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Case Report

Neonatal multiple long bone fractures: A case presentation due to nemaline myopathy and review of other potential causes

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ABSTRACT

Nemaline myopathy is a rare genetic disorder caused by a mutation in genes encoding skeletal muscle proteins resulting in generalized hypotonia. It can be associated with neonatal multiple long bone fractures. We present a female neonate who had bilateral humerus and left femur fractures. Due to fetal distress, her delivery was at 37 weeks gestation by emergency cesarean section. The child was splinted and followed up for 8 weeks. She had a good union of fractured bones, callus formation, and no deformity. Muscle biopsy showed nemaline myopathy. Other biochemical and genetic tests were normal. The aim of this case report was to describe the presentation of multiple long bone fractures in neonates as an obstetric complication. Therefore, identifying the potential risk factors and planning the mode of delivery in future pregnancies, is critical in their management.

Keywords: Neonate fractures, Multiple fractures, Nemaline myopathy, Congenital myopathy, Perinatal fractures, Congenital fractures, LMOD3

INTRODUCTION

Neonatal multiple long bone fractures (NMLBFs) are rare. It is believed that cesarean section (CS) reduces the incidence of long bone fractures in susceptible babies, yet it does not preclude the possibility of their occurrence.[1] Emergency CS carries an increased risk for long bone fractures. It has been shown that the highest reported fetal injury was found in CS after a failed vaginal delivery experience. A multicenter study, which included 2088 breech presentations, reported that the incidence of long bone fractures is 0.1% after CS and 0.6% after vaginal delivery.[2] Published reports have identified a correlation between birth-associated long bone fractures and trauma as well as metabolic bone diseases, prematurity, low birth weight, and bone dysplasia. Therefore, identifying underlying diseases and risk factors are critical to their management.[3,4] In this paper, we report a neonate with nemaline myopathy who had bilateral humerus and left femur fractures after emergency CS that was performed because of indeterminate fetal distress and breech presentation.

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Nemaline myopathy is a rare hereditary autosomal recessive disorder with an estimated incidence of 1/50,000 and is responsible for 17% of all cases of congenital myopathy. It is underreported in the Middle East.[5,6] It was first described by Shy *et al*. and is typically present early in life.[6,7] It is classified into six types according to mutation and mode of inheritance. The most common mutation is in the genes encoding skeletal muscle proteins (e.g., α-actin and nebulin), which can result in generalized hypotonia, weakness, and feeding problems.[7,8] The prognosis varies according to the age of onset and mutation type, whereas severe congenital cases may have a fatal course due to respiratory failure. The diagnosis is made through muscles biopsy and genetic analysis.[8] The aim of this case report was to describe the presentation of multiple long bone fractures in a neonate as an obstetric complication and identify the possible risk factors.

CASE REPORT

A female neonate was born to a primigravida mother at 37 weeks of gestation with a birth weight of 2150 g. Prenatal history was insignificant except for the prenatal ultrasound examination, which showed reduced fetal movements. The medical history of the parents and their families was unremarkable and neither parents nor grandparents were related. The Apgar scores at 1 and 5 min were 6 and 7, respectively. She required respiratory support; therefore, she was shifted to the neonatal intensive care unit. An orthopedic consultation was requested immediately post-delivery because of decreased limb movements and abnormal shape of the left lower limb and both upper limbs.

Examination revealed both humerus and left femur fractures, with crepitus and intact distal circulation. There were no dysmorphic features and no flexion deformities. However, she had severe swelling of the limbs, lack of motion, and discomfort when moving the limbs. Chest radiographs showed normal dome-shaped hemi-diaphragms adjacent to normal lungs. Limbs radiographs showed bilateral humerus and left femur fractures [Figure 1]. Both upper limbs were splinted and lower limbs were put in gallows traction. The genetic and metabolic investigations were sent for the child.

The immobilization was removed at 2 weeks, and she was able to actively move her arms and legs; the radiographs showed callus formation at fracture sites. After 4 weeks, she was weaned off mechanical ventilatory support, but she needed a nasogastric tube for feeding due to hypotonia of epiglottis and pharyngeal muscles. At 6 weeks, follow-up radiographs showed good callus formation and no deformity [Figure 2]. Biochemical and genetic tests were normal; however, her muscle biopsy showed nemaline myopathy.

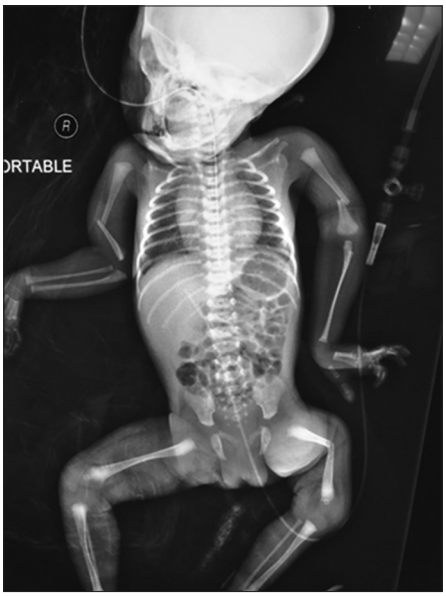

Figure 1: Radiograph at presentation showing bilateral humerus and left femur shaft fractures.

