American Journal of Medical Case Reports, 2020, Vol. 8, No. 4, 98-99
Available online at http://pubs.sciepub.com/ajmcr/8/4/1
Published by Science and Education Publishing
DOI:10.12691/ajmcr-8-4-1

Femur Fracture Caused by Falling of a Boy with Osteogenesis Imperfecta

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Received December 05, 2019; Revised January 06, 2020; Accepted February 03, 2020

Abstract Osteogenesis imperfecta (OI) is the most common hereditable cause of bone fragility in children. A Japanese 2-year-old boy fell accidentally from a slide, sustaining a femur shaft fracture. Several maternal family members had experienced episodes of hearing impairment and bone fracture. The injury mechanism was clear and competent. Child abuse was unlikely. Strong family history and presence of blue sclera confirmed the OI diagnosis. Any doctor treating pediatric femur fractures should recall the rare cause of OI. Clinical evaluation alone without a family interview might not complete the diagnosis with OI. We continued proposing to his parents the benefit of receiving bisphosphonate treatment.

Keywords: *blue sclera, bone fragility, hearing loss, non-accidental injury, spica cast*

Cite This Article: Hiro Matsukura, and Yuto Takashima, "Femur Fracture Caused by Falling of a Boy with Osteogenesis Imperfecta." *American Journal of Medical Case Reports*, vol. 8, no. 4 (2020): 98-99. doi: 10.12691/ajmcr-8-4-1.

1. Introduction

Osteogenesis imperfecta (OI), an inherited disorder, is characterized by bone fragility, fractures, blue sclera, adult onset hearing loss, and joint laxity [1,2,3,4]. Its prevalence is as high as 1 in 10,000–20,000 births [2]. Inheritance is usually autosomal dominant, but new mutations and recessive inheritance have also been reported [1]. A mutation in either *COL1A1* or *COL1A2* genes encoding the chains of type I collagen, the major protein of bone, exhibits the phenotype in most instances: about 90% [1]. OI is a heterogeneous disease. Type I is the most common and mild form [2]. At present, no specific therapy exists for OI [2]. The first line of diagnosis is a detailed medical and family history, physical examination, proper radiograph, and routine laboratory examination [1]. If these will not accomplish the diagnosis, then more specific genetic testing is advisable [1]. This report describes a case of accidental femur fracture in a boy with OI.

2. Case Presentation

A 2-year-old Japanese boy with femur shaft fracture was transferred to our hospital by ambulance. The infant was born at term after an uncomplicated pregnancy as the second child of non-consanguineous parents. He was breastfed with no delayed eruption of milk teeth. He showed normal neurodevelopmental milestones. When playing in a park with his grandmother, he accidentally fell from a 1-m-high slide. Physical examination showed a boy in great pain, but well-kempt, well-nourished, and exhibiting growth in accordance with his age. His height was 89 cm (75th percentile), with 13 kg weight (75th percentile), and a Kaup index of 16.4 kg/m^2. He had no beading of the ribs, skeletal deformity, hearing loss, heart murmur or shortness of breath. The neurological examination results were normal. He had blue-gray sclera without dentinogenesis imperfecta. His white blood cell count was 6,700/μL, hemoglobin 13.1 g/dl, and C-reactive protein 0.7 mg/dl. Blood urea, serum creatinine, alkaline phosphatase, and serum electrolytes were within normal limits. His upper left leg was swollen and fixed in exorotation. Full body physical inspection revealed no bruise, injury, or scar on the body. A radiograph of his left leg showed a spiral fracture of the left femur (Figure 1). His bone mineral density was not measured.

He was treated with Weber's traction for 16 days, followed by immobilization in a spica cast. He had no documented medical or psychiatric history. No previous incident or fracture was recorded. The family history was remarkable for the following: his maternal grandfather had comminuted fracture of the right hand joints at three years and hearing loss at 40 years; his mother had comminuted fracture of the right hand joints at three years, leg fracture at 12 years with frequent visits to an otorhinolaryngologist; and his elder sister had a skull fracture at four years. On hospital day 24, he was discharged. The fracture healed with no complications. After 3 months, he visited the hospital and was able to run well without falling. The parents objected to genetic analysis and bisphosphonate treatment.

