Case report

Cesarean section newborn with multiple fractures: an Osteogenesis Imperfecta case report

Mohammad Hossein Kariminasab[1] , Masoud Shayeste-Azar[2], Mohammad Hossein Kariminasab[2], MajidSajjadi Saravi[1], Seyed Mohamad Mehdi Daneshpoor[3,] MiladBahari[4],Mehran Fazli[4]*, Mahdi shayesteh azar

1- MD, Assistance professor Department of Orthopedic Surgery, School of Medicine, Mazandaran University of Medical Sciences, Sari, Iran.
2- MD, Assosiated professor, Department of Orthopedic Surgery, School of Medicine, Mazandaran University of Medical Sciences, Sari, Iran.
3- MD, Assistance professor Department of Orthopedic Surgery, School of Medicine, Mazandaran University of Medical Sciences, Sari, Iran.
4- MD, Orthopedic Resident, Department of Orthopedic Surgery, School of Medicine, Mazandaran University of Medical Sciences, Sari, Iran.
5- Medical student, Student research committee Faculty of medicine Mazandaran University of medical sciences, Sari, Iran.
6- Dentist student, Dubai, Emirate

Coreponding thour: Masoud Shayeste-Azar Mshayestehazar@yahoo.com

Abstract: Osteogenesis Imperfecta (OI) or Brittle bone disease, is a rare genetic disorder that causes type I collagen synthesis disturbance results in bone fragility.
We present a female newborn which had numerous fractures in the arm and femur. Her delivery was at 38 weeks gestation by caesarean section. Prenatal ultrasound revealed bone swelling and long bones shortness. The patient is pale blue sclera. 36 days after birth, the patient had a fracture in her left femur. Genetic analysis of the patient was reported. Protests by clinical, ultrasound and x-ray taken during pregnancy for this patient reported Osteogenesis Imperfecta type V.
shortening, swelling and deformity of the long bones during prenatal sonography can prove Osteogenesis Imperfecta before birth and select the correct orthopedic plan for treatment.
Key words: Osteogenesis Imperfecta, multiple fractures, collagen type I

1. Introduction

Osteogenesis Imperfecta is a rare genetic disorder characterized by type I collagen synthesis disturbance results in bone fragility and its growth shows deficiency (1). The literature reported that this disease have an incidence 10000/1 and 20000/1 (2). Clinical Classification of the disease from mild (Type I) or moderate (types V and VII) to severe (type III and IV) and even fatal have been reported (3). Type I is the most common form of the disease (more than 60% of cases) and clinical symptoms are usually mild. Type II is the most severe form of this disturbance and Stillbirth or death during infancy or early childhood have been reported. poor bone quality is more than other types and also low birth weights is reported. respiratory failure (Hypoplastic lung) or intracerebral hemorrhage are more common cause of mortality in the first year of life. Other factors such as, Infectious complications and cardiac disorders are also play in part . In this type new dominant mutation in a type 1 collagen gene or parental mosaicism exists. (3,4).
Type III and IV disease are severe form but not life threatening. Type III is the most severe form of this disorder in children who survive. Osteogenesis Imperfecta type IV is determined by moderate deformity and short stature. Furthermore, three other types of diseases (V, VI and VII) have recently been introduced (5,6).
.

2. Case presentation:

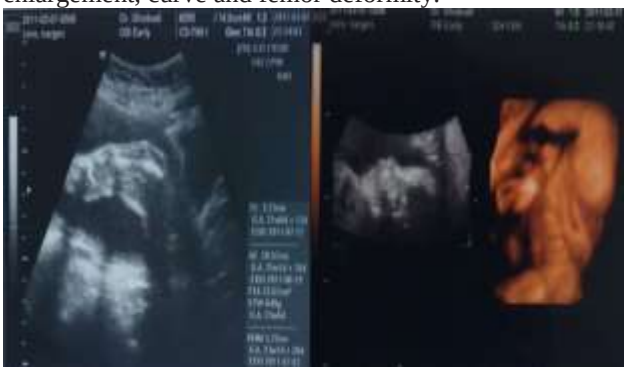
We present a 4 months old female infant with short stature and multiple fracture. At the 36 day of life she reffered to our

Neonatal out Patient clinic with short stature.

She was born at gestational age of 38 weeks by Caesarean section, at a teaching hospital. Her Apgar score was 7 and 9 at 1 and 5 minutes respectively, Her birth weight was 3kg and head circumference was 31.5cm. Pernatal care for her was complete and attended by the same hospital that section was done. Her mother age at the time of delivery was 25 years old and she didn't take any drug during pregnancy. Also she denied any medical history or febrile disease before and during pregnancy. Her family history for similar disorder and other hereditary disease was negative. Lab tests such as CBC, ESR, CRP, Ca, P, Alkp, Liver tests, electrolyte, urea, creatinine was in normal range.

