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OSTEOGENESIS IMPERFECTA: A CASE REPORT

Orthopaedics

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ABSTRACT

Osteogenesis imperfecta (OI) is the most common cause of multiple fractures in newborns. It is a rare genetic disorder that causes Type I collagen synthesis disturbance resulting in bone fragility. Incidence of OI- 1:20,000 . Inheritance is generally autosomal dominant but new mutations are common and recessive inheritance occurs. Regardless of the time at which the diagnosis is suspected, the first line of evaluation is a detailed medical history, family history, physical examination, appropriate radiographs, and routine lab testing. If these do not lead to the diagnosis, then specialized genetic testing is warranted. Treatment for baby born with OI is a multidisciplinary approach –for fracture prevention with bisphosphonates, fracture management when present, realignment osteotomies for long bone deformities.

KEYWORDS

Osteogenesis imperfecta, brittle bone disease,bone fractures.

INTRODUCTION

Osteogenesis Imperfecta is a rare monogenic disorder of bone fragility, also known as a "brittle bone disease".The term OI encompasses a broad range of clinical presentations that may be first apparent from early in pregnancies to late in life, reflecting the extent of bone deformity and fracture predisposition at different stages of development or postnatal ages. It is caused by a mutation in COL1A1 or COL1A2 genes, resulting in abnormal collagen cross-linking and an overall decrease in type 1 collagen.

The condition is characterized by easily occurring bone fractures, skeletal deformities, shortness of stature and bluish eye sclera, hearing loss, joint hypermobility, dentinogenesis imperfecta, and cardiovascular and pulmonary complications.

The diagnosis of OI is considered at different times: during fetal development, at birth, in childhood, or less often in adults.

Pre-pregnancy preparation and family planning allow a wider variety of reproductive options, reduce associated risks, and enable the arrangement of OI pregnancy, delivery, and early treatment options where necessary.

Early prenatal diagnosis of OI is beneficial, as it provides enough time for a pregnancy management decision.

It also allows consideration of delivery management and early OI treatment directly after birth, or even antenatally, with the developing method of mesenchymal stem cell transplantation.

Case Report

We present a case of a female baby born with numerous fractures of the diaphysis of the left humerus, right radius and ulna, and bilateral femur.

My patient had her delivery at 33 weeks +5 days gestation by Cesarean Section(Indication-previous 1 CS with scar tenderness with preterm breech with intractable hypertension with severe oligohydramnios). A female baby born and baby did not cry immediately after birth, was intubated but expired after 30 min of birth.

My patient, Mrs. K, aged 26 years, is Sikh by religion.

The patient was booked, G2P1+0L0 at 33+3 weeks POG (by dates) with previous 1 C-Section with Gestational Diabetes mellitus on Medical Nutrition Therapy who came to OPD on 1/11/23 for regular ANC checkup.

Her Antenatal period was uneventful.

Obstetric History-M/L – 2 years. Para 1 – Her first child was born 1 year back by Emergency LSCS i/v/o preterm breech with oligohydramnios.

Baby died just after birth.

There was no significant medical, surgical, past, or family history.

On examination the patient was conscious and oriented to time, place, and person.Vitals were stable.On Per abdomen examination, fundal height corresponds to 30weeks, frank breech, liquor seems reduced,SFH-29cms, scar tenderness was absent at the time of admission. FHS/+/R/134bpm

As there was repetition of breech presentation with oligohydraminos ,a risk of congenital anomaly in this baby was kept in mind although level II scan was normal.

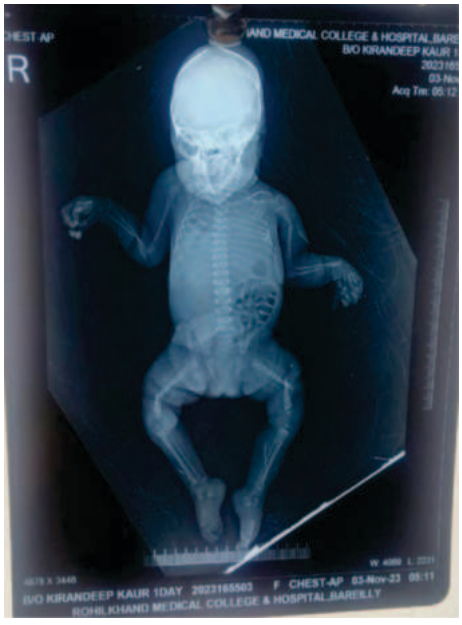
The patient was admitted for steroid coverage and for conservative management; USG done on admission also did not show any anomaly.

