“Management of Femur Shaft Fracture with Nancy Nail in the Setting of Dystrophic Epidermolysis Bullosa”: A case report

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Abstract

Background

The management of bone fractures must achieve both the reduction and stability provision. However, dermatological conditions such as dystrophic epidermolysis bullosa (DEB) for instance can lead to catastrophic events when operating on the patient’s bone fracture. This can lead to wound infections and possible failure of bone healing leading to fracture nonunion. This dermatological disorder leads to heterogenous bullous dermatoses including cutaneous fragility leading to cutaneous bullous formation after exposure to any type of trauma. DEB is a rare inherited form of the disease characterized by the formation of cutaneous bullae. DEB is associated with a genetic mutation of COL7A1 gene that encodes collagen type VII. Due to the rarity and uniqueness of the disease, special modifications due to the challenges faced during the patient care approach were accomplished to prevent any possible harm to the patient. In this study, we propose a case report that is followed by the anesthetic and surgical challenges faced and how they were modified upon.

Case Presentation

A 20-year-old female presented to the emergency department with a spiral mid-diaphyseal fracture of the femur after an incidence of falling from bed. The patient was previously diagnosed with dystrophic epidermolysis bullosa which made this case unique and complex. As the patient was examined by the team, her skin was covered with old blisters and wounds that have developed with the consequence of the disease. Due to the sophisticated dermatological condition and the unique presentation, the established multidisciplinary team took a decision to treat the patient with flexible intramedullary nailing in an open versus closed reduction technique, and modifications of the treatment approach were done based on the challenges in this case. The goal was to provide the management while minimizing the risk of infections and complications that would have arisen. The proposed case will set a baseline for the management of similar cases.

Conclusions

We suggest that in order to manage femur shaft fracture in the setting of dystrophic epidermolysis bullosa, modifying the management to avoid the least possible skin harm at any expense while managing the bone fracture is the golden approach.

Case Presentation

A 20-year-old female with a background history of DEB has presented to our institution’s emergency room department due to an incidence of a fall from bed. Consequently, a sustainment of a spiral mid-diaphyseal fracture in the right femur has occurred. Upon initial primary examination in the emergency department, the patient was fully conscious, oriented with a Glasgow Coma Scale of 15/15, pale, and cachetic in appearance. Moreover, the patient's physical appearance was not suitable for her current
chronological age. Furtherly, the patient was vitally hypotensive in a third-grade hypovolemic shock. The Advanced Trauma Life Support protocol was initiated, and 2-liters of Ringer Lactate were started along with antibiotics and analgesia that was administered in the form of Paracetamol although an intravenous access was extremely sophisticated to be sought due to the present skin condition. Afterwards, the patient moved to a state of transient normotensive shock. Severe pain and an obvious deformity of the right femur were notable. However, no observable wounds or new skin blisters were noted. Thereby, it was a closed femur fracture.

Due to the situation, the managing team was not capable of performing skin traction due to possible skin sloughing and catastrophic deterioration of the condition.

The patient's past medical history was positive for DEB that was managed and followed up by a dermatologist where daily dressing was prescribed to be applied on a daily basis. Dilated cardiomyopathy and anal fissures were mentioned to be very painful and kept the patient from eating normally due to the fear of gaining more pain, severe malnutrition that has not been managed with a gastrostomy tube or nutritional supplements, and iron deficiency anemia. Additionally, the patient had complained of on-and-off abdominal cramps, severe constipation, epistaxis, difficulty swallowing, adhesive tight tongue, unexplained shortness of breath, and dental caries despite brushing the teeth. However, no constitutional symptoms, or current infections were instantly present. Furthermore, there were no previous similar trauma events or surgeries upon patient's history but a hospital admission 10-years ago due to shortness of breath was reported and 3-units of blood were given due to a hemoglobin level of 5 gm/dL at that past setting.

Family history was positive for DEB in two of the siblings. However, the eldest male child who was diagnosed with DEB since birth had died in the age of ten-years due to severe malnutrition and accompanying iron deficiency anemia. The other male sibling who was also diagnosed to have DEB had also died at the age of five-years. The youngest female sibling in the other hand had died in the age of 16-days and the cause of death was not established. In addition, the mother has reported that the older two female siblings were healthy with no current medical illnesses.