In our clinic, infant was afebrile, with short stature. In ultrasound which was done at 25 weeks gestational age, bony swelling and short bones such as the femur relative to gestational age (22 weeks), tibia (23 weeks) and arm (23 weeks) reported (Figure 1).

Figure 1; Sonography in 25±1weeks that shown shortage of long bone include of femor (22 weeks), tibia (23 weeks) and humerus (23 weeks) with enlargement, curve and femor deformity.



But heart, brain and other organs of the fetus was normal. She had pale blue sclera After birth. According to the deformity of the bone in ultrasound during pregnancy radiography was requested for the patient. In X-ray radiography, multiple bone fractures in the left arm and the right femural shaft was reported (Figures 2 and 3).

Figure 2; Left shoulder and humerus X-ray that shown left shaft humerus fracture.

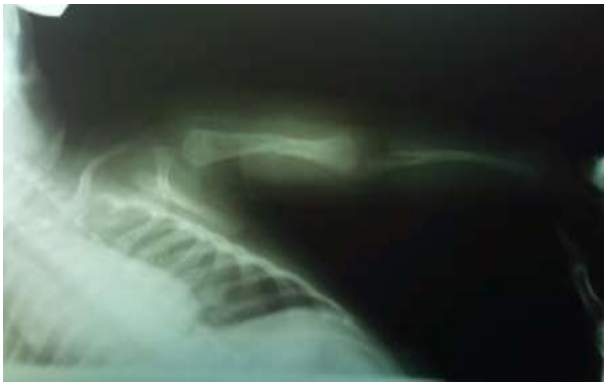
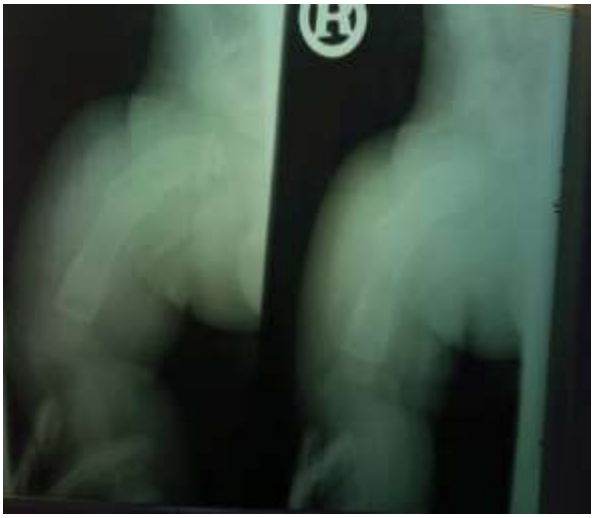
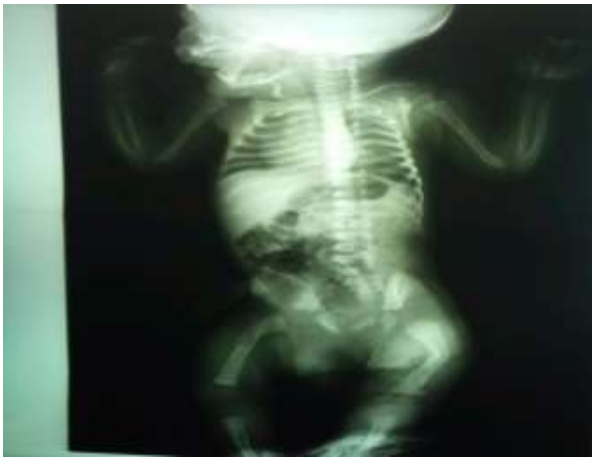


Figure 3; Right femor X-ray that shown right shaft femor fracture.



Genetic analysis of chromosomes was observed in patients with no important findings. After 36 days, 4.2Kg weight and other spontaneous fracture was seen in the left femur without trauma (Figures 4).

Figure 4; whole body X-ray that shown multiple bone fracture in both femor.



Based on clinical features, ultrasound and X-ray findings and lack of findings in chromosomes study, Osteogenesis Imperfecta type V was diagnosed.

