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Case Report

Osteogenesis imperfecta: a case report

Kambiakdik T.[1]*, Inderpreet Sohi[2], Toli Nyorak[1], Pawam Kumar[1], Sengseng R. Sangma[1], Zosangliani[1]

[1]Department of Paediatrics, Regional Institute of Medical Sciences, Imphal, Manipur, India
[2]Department of Paediatrics, Christian Medical College and Hospital, Ludhiana, Panjab, India

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***Correspondence:**
Dr. Kambiakdik T.,
E-mail:tkambiakdik80@gmail.com

ABSTRACT

Osteogenesis imperfecta (OI) is a group of rare inherited disorders of connective tissue with the common feature of excessive fragility of bones caused by mutations in collagen. Diagnosis is mainly based on the clinical features of the disorder. We report a late preterm a male neonate born to a 20 years old primigravida. He had clinical features of a type II OI and severe birth asphyxia.

Keywords: Alkaline phosphatase, Bluish sclera, Fragile bones, Osteogenesis Imperfecta, Wormian bones

INTRODUCTION

Osteogenesis imperfecta, the most common genetic cause of osteoporosis, is a generalized disorder of connective tissue. The spectrum of OI is extremely broad ranging from forms that are lethal in the perinatal period to a mild form in which the diagnosis may be equivocal in an adult. Approximately 1 in 20000 cases of OI are detectable during infancy. The severity varies greatly, even among individuals of the same family. Four main types of OI have been identified. Type I is the most common and the mildest while Type II is the most severe.[1]

CASE REPORT

Authors present a case of a late preterm male neonate, gestational age of 35 weeks, born to a 20 year old primigravida who underwent regular ante-natal check-ups throughout the pregnancy in a primary health centre and had received all routine medications and vaccines as prescribed under the prevailing health system. The antenatal ultrasonogram showed no fetal anomaly at the end of the second trimester. The baby was delivered by normal vaginal delivery and cried with the initial steps of resuscitation. On examination, he was clinically preterm 34 weeks with a birth weight of 1600 grams, length of 42cms and head circumference of 28 cms. The baby had a frog legged posturing, a protuberant abdomen, large anterior fontanelle and an open posterior fontanelle with widely separated cranial sutures. He was also noticed to have deformity of lower limbs. On the right thigh, crepitus was felt while on the left thigh an abnormal bony prominence was felt along with forward bending of the thigh. The chest showed paradoxical movements with respiration. Based on these findings, a babygram was done which showed generalised decrease in bone density with multiple fractures of the long bones, bell shaped chest, with multiple fractures of different ages while the skull roentgenogram showed wormian bones. Hearing screen with OAE for both ears suggested right ear pass and left ear refer. Serum alkaline phosphatase, serum calcium and phosphate along with cranial USG were done. The serum alkaline phosphatase level was 137 U/L and the patient was managed conservatively with

minimal handling, as advised by the orthopaedic consultant. They suggested splinting at 4 weeks of life. Cholecalciferol and calcium were supplemented at 400IU per day and 150mg/kg/day respectively. Ophthalmologic assessment showed mild bluish discoloration of the sclera.

DISCUSSION

Osteogenesis Imperfecta is a group of rare disorders affecting the connective tissues and characterized by extremely fragile bones that fracture easily, often without apparent causes. In all types of OI, the symptoms vary greatly from case to case even within families. OI have an autosomal dominant pattern of inheritance. They may range from a mild disorder with few symptoms to a severe debilitating disorder. The age of onset of fractures also varies from case to case.[1,2]

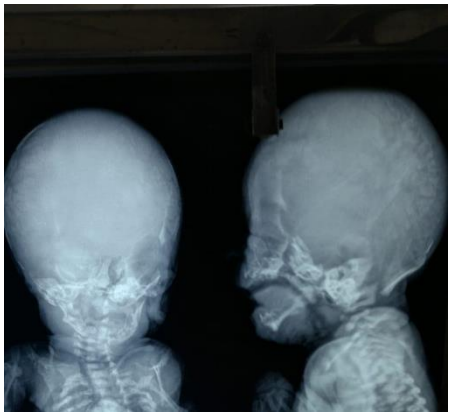

Figure 1: X-ray of skull showing wormian bones.