On clinical examination, scoliosis was evident along with the right thigh deformity and flexion contracture of both hip joints in an approximation of 20 degrees. In addition to that, both knees were noted for flexion contracture in about 40 degrees with both feet being equinus and showed “mitten” appearance. The right thigh has scattered healed wounds due to the skin condition from all the proximolateral aspect up to the middle region of the thigh. However, the distal thigh skin was noted for several unhealed wounds all over the limb aspects. The range of motion in both of the lower limbs were normal up to the contractures. Examining the hands, flexed syndactyly in both due to the old skin blisters and wounds as it is the nature of DEB and mitten appearance were also noted on the inspection. The distal pulses were intact and motor and sensory components of a conducted neurological exam were scored as 5/5 and there was no evidence for signs of compartment syndrome.
**Investigations**

During the primary emergency department management, anteroposterior and lateral views of the pelvis and right hip X-rays were obtained demonstrating femur spiral mid-diaphyseal fracture. *(Figure.1-4)* Routine complete blood count showed iron deficiency anemia with a hemoglobin level of 2.5 mg/dl. Other laboratory investigations included the INR (1.2), PT (14.9), PTT (35.3), Albumin (7 g/L), Creatinine Kinase (46), Calcium (1.83 mmol/L), Potassium (3.1 mEq/L), Sodium (135 mEq/L), Phosphorus (1.35 mg/dl), Creatinine (23 mg/dl), BUN (1.4 mg/dl). Due to the previously mentioned past medical history of dilated cardiomyopathy and in preparation for the management, an echocardiogram was done to show an ejection fraction of 48% and a systolic murmur was noted.

**Differential Diagnosis**

This case scenario was a typical presentation of femur fracture. This conclusive diagnosis was approached by conducting a detailed history, thorough clinical examination, laboratory investigations, and radiographic imaging. All the flow approach steps have guided to a diagnosis of spiral mid-diaphyseal fracture.

**Treatment**

*Preoperative:*

The patient was admitted to the in-patient ward where she received 3-units of whole blood and albumin. Due to the unique case, the literature review has shown a gap regarding the treatment of femur fracture in a case that is complicated by the background of DEB, thereby, the team has discussed the case thoroughly to approach the patient through a method that is unharmed to the skin to save the patient at any cost. However, the managing team have found that the most suitable surgical treatment in this setting is the use of Nancy nail which is a flexible intramedullary nail with the aid of Steinmann pin in the closed versus open reduction and internal fixation for the femur fracture. The open reduction was not a possible treatment option due to the DEB condition. We have chosen this treatment path to avoid traumatizing the skin with countertraction, decrease the potentiality of infection development, and reduce the risk of skin sloughing. The team has discussed the management plan with the patient and her family with listing all the pros-and-cons as well as the list of possible intraoperative and postoperative complications. The list of complications that could have arisen included wound infection, delay in the healing process, risk of bleeding, iatrogenic nerve injury, malunited bone, and delayed bone union. Eventually, a consent was signed to conduct the operation. Moreover, previous agreement on hospitalizing the patient 3-days prior to the day of surgery for preparation was done.

*Day of Surgery:*

The patient was evaluated to have a Malampati score of 4. In spite, she was at a high risk of possible esophageal perforation and difficult general intubation. Thereby, the team has agreed to do a spinal
anesthesia and admit the patient for postoperative intensive care unit. After prepping and draping, we have done two 3-cm longitudinal incisions in the distal medial and lateral right distal thigh. Afterwards, 2.5 mm and 3 mm Nancy nail were introduced and have used Steinmann pin during the closed reduction technique. Thereafter, the layers were closed in sequence with Vicryl suture and the application of wet dressing. It is important to mention that the surgical team faced the challenge of dealing with fragile bone during the operation as all the steps had to be performed very delicately to avoid any harm. However, with the option of Nancy nail, the team was capable of managing with the fracture. The patient was admitted to the intensive care unit for close monitoring for one day followed by 2-days in the burn unit to continue the close monitoring and special skin condition treatment. Along with the skin dressing, antibiotics, analgesics, and parenteral feeding with electrolytes and albumin correction were done. Fortunately, the patient was discharged home without any of the complications that include adhesions, wound infection, skin sloughing, or blistering, and the patient’s family were informed about all the needed considerations to take optimum care of the patient.

