

## Case Report

# An Unusual Presentation of Perinatal Osteogenesis Imperfecta Mimicking Fracture Femur – A Case Report

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**Abstract:** Osteogenesis imperfecta (OI) is a genetic disease characterized by fragile bones, skeletal deformities and, in severe cases, prenatal death that affects more than 1 in 10,000 individuals. We report a case of a primigravida, a 24 yr old lady who presented in the emergency room of a secondary care hospital in Howrah, West Bengal, India (Silver Jubilee Matrisadan) at 36 weeks of gestation with breech presentation. Upon delivery by Em LSCS the Obstetrician & Neonatologist found that there was fracture in right femur and abnormally stunted left leg. Fracture femur during delivery was suspected. Radiological examination of both lower limbs showed fracture of right femur, with abnormal acute bowing of left femur with no associated shadow showing fracture haematoma. The neonate was transferred to a tertiary care centre for investigation and further diagnosis and management. Neonate was diagnosed to have Osteogenesis Imperfecta. This peculiar puzzling presentation which was primarily suspected as fracture of femur due to manipulation during breech delivery was subsequently diagnosed to be an undiagnosed case of osteogenesis imperfecta with intrauterine fractures.

**Keywords:** Osteogenesis imperfecta, primigravida, Breech delivery, intrauterine fracture.

## INTRODUCTION

OI was formerly referred to as Lobstein's disease, named after the first to correctly identify the pathophysiology of the disease [1]. Oakley and Reece (2010) stated that OI is one of the most prevalent skeletal dysplasias and that it occurs in approximately 1 in every 20,000 births [1, 2]. Mutations in genes in type I collagen account for 90% of OI cases with type I as the most common and mild form. Individuals with this type have blue sclera and the majority of their fractures occur prior to puberty. More serious variants exhibit a varied range of fractures including intrauterine fractures [1-3].

## CASE REPORT

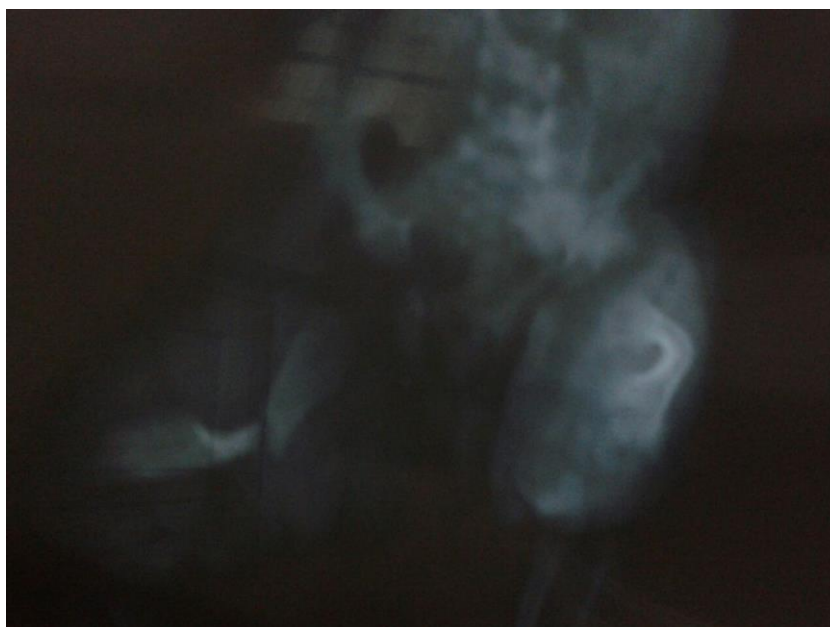
A young primigravida twenty four years old was booked in a municipality maternity hospital of Howrah (Liluah Silver Jubilee Matrisadan). She underwent all necessary antenatal check-up throughout her pregnancy period. Detailed history of both the patient and the family was not suggestive of any medical, surgical or congenital abnormality. All requisite routine antenatal investigations including ultrasonography in 1<sup>st</sup> and 2<sup>nd</sup> trimester were done. All investigations were well within normal limits. The

patient had breech presentation from 28<sup>th</sup> weeks of gestation. Throughout the antenatal period all parameters like body weight, blood pressure and laboratory investigations were within normal limits. At 36<sup>th</sup> week of gestation the patient presented with dribbling and foetal distress and decision for Emergency Caesarean section was taken and performed under spinal anaesthesia. The patient was hydrated with Ringers lactate 1lt and spinal anaesthesia was given at L3, 4 interspace with 2.2ml (12mg) of bupivacaine with 26G Spinocan needle. The patient was haemodynamically stable throughout the operation. Mild sedation was given with 2ml of midazolam. Abdomen was opened with a Phannensteil incision and was opened in layers. Bleeding vessels were secured and the peritoneum breached. After uterine incision, the fetus which had a breech presentation was delivered by breech delivery with Burns Marshall Technique for the after coming head. After delivery of the baby, the baby cried immediately and was handed over to the paediatrician who graded the baby APGAR 7.