3. Discussion:

Osteogenesis Imperfecta is a rare inherited disorder of connective tissue that characterized by bone fragility. In this condition, COL1A1 and COL1A2 mutations affect the amount of type I collagen (7,8). The clinical findings in this disease include bone fragility(Hallmark), short stature, blue sclera, deformity of the long bones, partial hearing loss(Hypoacusis) and ligament laxity (8). Based on the classification that was used previously, Osteogenesis Imperfecta four main types (I to IV) (9), But recently, three new types of the disease have been identified including type V, VI and Rhizomelic or type VII or (10,11). In this category, the most severe forms of OI is type II which is associated with intrauterine death (3). Type III and IV are associated with severe disorder and type I is mild and classic form of this disease. Type II, III and IV have molecular model of a dominant negative while the type I pattern is haploid insufficiency. Molecular model of type V, VI and VII are unknown(3).

Severe hearing loss in Osteogenesis Imperfecta type I, mild hearing loss in types II, III, IV and normal hearing in the types V, VI and VII have been reported(3). Our patient had a good response to sound and her hearing examinations was normal. Her sclera color was pale blue.

According to the Previous studies sclera colors is dark blue in types I and II, sky blue in type III, white in type IV , white to pale blue in type V and VI.(3) The Type VII is still not known

Choice of diagnostic method during pregnancy for fetal morphology is ultrasound as a screening test. According to previous manuscripts, this way can be useful in prenatal diagnosis of Osteogenesis Imperfecta. The sonographic findings in Osteogenesis Imperfecta during pregnancy include reducing the acoustic shadow, shortening of the long bones, and the long bones swelling.(2,6,12) In the present study the prenatal sonographic evidence of shortening, swelling and deformity of the long bones that was consistent with previous reports.

finally, shortening, swelling and deformity of the long bones can cause prenatal diagnosis and select of best treatment of Osteogenesis Imperfecta before birth.

We should suggest the clinicians to consider Osteogenesis Imperfecta in mind in every neonate suspected to this rare disorder and thus physical examination should be complete and accurateand finally should help from clinical and radiographic tools for better result and decrease the perinatal deaths rate.

Corresponding Author:
Masoud Shayeste-Azar
MD, Assistance professor Department of Orthopedic Surgery, School of Medicine, Mazandaran University of Medical Sciences, Sari, Iran.
E-mail: mshayestehazar@yahoo.com

References

1- Marini JC. Osteogenesis imperfecta. In: Behrman RE, Kliegman RM, Jensen HB. Nelson textbook of pediatrics. 17th ed. Philadelphia: W.B. Saunders; 2004: 2336-2338.

2- Bastos F, Perez LT, Narváes CPL, Costa O, Souto da Silva RC, Van-Dunem JCVD, Miranda SMRN, Cordeiro LB, Maia PCS, Airosa LFLM, et al. Severe osteogenesis imperfecta: case report. Einstein 2010; 8:480-2

3- Solopova A, Wisser J, Huisman TAGM. Osteogenesis Imperfecta Type II: Fetal Magnetic Resonance Imaging Findings. Fetal Diagn Ther 2008; 24: 361-7

4. Lehmann HW, Herbold M, Bodman JV, Karbowski A, Stücker R. Osteogenesis imperfecta. Monatsschr Kinderheilkd 2000; 148: 1024.

5- Li M, Xia WBB , Xing XPP, Yu W, H YY, Jiang Y, Wang O, Liu HJJ, Han LWW, Heng XWW, et al. Benefit of infusions with ibandronate treatment in children with osteogenesis imperfect. Chin Med J 2011;124(19):3049-53.

6- Van Dijk FS, Pals G, Van Rijn RR, Nikkels PG, Cobben JM. Classification of osteogenesis imperfecta revisited. Eur J Med Genet 2010; 53: 1-5.

7- Cheung MS, Arponen H, Roughley P, Azouz ME, Glorieux FH, Waltimo-Sirén J, Rauch F. Cranial base abnormalities in osteogenesis

imperfecta: Phenotypic and genotypic determinants. J Bone Miner Res 2011; 26: 405-13.

8- Rauch F, Lalic L, Roughley P, Glorieux FH. Relationship between genotype and skeletal phenotype in children and adolescents with osteogenesis imperfecta. J Bone Miner Res 2010; 25: 1367-74.

9- Sillence DO, Rimoin DL. Classification of osteogenesis imperfect. Lancet 1978;1:1041–2.

10- Lehmann HW, Herbold M, Bodman JV, Karbowski A, Stücker R. Osteogenesis imperfecta. Monatsschr Kinderheilkd 2000; 148: 1024.

11- Primorac D, Rowe DW, Mottes M, Barisic I, Anticevic D, Mirandola S, Gomez Lira M, Kalajzic I, Kusec V, Glorieux FH: Osteogenesis imperfecta at the beginning of bone and joint decade. Croat Med J 2001; 42: 393–415.

12- Morgan JA, Marcus PS. Prenatal diagnosis and management of intrauterine fracture. Obstet Gynecol Surv 2010; 65(4):249-59.