Outcome And Follow-up

The wound was completely healed with no ulcers, sinus tracts, or infection around the surgical wound. (Figure.5-6) Two months postoperatively, the patient was seen for follow-up; an x-ray was done and has shown an ingoing healing femur that was incomplete, with the complete heal established only after 18 weeks. (Figure.7-8) Although the continuous rehabilitation program was performed, the patient did not gain her weight bearing until after 8 months of rehabilitation including range of motion, stretching, and strengthening program exercises. Moreover, she was not able to weight bear because of the pain and contracture of the knee and achilles tendon. The patient started walking with walking aids almost 4 months postoperatively, but she did not gain all her weight bearing capability with the accompanied decrease in the range of motion compared to her previous state before the event. The patient may need another operation to correct her femur mechanical access because she gained a deformity that affect the ability to walk. This deformity occurred as result of the fracture pattern and the deforming force of the muscle of the thigh. The aim was to regain the length, correct the rotation, and prevent the sagittal and coronal deformity as possible. However, we did not achieve the anatomical reduction because the team wanted to prevent gaining more traumatic injury to the soft tissue in the form of sloughing to the skin as mechanical trauma or establishing an iatrogenic fracture. Due to the physis not being completely fused, the body weight according to her chronological age, the skin condition because of the nature of the disease, all have affected the decision of the surgery type and the instruments chosen. The plate and screw use with closed reduction could not be used due to the huge open approach in the lateral thigh in the presence of old scars and new ulcers and blisters increasing the potential risk of infection and the risk of delayed wound healing which may furtherly lead to soft tissue bleeding, fibrosis, and adhesions or hematoma. Moreover, the diameter of the canal was very narrow and that justifies why we have chosen the Nancy nail. In choosing the best treatment modality for this patient, we considered that we need an acceptable reduction and to initiate rapid rehabilitation. During the preoperative, operative, and the postoperative areas of treatment, the team has handled the case in a very
considerable manner. The soft tissue in the area of the fracture was well taken care of in spite of the critical skin condition and the risk of potential infections.

**Discussion**

A description of children's skin who suffer of epidermolysis bullosa is often referred to as “Butterfly children” as their skin is as vulnerable as the wings of a butterfly. (1) Epidermolysis bullosa is an entity of heterogenous bullous dermatoses including cutaneous fragility leading to cutaneous bullous formation after exposure to trauma despite how minimal it is. Dystrophic epidermolysis bullosa (DEB) is a rare inherited form of the disease, characterized by the formation of bullae that further evolve to atrophic scars. DEB is associated with a genetic mutation of COL7A1 gene that encodes collagen type VII. (2) In turn, the sublamina densa layer of the basement membrane separates. (7) The estimated prevalence of DEB was reported to be 2-6 affected individuals per a million births and 1:1 male to female ratio.

Moreover, the symptom manifestation of DEB starts since birth. (3) Type VII collagen is responsible for the formation of a dermal-epidermal adhesion that is stable in nature as it is one of the building blocks of the anchoring fibrils. (4) The patterns of inheritance associated with DEB are autosomal dominant and recessive. Comparing both modes of inheritance, the dominant type often manifests at birth or during the period of early childhood and is described to form generalized blistering that becomes more localized with the aging process. However, the recessive type of DEB range in severity from mild to severe form. Feet and hands pseudosyndactyly; referred to as “boxing glove hands”, deformed nails and affected teeth, flexural contractures, esophageal obstruction due to the disease process affecting the mucosa, and iron deficiency anemia caused by the associated malabsorption are all manifestations of DEB. (5) Scar tissue formation is often the sequela of the chronic skin ulcers that occur in a recurrent cycle which can lead to major dysfunction. In addition, the recurrent skin lesions can cause squamous cell carcinoma on its run leading to mortality. (3) The optimum management of DEB is sophisticated, and no ultimate curative treatment is present instantly. However, the main concept followed in treating patients suffering from DEB is to treat the blisters and the skin wounds while preventing infection occurrence and other potential complications from taking place. As pain is a characteristic feature of DEB, managing the accompanying pain and itching will leave crucial impact and relieve a burden in the patient’s quality of life. Justifying upon, lancing the blisters, applying skin dressings that are featured to be non-adhesive are favorable skin treatments for the erosions while they apply pain relief simultaneously. Additionally, opioids such as morphine for instance were reported to relieve the pain associated with recessive DEB itself. (6) Back in 1968, Becker and Swinyard (8) were the first to find the association between DEB and reduced long bones mineralization and the manifestation of dental caries as they have reported four cases of DEB in children suffering from DEB. This in turn, rises the risk of developing all the osteopenia, osteoporosis, and pathological fractures as consequences. (7) Factors that play roles in the development of decreased bone mineralization in the setting of DEB include reduced intake and absorption of calcium, decreased overall production of vitamin D, all accompanied by existing malnutrition, reduced body mass index, and history of a fracture event, all lead to increased risk of osteoporosis occurrence as they are reported risk factors. (9) Moreover, the low hemoglobin levels found in DEB patients along with the
reduced plasma albumin are factors that increase the potential risk of low bone mass. (10) In order to monitor the bone status in this case; blood tests and bone radiographs along with monitoring the levels of calcium and vitamin D status are necessary. Unfortunately, the ultimate management of osteoporosis in the setting of epidermolysis bullosa is still a gap in the medical literature. However, additional nutritional support and accommodating weightbearing activities are all advantageous options. Pharmacological agent of therapy includes cholecalciferol as an option. (11) Regarding the femur spiral fracture management in our presented case, we have chosen to treat it surgically using Nancy nail which is a flexible intramedullary nail with the aid of Steinmann pin in the closed versus open reduction and internal fixation for the femur fracture. We have chosen this approach after a condense literature review and multiple benefit-and-risk outweighing discussions between the team members. We concluded that an open reduction was a possibly harmful surgical option due to the increased risk of infection and complications that could have arisen due to the DEB complex skin condition. Aksoy and colleagues (12) have noted their study that the flexible intramedullary nailing is associated with a shorter intraoperative duration and was found to have quicker healing process. In addition, introducing this nail requires only small incisions. (12) The small incisions needed to introduce the nail is an advantage we have needed in this case to preserve the skin and avoid harm. Lastly, Aksoy et al (12) have also noted that the postoperative period of immobilization related to the use of this nail is shorter. This key point is considered a benefit in facing the postoperative challenges. Lohiya et al (13) have described the flexible intramedullary nailing to have a reduced infection risk and promotes the process of bone healing while being inserted in a closed technique.