The paediatrician, on routine examination of the neonate, felt a gap in the right femur bone, which was

first suspected as a fracture which inadvertently occurred during manipulation of the breech. But on further examination of the left limb, a knob like structure was felt in place of the femur bone. A congenital abnormality was suspected and X-Ray of the neonate was done. The radiographic images showed that there was a fracture in the right femur of the neonate with the absence of any fracture haematoma. In place of the left femur a knob –

like structure was present which presented like a bowed femur most likely due to intrauterine fracture followed by malunion. Osteogenesis Imperfecta was suspected by our team and the neonate was referred to a tertiary paediatric centre for further diagnosis and management. The neonate was transferred to Calcutta Medical College and later the diagnosis of Osteogenesis imperfecta was confirmed.



**Fig-1: Image showing fracture rt femur with acute bowing of left femur.**

## DISCUSSION

Osteogenesis imperfecta is the result of a mutation in one of the two genes that carry instructions for type 1 collagen - the major protein in bone and skin [1-3]. The mutation may result in either a change in the structure of type 1 collagen molecules or in the number of collagen molecules. Either of these changes results in weak bones that fracture easily and other connective tissue symptoms [2, 3].

Results of studies in recent years show that very frequently people with OI, , have dominantly inherited forms of the disorder. It is possible that the child of a person with OI will have a spontaneous genetic mutation resulting in a different type of OI. Some individuals with very mild OI have been known to have a child with more severe symptom. Excluding OI, the risk of other congenital disorders in pregnancies in which one parent has OI is the same as that of the general population [1, 4].

Most researchers now agree that recessive inheritance rarely causes osteogenesis imperfecta [4]. About 25 percent of children with OI are born into a family with no history of the disorder. That is, a child is born with a dominant genetic mutation that causes OI, yet neither parent has OI. This occurs when the child has a "new" or "spontaneous" dominant mutation. The gene

spontaneously mutated in either the sperm or the egg before the child's conception [1, 3, 4]. Now that the child has a dominant gene for OI, he or she has a 50 percent chance of passing the disorder on to his or her children. There are no known environmental, dietary, or behavioural triggers for this type of mutation. This appears to be the reason in our case.

In most cases, when a family with no history of OI has a child with OI, they are not at any greater risk than the general population for having a second child with OI [3, 4].

More recently, however, researchers have concluded that the rare recurrence of OI in a previously unaffected family is more likely due to a phenomenon called mosaicism [1, 4]. Studies suggested that the mutation, instead of occurring in an individual sperm or egg, occurred in a percentage of cells that give rise to multiple sperms or eggs. Thus, although the parents are not affected by the disorder, the mutation present in a percentage of his or her reproductive cells can result in more than one affected child [5].

Ultrasound examination of the foetus identifies a variety of skeletal and bony abnormalities within the foetus and allows for the differentiation of lethal and non-lethal skeletal dysplasias. However, in isolation it

cannot discriminate between the various skeletal dysplasias with certainty. The skeleton begins to ossify early in development, and can therefore be assessed by ultrasound throughout most of pregnancy. The clavicle, mandible, ileum, scapula, and long bones ossify by 12 weeks of gestation. The detection of skeletal diseases of prenatal onset has improved enormously with advances in 2-D imaging. However, the sensitivity of 2-D ultrasound, which represents the current standard of care for prenatal diagnosis, remains limited, ranging most typically between 40% and 60%. The diagnostic accuracy ranges from 31% by Kurtz and Wapner in 1983 to 68% by Schramm et al in 2009 [6].

3D ultrasound definitely broadens the technical horizons achieved by 2-D imaging, but does not replace it; rather, the two should be considered complementary techniques. 3-D ultrasound allows the acquisition and storage of multiple 2-D planes, which with the use of increasingly sophisticated computer algorithms, allows this volumetric data set to be reconstructed and displayed in any plane. Recent data supports the use of 3-D ultrasound to improve the accuracy of standard 2-D imaging in the evaluation of skeletal abnormalities [7]. It should be noted that it can be difficult to tell from an ultrasound whether the foetus has OI Type II or Type III. Ultrasound can be used to examine the foetal skeleton for bowing, fractures, shortening, or other bone abnormalities consistent with OI. Ultrasound is generally most helpful for prenatal diagnosis of the more severe forms of OI. The foetal skeleton shows signs of OI as early as 16 weeks in OI Type II, and 18 weeks in OI Type III. Foetuses with mild OI seldom show evidence of fractures or deformity before birth. 3D US can detect foetal OI precisely, and provide additional vivid illustration after various modes of reconstruction that 2D US cannot [6, 7].

According to a recent study by Rachel *et al*, caesarean delivery did not decrease fracture rates at birth in infants with nonlethal OI, and in non lethal forms there was no benefit of C.S.[8]. This study also found that most caesarean deliveries were done for the usual obstetric indications and not specifically because OI was detected in the foetus [9].

It is recommended that couples at risk of having a child with OI seek genetic counselling before conception, or as early in the pregnancy as possible. A genetic counsellor can provide information on OI genetics and prenatal diagnosis. Many of the secondary centres in India have to make do with 2D ultrasound machines in which a wide margin of error remains. So a screening with 3D ultrasound machine in the 2<sup>nd</sup> trimester would be advisable for detection of skeletal abnormalities in order to come to a diagnosis at an earlier stage and advise the couples accordingly [10, 11].

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