A Newborn with Multiple Fractures in Osteogenesis Imperfecta: A Case Report

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Introduction: Multiple bone fractures in a newborn can be associated with osteogenesis imperfect (OI). OI is a rare genetic disorder that causes Type I collagen synthesis disturbance resulting in bone fragility. Identification of the underlying disease correctly is crucial for multiple fracture treatment and care. Therefore, radiological, laboratory, and clinical evaluations, including genetic studies, must be done properly. Osteogenesis imperfecta (OI) is the most common cause of multiple fractures in newborns. OI, which is the most common cause of genetic osteoporosis, is seen in approximately 1/20,000 ratio [2]. In this case, we aimed at a newborn patient with bilateral clavicle, bilateral femur, and left humerus fracture with OI without any history of trauma.

Case Report

The prob and was the first child of healthy, 18-year-old mother. Parents were Syrian refugee. No relevant history was found. She was delivered at 36 gestation week by spontaneous vaginal delivery caused by premature rupture of membrane. She was 18 years old and G1P1. The delivery and pregnancy were uneventful. When examined the baby was active and pink. She weighed 2870g. She was not dyspneic. The chest was clear with normal heart sound. The abdomen was soft with no enlarged organs. The cord was tied without any problems. The baby was conscious with normotensive. Her parents did not describe any trauma. During the 2nd week of life, the baby became distressed whenever she was handled and bilateral arm and legs movements decreased spontaneously. She was admitted to a hospital. She was in good general condition, but she screamed in pain when bilateral legs and the left arm were moved, and bilateral clavicle was in pain with palpation in her examination. Blood tests revealed normal morphology and count of blood cells, and the C-reactive protein was negative. Laboratory values for Ca, P, and parathyroid hormone (PTH) were normal (PTH was 12, CA: 8.9, P: 3.7).
D level was 5ng/mL, between the normal range. X-ray examination was revealed the fractures: Bilateral clavicle, bilateral femur shaft, and left humerus shaft (Fig. 1). The fractures were initially suggested from the imaging study, so the baby was transferred to our clinic. When other clinic, genetic analyze performed, diagnosis of osteogenesis imperfect was suggested. The patient was assessed, and diagnosed with bilateral clavicle, bilateral femur, and left humerus fractures on the basis of the examination and obtained radiographs. Pavlik harness was performed for bilateral femur (Fig. 2) fixation, and the left humerus fracture was fixed with a needle to the clothes. Complete healing of the fractures was observed in the 2nd-month control examinations. The baby was followed regularly for 8 months. There was no complication during the follow-up. All fractures were healed with successfully (Fig. 3).

Discussion

There are several genetic disorders and congenital defect conditions that have been associated with bone fragility and fractures that can be misdiagnosed as child abuse [3]: OI, osteopenia or rickets of prematurity, infantile myofibromatosis, Cole-Carpenter syndrome, and lethal gracile bone dysplasias (LGBDs). OI is the most common genetic abnormality associated with multiple unexplained fractures in an infant or child that can be misdiagnosed as child abuse [4]. Generalized osteoporosis with underossified skull and spine, anisospondylly, multiple long bone, and rib fractures makes the diagnosis of OI usually easy. Of the osteogenesis subtypes, Type IIA is especially characterized by multiple fractures in rib fractures and also multiple fractures [5]. Due to these subtypes have diffuse hypomineralization in the bones, they can be diagnosed by prenatal ultrasonography [6]. In this subtype, respiratory problems are very common, and the patients are usually lost in the perinatal period. Preterm rickets was also with multiple unexplained fractures in an infant [7]. Infants were examined for clinical signs of rickets. There were also X-rays of any site of body taken for clinically suspected fractures. However, it is known that a large number of genetic disease groups are involved in the newborn multiple fractures. Disease groups such as LGBD, which have significant facial dysmorphism, short extremity, and short hand-foot characteristics, can also cause multiple fractures [8]. Femur fracture can be treated with Pavlik harness successfully [9]. Advantages of the Pavlik harness in the treatment of femur fractures include ease of application, minimal cost, reduced hospitalization, and ease of nursing care and diapering for infant. For this reason, the use of the Pavlik harness is recommended by the American Academy of Orthopaedic Surgeons for patients younger than 6 months of age.

Conclusion

Multiple fractures following delivery should be looked out for genetic disease groups which are involved in the newborn multiple fractures such as OI, especially in babies with decreased extremity movements. Thorough clinical examination and proper orthopedic consult in the event of doubtful presentation would help. These fractures have very good prognosis and show complete healing following appropriate immobilization.
Multiple bone fractures in the newborn can be associated with trauma as well as metabolic bone disease or hereditary bone dysplasia. Thorough clinical examination and proper orthopedic consult in the event of doubtful presentation would help.

References


Conflict of Interest: Nil
Source of Support: Nil

Consent: The authors confirm that Informed consent of the patient is taken for publication of this case report

How to Cite this Article