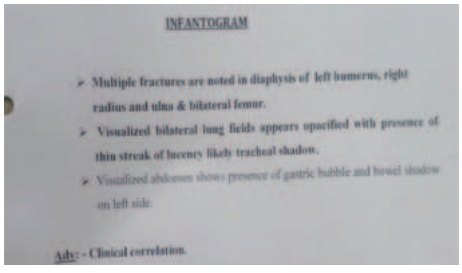
On day 3 of admission she started having pain abdomen and her BP was raised ,she was also complaining of decreased fetal movements, scar tenderness was present on examination for which she was taken up for emergency Cesarean section.

During cesarean section for breech extraction, the obstetrician noticed multiple crackling sounds, likely caused by brittle bones. The baby did not cry immediately after birth and was handed over to the pediatrician.Despite all resuscitative measures, the baby could not be revived and was declared dead.

O/E the baby was found to have cleft palate and multiple fractures of B/L upper and lower limbs. After examination of baby pediatrician had a suspicion of Osteogenesis imperfecta.In view of suspicion of OI full body X ray of baby and autopsy was advised.

The family refused for autopsy.



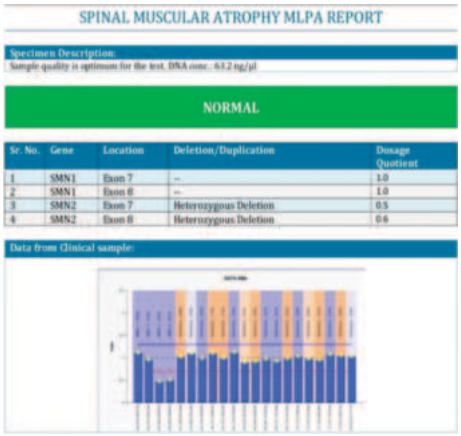


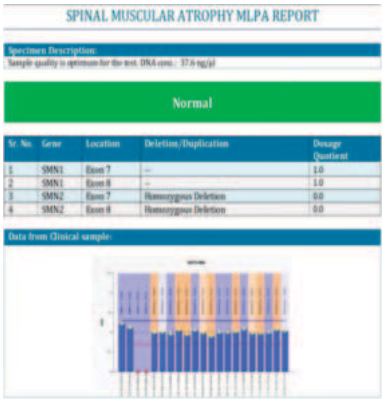
With due consent of the baby's father whole body X ray of baby was done which showed –numerous fractures of upper and lower limb ie the diaphysis of left humerus, right radius and ulna and bilateral femur.

Visualized bilateral lung fields were opacified with presence of thin streak of lucency like tracheal shadow.Visualized abdomen shows presence of gastric bubble and bowel shadow on left side.

After the delivery when the family was asked repeatedly about history of previous child birth, they revealed that previous child born also had multiple fractures and was not revived ,this history was kept hidden by the attendents and was not told earlier during her antenatal visits.

On follow up, the couple were advised for Spinal Muscular Atrophy MLPA testing. On the basis of the report, couple referred to genetic clinic.





DISCUSSION

Skeletal dysplasia, also known as osteochondrodysplasia, is the name given to a heterogeneous group of diseases that comprise abnormalities in the bone and cartilage due to genetic mutations. Thanatophoric dysplasia, achondrogenesis II, and hypochondrogenesis are the most common lethal skeletal dysplasias. Regarding the non-lethal ones or the ones of variable lethality, there are OI, congenital spondyloepiphyseal dysplasia, and heterozygous achondroplasia

Sillence classification divides patients into categories I-IV, all of them considered autosomal dominant, and an X-linked variant in the type I presentation was added later (2013). Moreover, due to genetic and radiological advances, new types have recently been recognized, and the classification has been extended, including types V-VII, being types I-V predominantly autosomal dominant and the rest from VI to VII considered to be autosomal recessive.

Regarding the diagnosis of OI, it is mainly clinical and radiological. It can be made prenatally or postnatally. The clinical manifestations are variable, from mild forms to severe presentations, with the most distinctive characteristic being bone fragility, vulnerability to fracture due to minimal trauma or even its absence, and deformity or low growth. Extraskeletal manifestations include joint hypermobility, dentinogenesis imperfecta, blue sclera, and hearing loss. Muscle weakness and pulmonary and cardiovascular complications are rare.

CONCLUSION

OI is a disease with multiple clinical findings and genetic variations. An early diagnosis is important; it allows a comprehensive and multidisciplinary approach to the management of the patient. On the contrary, the consequent fractures and bone deformities will decrease the quality of life of these patients.