Patient Perspective

As a 20-year-old girl, I fear for myself that I will be identified by my fears. The fear of an upcoming skin infection or a wound that leaves me in pain. Although the pain of the Epidermolysis Bullosa is what I fear, the pain of not being able to be held by my close ones due to my skin condition associated pain is more painful. Nightmares tend to end after falling from bed at night. I had a nightmare that started after waking up from a fall; the pain was excruciating, I could not stand, hold myself, or change my position. It was a sharp stabbing nonspecific pain. My hip was in pain from inside, and my skin was burning me from the outside. I was rushed to the hospital in a state of confusion wondering what my x-ray will reveal. In retrospect, I was informed that I am malnourished. As I am sleep-walking through my experience, I find it difficult to eat and it is hard to use the toilet due to the continuous constipation I have. During my childhood, I thought that Epidermolysis Bullosa is just a bad season in my life. I saw the leaves of trees falling down in Autumn; however, my skin falls down all the year around and I am wondering when will the season where my skin regrows will come around. Today, I realize that hope is a word that describes something that is way bigger than emotions. My miracle consent is an act of hope, my hope that the story inspires the medical community to find a cure for Epidermolysis Bullosa.

Declarations
Consent:

A written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

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Author details:

Authors’ contributions:

Mohammed Tarabishi and Shahd Almonaie have written and edited the manuscript, and Mohammed Tarabishi, Mohamed Taha Abdelaty Mohamed, and Weam Farid Mousa have diagnosed and managed the patient. All the authors have participated in the final editing of the manuscript.

Competing interests:

The authors declare that they have no competing interests.

Declarations:

The ethical approval to conduct the case report was obtained from the committee of King Fahad Hospital and Department of Scientific Research Ethics in Health Affairs in Madinah.

Data sharing not applicable to this article as no datasets were generated or analyzed during the current study.

Moreover, the use of any animal or human data or tissue is not applicable in this project.

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References

Figures
Figure 1

Day of admission. Anteroposterior view radiographic image of the pelvis showing no fracture in the neck of the femur and right spiral mid-diaphyseal fracture of the femur can be seen.
Figure 2

Day of admission. Lateral view radiographic image showing right spiral mid-diaphyseal fracture of the femur.
Figure 3

Lateral x-ray image taken on the day of admission. Fracture of the femur and a crack in the distal diaphyseal region can be appreciated.
A radiographic image of the knee showing the knee joint and the distal femur physis still not fused.
Figure 5

Postoperative image of the lateral aspect of the thigh.
Figure 6

Postoperative image of the medial aspect of the thigh.
Figure 7

8 months, anteroposterior radiographic image showing Nancy nail. Showing delayed bone healing that took almost 4 months to heal completely. After close technique as anatomical reduction was not an option in this case.
Figure 8

Lateral radiographic image after sagittal reduction with good bone healing.

Supplementary Files

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