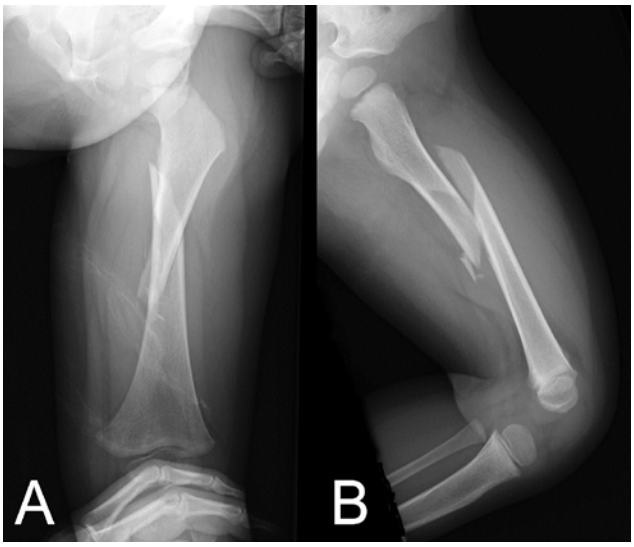

Figure 1. (A) Anteroposterior and (B) lateral radiograph of the left femur of a 2-year-old male showing a spiral mid-diaphyseal femur fracture.

3. Discussion

This case highlights two clinically important issues. First, doctors treating pediatric femur fractures should be conscious of the rare cause of OI. Femur fractures are the most common orthopedic injury in children [5,6]. Boys are predominantly affected more than the girls, with a ratio of 2.6:1 [5,6]. Falling from a height, after stumbling over some object, or while running are the most common events causing the injury [5,6]. Pediatric femur fractures occurred more frequently from short falls of less than 60 cm than from high falls over 1.2 m [5,6]. The most common hereditary bone disease causing fractures in children was OI [4,7,8]. Unexpected bone fractures are always a major concern in affected children and their parents [8]. Children with OI showed no risk of femur fracture in standing and normal gait modes, although jumping modes lead to femur fractures [3,8]. The 1 m height was sufficiently high for the necessary force to cause femur fracture in this boy with OI [5].

Second, this case underscored the importance of taking a family history to make or refine a diagnosis of OI. With recent advances in molecular genetic technologies for biomedical research, taking a family history has come to be regarded as old-fashioned [9]. Clinicians often undervalue the importance of the family history [9]. They are too busy to collect, classify, and analyze family history information during daily medical care. The author is no exception [9]. The author did not conduct this process, even after meeting the boy several times for routine care. Some orthopedist had a quick eye for blue sclera and obtained meaningful information successfully. The clear explanation of the injury and lack of risk factors related to child abuse and neglect successfully excluded non-accidental injury. The patient showed no musculoskeletal pain associated with joint hypermobility, short stature, and recurrent fractures. Lacking a family history interview, the definitive diagnosis of OI would have been missed. A missed diagnosis of OI might have put him at risk of incurring additional fractures.

Several times, we have proposed treatment by bisphosphonate administration and genetic testing for the definite diagnosis of OI. Genetic testing is useful for etiological diagnosis and for identifying the types of OI [10]. Oral and intravenous administration of bisphosphonate has shown effectiveness for improvement of bone mineral density and the remission of clinical features, although bisphosphonate treatment does not promise a complete cure for OI [10]. Unfortunately, the child's father did not agree to treatment. Even if he were to have a mild form of OI type I, he still faces a potential risk of repeated fractures. We intend to maintain contact with his parents and to continue to encourage them to choose bisphosphonate treatment.

4. Conclusion

Physicians encountering pediatric femur fractures should recall the rare cause of OI. Taking a family history is a powerful and reliable tool to make or refine an OI diagnosis, and to rule out non-accidental injury.

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