Perinatallethal Osteogenesis Imperfecta Type II A with white sclerae:A Case Report

Dr. Rinku Saini[1], Dr. R. N. Sehra[2], Dr. Lovesh Saini[3*], Dr. Priti Saini[4]

[1]Assistant Professor, Department of Pediatric Medicine, S.M.S Medical College, Jaipur(Rajasthan)India
[2]Professor, Incharge NICU, Mahila Chikitsalaya, S.M.S Medical College, Jaipur(Rajasthan)India
[3]Senior Demonstrator, Department of Preventive and Social Medicine, SMS Medical College, Jaipur(Rajasthan)India
[4]Assistant Professor, Department of Pathology, SMS Medical College, Jaipur(Rajasthan)

Abstract— *Osteogenesis Imperfecta is a genetic connective tissue disorder. It has clinically heterogeneous four types. Type 2 is the most severe and perinatally lethal form having small thorax, curved limbs and blue/gray sclerae. It is further subclassified into 3 types. A baby with sign and symptoms with macrocephaly, retrognathia, low set ears, widely placed eyes with white sclerae, complete cleft palate, narrow chest, curved and shortened limbs, B/L CTEV, left undescended testis with hypospadiasis was reported. This rare case was thoroghly examined and investigated which came out to a case of 'Osteogenesis Imperfecta type 2 A' having white sclerae. So case having such symptoms were should be investigated for Osteogenesis Imperfecta.*

Keywords: Osteogenesis Imperfecta Type 2, White Sclerae, Genetic disorder, Perinatally lethal condition.

I. INTRODUCTION

Osteogenesis Imperfecta (OI), is a rare inherited bone disease. It is the most common genetic cause of osteoporosis, a generalized disorder of connective tissue.[1]

It can be identified in perinatal period. Its spectrum is extremely broad varying from mild to lethal forms. In the perinatal period. It's cause being structural or quantitative defect in type I collagen.[2]

It is classified mainly into 4 types i.e. Type I –Type IV according to Silence classification based on the varying phenotypic features.[3] Type I is the common mild form, type II is the perinatal lethal form, type III is a severe form and type IV is a moderately severe form.[4,5]

Osteogenesisimperfecta type II is the most severe form leading to inutero or perinatal death and has been subclassified into groups i.e. group A, B and C on the basis of radiological features.[6] Cases of perinatally lethal forms of OI have already been reported in the past.[7,8,9]

At birth, infants with OI type II A have very short and curved limbs, long bone fractures, small chest and soft skull, sclerae are usually blue or gray.[4,5,6,7,8]

This rare case of OI type II A who was having all the characteristic features for its type[4,5] but differed in having white sclerae was reported.

II. METHODOLOGY

A rare case of OI type II A who was having all the characteristic features for its type but differed in having white sclerae was admitted in NICU, Mahila Chikitsalaya, SMS Medical College, Jaipur (Rajasthan) India. So case study was done thoroughly and case report was prepared to publish this rare case.

III. CASE REPORT

A late preterm (34-36 wks) appropriate for gestational age male baby with birth weight of 2.2 kg was born to a second gravida mother by LSCS. Indication for LSCS was restricted fetal movements with oligohydramnios. There was no consanguinity, no family history of OI.

Pregnancy was uneventful. Antenatal ultrasound examination done in first trimester which was suggestive of poly hydramnios and skeletal dysplasia. A second ultrasound done in third trimester which was suggestive of oligohydramnios. Index case was second in birth order. Previous one was an abortion in first trimester. There was a history of leaking per vaginuum for about 24 hours but there was no history of fever.

At birth, baby had a good cry; Apgar at '1' being 6/10 but baby had respiratory distress for which he was admitted in NICU.

Postnatal examination showed macrocephaly, retrognathia, low set ears, widely placed eyes with white sclerae, complete cleft palate, narrow chest, curved and shortened limbs, B/L CTEV, left undescended testis with hypospadiasis. (Figure 1)