OI Type I is the commonest and usually the mildest form in most cases. It is characterized by multiple bone fractures usually through childhood and puberty. Fractures usually begin when the child begins to walk: fractures during newborn period are rare. The frequency of fractures usually declines after puberty.[1-3]

OI type II is the most severe form. Affected infants often experience life threatening complications at or shortly after birth. Infants often have low birth weight, abnormally short arms and legs and bluish discoloration of the white of the eyes. Affected infants may have extremely fragile bones and numerous fractures at birth. They may also have a small narrow nose, small jaw and an abnormally soft calvarium with an abnormally large fontanelle. They may also have an abnormally thin fragile skin and hypotonia.[1-4]

OI Type III is characterized by extremely fragile bones, multiple fractures and malformed bones. Multiple fractures are often present at birth. Fractures and malformation of various bones may progress as the affected infants and children ages. In some cases, affected infants may develop pulmonary insufficiency and respiratory problems.[1,6,7] They may have a slight bluish discoloration of the sclera but most often fades during the first year of life. Hearing impairment and dentinogenesis imperfect may also be present.[1-4]

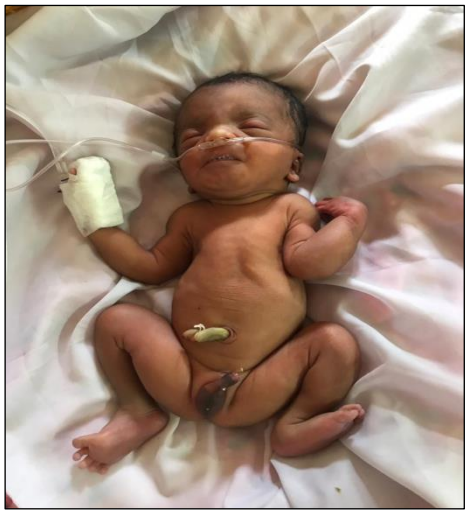

Figure 2: Frog legged posture, bell shaped chest, protuberant abdomen.

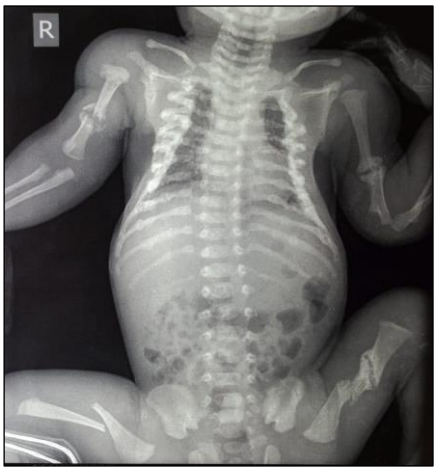

Figure 3: Multiple fractures of different ages.

Individuals with OI type IV have fragile bones yet fractures may be more common before puberty.

Follow up includes a good orthopaedic management and rehabilitation. Regular hearing assessment is vital for early detection of hearing deficit. Respiratory infections and neurological complications like basilar invagination and brainstem compression are frequent. Dentinogenesis imperfecta is also a common association, which can become apparent after few months. Cutaneous laxity and occasional bleeding diathesis are also reported. No curative therapy exists and using bisphosphonates to reduce osteoclast mediated bone resorption has targeted increased born turnover. Intravenous pamidronate and calcitriol administration reduces bone pain and fracture incidence, with increased bone density and level of ambulation, with minimal side effects. Growth hormone

therapy had also been tried with variable results. Gene therapy may be the answer in the future.[4,5]

Genetic screening and counselling is of paramount importance since the risk of an affected individual passing the gene to the offspring is 50% and the recurrence risk for an unaffected couple of having a subsequently affected offspring is 5-7%. Antenatal prediction can be done at 14-16 weeks of gestation by ultrasonography with severe shortening of the long bones, femoral length- abdominal circumference ratio of less than 0.16, hypoplastic thorax and marked bowing or fractures. Chorionic villous sampling can be done for recurrent cases.[4-7]

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