer (Table 1). Four (80%) of these patients were on chronic steroid therapy at baseline. Four (80%) of these patients were nonambulatory prior to injury and remained nonambulatory at latest follow-up. One patient was ambulatory prior to fracture. This patient failed to regain his baseline ambulatory status postoperatively. There were no complications associated with immobilization, and all fractures demonstrated evidence of healing at final follow-up.

Table 1. Cohort Characteristics & Treatment

	Fracture Age	Injury Mechanism	Fracture Location	Treatment	Immobilization Duration	Weight- Bearing Restrictions	Pre-Injury Functional Status	Post-Injury Functional Status	Age at Last Ambulation
Group A									
1	14	Fall from scooter	Left IT	Locked IMN	None	50% WB x2 weeks	SDA	SDA	-
2	11	Motor vehicle accident	Right femoral neck	CRPP	None	NWB x7 weeks	WD	WD	10
3	12	GLF	Right femoral shaft	Locked IMN	None	50% WB x3 weeks	SDA	SDA	-
4	12	No discrete injury	Right femoral neck	CRPP	None	None	WD	WD	8
5	10	GLF	Right segmental femoral shaft & supracondylar	Locked IMN & CRPP	6 weeks in knee immobilizer	NWB x6 weeks	SDA	SDA	11
Group B									
6	16	GLF	Right supracondylar buckle	Knee immobilizer	5 weeks	NWB x5 weeks	WD	WD	11
7	12	GLF	Left supracondylar	Spica cast	4 weeks	NWB x4 weeks	SDA	WD	12
8	12	Fall from wheelchair	Right supracondylar buckle	Long leg cast	6 weeks	NWB x6 weeks	WD	WD	10
9	12	Fall from wheelchair	Left supracondylar	Hinged knee brace	6 weeks	NWB x3 weeks	WD	WD	11
10	16	Fall from wheelchair	Bilateral supracondylar	Long leg cast	6 weeks	None	WD	WD	9

GLF: ground level fall; IMN: intramedullary nail; CRPP: closed reduction and percutaneous pinning; IT: intertrochanteric; WB: weight-bearing; NWB: non-weight-bearing; SDA: short distance ambulator; WD: wheelchair-dependent.

Physical therapy was initiated for every patient on postoperative or post-casting day #1 while an inpatient. The physical therapy protocol was tailored to each patient based on their physical limitations and weight-bearing restrictions. Outpatient physical therapy was prescribed for each patient upon discharge and records were reviewed for each patient, when available, to assess functional status.

Across both groups, patients had an average radiographic follow-up of 221 days (SD: 278 days) and an average clinical follow-up of 286 days (SD: 466 days). All fractures went on to clinical and radiographic union without evidence of clinically relevant malunion. While there were no complications associated with orthopaedic treatment of femur fractures, Patient #10 in Group B with bilateral femur fractures suffered a fat embolus and resultant acute respiratory distress syndrome (Table 1). During placement of a central venous catheter, the aorta was inadvertently cannulated, which required open surgical repair and veno-arterial extracorporeal membrane oxygenation (VA-ECMO) support. The patient was ultimately decannulated from ECMO but required persistent gastrotomy tube and tracheostomy support.

Discussion

Osteoporosis in the setting of DMD is nearly ubiquitous as patients age and may be accelerated by concomitant use of systemic oral steroids.[6-8,14] As demonstrated in this case series, DMD patients are at increased risk of fracture throughout the femur, often resulting from low-energy mechanisms. Treatment of these fractures predominately focuses on preserving function in these patients while minimizing morbidity associated with surgery and/or immobilization. Data surrounding these treatment options is limited to small case series with heterogenous patient populations often including patients with differing myopathies.[15-19] To the authors' knowledge, this is the largest series of exclusively DMD patients that compares surgical versus nonsurgical management of femur fractures.