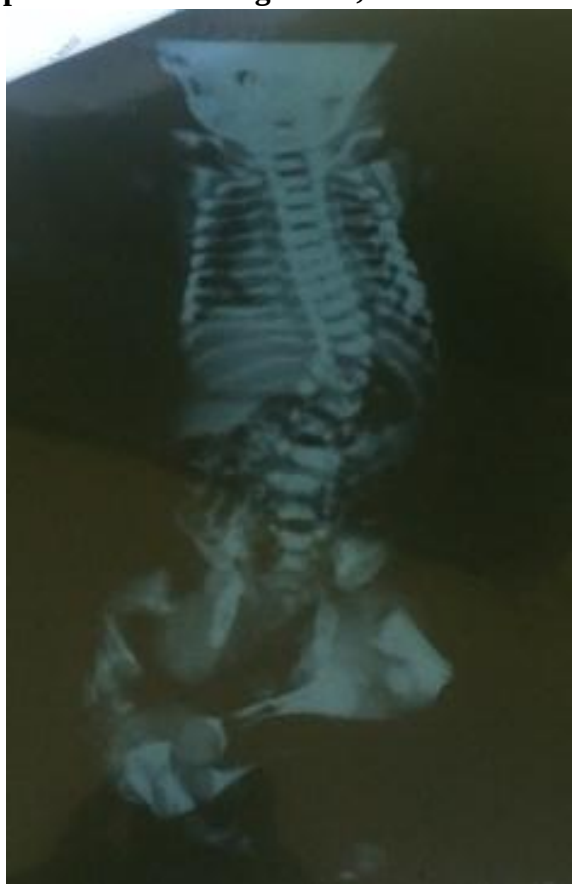
Postnatal bone radiographs showed hypomineralization, scolisis, narrow thorax, multiple beaded ribs, long bone fractures and wormian bones in skull. Among other routine investigations done, CBC, CRP, RFTS, LFTS were within normal limits. (Figure 2)

Figure 1

Infant with OsteogenesisImperfecta Type II A

Figure 1

Bone demineralization, scoliosis, crumpled appearance of all long bones, beaded ribs



Based on the characteristic clinical features and radiographs, a diagnosis of oteogenesis imperfecta type II A was made.

Baby initially was taken on Continuous Positive Airway Pressure (CPAP), Intravenous fluids and antibiotics. But he got deteriorated on day 3 of life (LD3), so was taken on ventilator with other supportive measures. But baby expired on day 7 of life (LD7) due to respiratory failure secondary to pulmonary hypoplasia.

IV. DISCUSSION

OsteogenesisImperfecta is a genetically determined disorder of the connective tissues.[1] A variety of bio-chemical defects in type I procollagen resulting in disruption of triple helical conformation are responsible for the clinical features.[2] The major phenotypic features include bone fragility, dentinogenesisimperfecta, blue sclerae, deafness and ligamental laxity.[1,2]

Some cases are autosomal recessive but many are new dominant mutations. Autosomal dominant forms of OI occur equally in all racial and ethnic groups, whereas recessive forms occur predominantly in ethnic groups with consanguineous marriages.[1,2]

It is clinically heterogenous with mild, moderate, severe and lethal phenotypes, classified into main four types (I-IV) according to Sillence classification.[3]

Babies are classified as having OI – type II if they die in the perinatal period.[4,5] The radiographic appearances are used to subclassify OI – type II in to group A to C or group I to IV according to Sillence subclassification.[6]

In type II A OI, the babies are mostly small for gestation, have a hypoplastic face with micrognathia and deep blue- gray sclera, a large calvarium, chest being small and protuberant abdomen, limbs are short and bowed. One or more limbs are held in a severely deformed position.[7,8,9,10]

Radiographs show generalized osteopenia, long bones have a beaded appearance due to thin and crumpled cortex. There is generalized platyspondyly. Skull has numerous wormian bones. Such babies normally die at birth or are stillborn.

In type II B, ribs being less affected, respiratory distress are less and babies may survive. Other features being similar to type II A. And in type II C, babies are very small having severe osteopenia and numerous fractures. It is the least common subtype and such babies hardly survive.

This present case justifies for its type as it had most of the characteristic clinical and radiological features reported in literature.[6,7,8,9] A distinctive feature was the presence of white sclerae in place of blue/ grey sclerae. Cases of OI type II A reported in past have mentioned blue/gray sclera as an ocular feature[6,7,8,9,10] and it is difficult to find case reports of type II OI with white sclerae.

As OI type II is very lethal and such cases hardly survive might be a reason that such minor variations were not noticed and reported.

V. CONCLUSION

A baby with sign and symptoms with macrocephaly, retrognathia, low set ears, widely placed eyes with white sclerae, complete cleft palate, narrow chest, curved and shortened limbs, B/L CTEV, left undescended testis with hypospadiasis should be investigated for Osteogenesis Imperfecta. As in this

cases which were investigated radiologicaly and found cases OsteogenesisImperfecta type II A.

CONFLICT OF INTEREST

None declared till now.

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