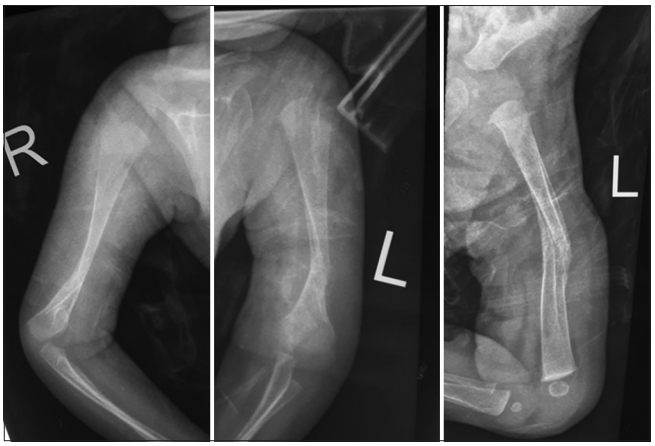

Figure 2: Radiographs illustrate 6-week follow-up showing a union of fractured bones, with good callus formation and no deformity.

DISCUSSION

Historically, NMLBF was thought to be unique to forceful obstetric breech maneuvers during vaginal delivery. However, reports have shown that birth-related injuries have been observed, with the increasing popularity of CS.[9] The present case has shown multiple factors, which may have contributed to the multiple bone fractures: Nemaline myopathy, breech presentation, and emergency CS. Some of these factors are modifiable and others can be controlled through pre-planning of delivery. For a better understanding of potential risk factors, it is easier to classify them. Neonatal musculoskeletal disorders, which can be associated with NMLBF, are either skeletal, metabolic, or neuromuscular

dysplasia. Examples of skeletal dysplasia are osteogenesis imperfecta, osteopenia of prematurity, campomelic dysplasia, and hypophosphatasia.[10] While the neuromuscular disorders, for example, nemaline myopathy, spinal muscular atrophy, central core disease, X-linked myotubular myopathy, Ehlers-Danlos syndrome, and infantile myofibromatosis.

NMLBF as an obstetric complication due to nemaline myopathy is rare. Monique *et al.* reported a series of 143 cases of nemaline myopathy, from which 49 cases had obstetric complications. These complications were distributed among different body systems wherein neonatal fractures occurred in 7 cases (4.9%) only.[6] On the other hand, obstetric risk factors which can lead to NMLBF may include macrosomia, malpresentation, primigravida, myomas, inadequate uterine relaxation, precipitous delivery, emergency CS, poor delivery techniques, and a small incision.[4,11] Reports have shown that elective CS can reduce intraoperative and post-operative complications compared to emergency CS.[12-14]

Identifying the potential risk factors in the prenatal period and proper planning may play a role in preventing long bone fractures. Therefore, the obstetrician must be very careful in the prenatal period to detect any genetic disorders and intrauterine growth retardation that may lead to long bone fractures and to be able to plan for the delivery mode. Thus, adequate uterus relaxation, avoiding energetic traction, and planning CS incision needed to be attained for safe delivery. Furthermore, a prenatal examination can help an orthopedic surgeon find out the underlying cause of a long bone fracture and guide future investigations and treatment plans.

Fortunately, NMLBF generally has a good prognosis and healing after appropriate immobilization. In this case, fractures were treated by immobilization and have shown good healing potential. Other reports with NMLF have shown the same observation wherein humeral fractures of newborns can be treated with splinting while femoral fractures can be treated either with traction or splinting.[1] The American Academy of Orthopaedic Surgeons recommends splinting for femur fractures (e.g., Pavlik harness) for neonates to reduce hospitalization and for easy application and nursing.[15]

CONCLUSION

NMLF is a rare obstetric complication. Therefore, identifying the potential risk factors and planning the mode of delivery in future pregnancies, is critical in their management. Although recurrence of nemaline myopathy in the future pregnancies is difficult to control due to the pattern of inheritance, genetic counseling is important in planning future pregnancies.

AUTHORS' CONTRIBUTIONS

AAG and VA collected and organized the data and wrote the manuscript, and AAG and WB revised and provided critical input. All authors have critically reviewed and approved the final draft and are responsible for the content and similarity index of the manuscript.

DECLARATION OF PATIENT CONSENT

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given her consent for her images and other clinical information to be reported in the journal. The patient understands that her name and initials will not be published and due efforts will be made to conceal her identity, but anonymity cannot be guaranteed.

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CONFLICTS OF INTEREST

There are no conflicting relationships or activities.

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