Our series demonstrates that both operative and nonoperative management are acceptable treatment modalities in this patient population. Operative management may be more suitable for ambulatory patients to maintain functional status, as stable surgical fixation allows for earlier mobility and early weight-bearing. Regardless of treatment modality, physical therapy is integral to the treatment of femur fractures in patients with DMD to maintain and restore function post-fracture. All patients in this series received inpatient physical therapy as well as a prescription for outpatient therapy; however, there was no standardization of therapy modalities or treatment duration. Physical therapy (PT) protocols allow for a dynamic treatment plan for patients and families during recovery as patients progress through all levels of activity.[20] This may include land-based or water-based therapies depending on cast and wound status. In ambulatory patients, PT is essential for progressing to safe weight-bearing and assisted ambulation. In minimally ambulatory and nonambulatory patients, disuse muscle atrophy can be delayed with appropriate early exercise.[21] Early integration of PT is essential to prevent joint stiffness and contracture, which limit functional mobility in ambulatory and nonambulatory patients. Further, early therapy allows for functional, safe activity progression within restrictions.[22]

As seen in this case series, minimally displaced or incomplete fractures in the nonambulatory child can successfully be managed nonoperatively with a period of immobilization. Despite the prevalence of osteopenia, all patients in this study had evidence of successful healing at final follow-up. The success of nonoperative management in this series echoes similar reports in the literature.[18,23] Only one patient in Group B, however, was ambulatory prior to injury and did not achieve ambulatory status at final follow-up. As such, the utility of this treatment approach in the ambulatory patient remains uncertain.

Treatment with operative intervention in this series was reserved for patients who were ambulatory prior to injury and/or had displaced fractures of the femoral neck that threatened the vascularity of the femoral head. All three operative patients in Group A (patients #1, #3, and #5)

who were preoperative ambulators regained a similar level of function postoperatively. These three patients were managed with an initial period of partial weight-bearing in order to allow for early fracture healing prior to full weight-bearing. This limited restriction in weight-bearing, however, did not prevent return of function. Of note, patient #5 in Group A had subsequent deterioration in his function status roughly 1 year postoperatively, and he primarily utilized a wheelchair for mobility thereafter. This was thought to be unrelated to his fracture.

Given the retrospective nature of the study, we could not control for fracture morphology or location between patient groups. As such, there were notable discrepancies between Groups A and B. In this series, fractures of the femoral neck and shaft were more likely to undergo operative intervention, whereas all supracondylar fractures were managed nonoperatively, the majority of which occurred in nonambulators. This inherent selection bias limits the generalizability of this study's conclusions; however, these results demonstrate both the safety and efficacy of operative intervention in this patient population, regardless of ambulatory status. As such, we believe that operative intervention should be considered for more proximal fractures despite ambulatory status to prevent contractures and allow earlier facilitation of seated positioning. Further research is needed to conclusively define optimal treatment protocols based on fracture location and ambulatory status in this patient population.

This series has multiple limitations. As a small study, it is not appropriately powered to detect significant differences between Group A and B. As stated above, the retrospective nature of the study prevented prospective treatment allocation. As such, patients with minimally or nondisplaced fractures as well as those with a lower baseline functional status were more likely to receive nonoperative treatment. In addition, utilization of physical therapy protocols were not standardized amongst patients in this series, which limits analysis of its effectiveness.

In conclusion, management of femur fractures in patients with DMD should involve a multidisciplinary approach focused on restoring each patient's functional status. Nonoperative management with cast immobilization is suitable for nonambulatory patients and those with minimally displaced fractures not amenable to surgical intervention. However, nonambulatory patients are not precluded from operative management, and this option should be considered in fracture patterns that are amenable to surgical fixation to allow for earlier range of motion and expedited return to baseline mobility. Surgical intervention is recommended for patients with higher levels of ambulation with the goal of restoring baseline functional status. Regardless of fracture management, all patients should receive physical therapy to preserve strength, muscle mass, and range of motion, which are all directly related to functional level.

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