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Case Report

OSTEOGENESIS IMPERFECTA TYPE II RARE PRENATAL FATAL FORM – A CASE REPORT

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ABSTRACT
The present case being reported is a lethal variant of Osteogenesis Imperfecta considered as type II with multiple fractures , tubular long bones, severe limb shortening and the fatality of the present case could be due to hypoplasia of lungs leading to respiratory failure.The other less severe forms of Osteogenesis Imperfecta were described as type I, type III and type IV. In Type I form of OI fractures do not occur at the time of birth, occurence of the fractures after adolescence becomes rare, minimal bone deformity, hearing loss and blue sclera are evident. OI Type III show multiple fractures at birth, progressive bone deformity, and short stature, hearing loss and tooth deformity are the features blue sclera of infancy becomes normal after adolescence. OI Type IV has all the features of Type I except that this form does not have blue sclera.

***Key Words**: Osteogenesis Imperfecta, Multiple Fractures, Hypoplasia And Blue Sclera.*

INTRODUCTION

Osteogenesis Imperfecta means "Imperfectly formed Bones" in greek and latin otherwise known commonly as brittle bone disease The mythical danish prince Ivar Benlos was carried to the battle feild on a sheild as was a victim of Osteogenesis imperfecta . Osteogenesis Imperfecta type II a rare fatal congenital abnormality with an incidence of one in every fifty thousand births Wynbrandt (2004); characterised by multiple fractures, severe bone deformity, small chest with beaded ribs , demineralisation of skull and the hypoplasia of lungs leading to the fatality . Osteogenesis Imperfecta a relatively not so infrequent form of abnormality needs a clear distinction amongst fatal OI type II form from other less severe forms at the time of prenatal diagnosis.

CASES

A 27 year female with a non consanguious marriage, second gravid with a healthy first child, and no family history of congenital anomalies, delivered a 28 week stillborn male foetus with gross anatomical abnormalities at Maharajahs Institute of Medical Sciences after an abnormality was detected in antenatal scan ordered as the patient was complaining of severe pain in the abdomen and the following were the observations.

Foetal Autopsy - Findings

External Features:

The face appeared triangular .

Multiple fractures were evident to the naked eye ,which was due to the displacement of the distal fractured segments from the alignment of the proximal segments both in upper and lower limbs leading to angulated appearance at the site of fracture. Chest was short and Tubular in appearance (Fig. 1).

Foetogram revealed

Generalised osteopenia of all the bones was observed, ribs appeared Beaded due to the fractures which occured and healed.

Long bones seemed to be Crumpled and bowed due to the callus formation at the site of the fracture.

Cortex of the bones was thin due to poor ossification **(Fig. 2).**

Internal Features:

The prominent feature noticed was Bilateral Hypoplastic Lungs.

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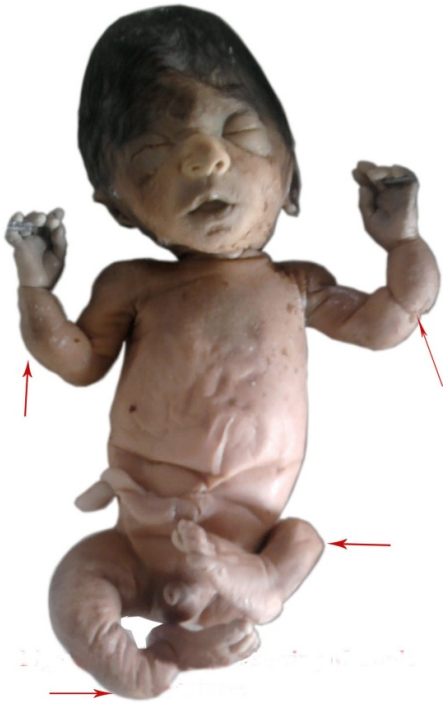

Figure 1: Arrows showing multiple fractures

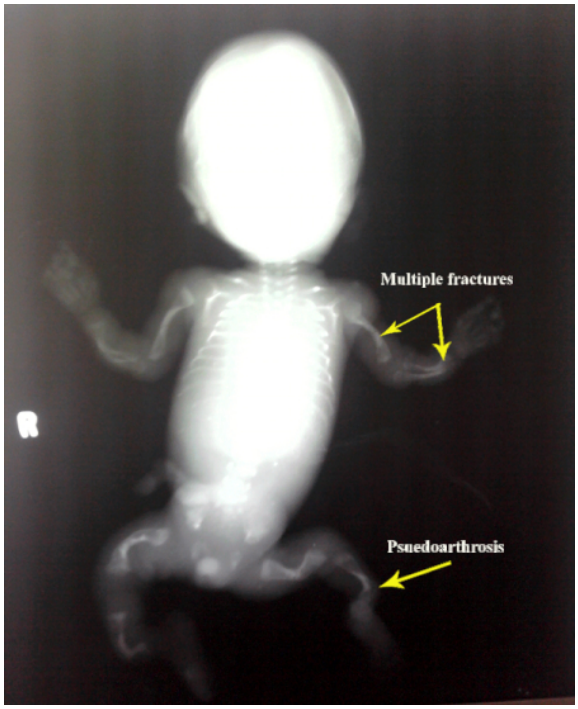

Figure 2: Foetogram showing multiple fractures and Psuedoarthrosis.

DISCUSSION

James (2004) stated that OI type II is characterised by multiple fractures, severe bone deformity results usually from sporadic mutations due to a possible germ line Mosaicism, with a recurrence risk of 6 % in the subsequent pregnancy.

Mutations in COL1A1,COL1A2 genes of type 1 collagen with a reccurence risk of 3 % attributing to gonadal mosacism was considered responsible for OI type II by Woodward,(2004).

Sillence(1979) (1984) classified the various forms of OI into four major groups on the basis of the phenotype and concluded that all forms of type II OI are likely to be autosomal recessive inheritence.

A study on family data and radiographs of 71 probands of the OI type II phenotype by Peter (1988) recorded a recurrence of (8.6%) amongst that some were due to autosomal recessive trait in consanguinous families,in one of the non consanguinous families gonadal mosiacism was noted in the mother while in the rest of the cases with no history of consanguity the attributed cause was other mutations, hence forth this OI type II is being considered as a Biochemically heterogeneous disorder usually due to new Mutations in the Genes for Type I Collagen.

Type II OI has been subdivided into three forms Young (1987), designated A, B, and C on the basis of radiological findings, contradicting the earlier view autosomal dominant inheritance was propounded.

The present case represents OI type II B manifesting a triangular facies which is considered as a feature of OI type III while others reported a round face, multiple intranatal fractures, tubular bones with thin cortices, short and tubular chest, discontinuous minimaly beaded ribs unlike OI type A where ribs would be much more beaded continuously and also differing with type C of OI where in ribs would be long without any beading and relatively longer and thinner long bones. With history of consanguity being absent autosomal dominant and autosomal recessive forms of inheritance were overruled, while germline mosaicsm could not be ascertained as the family was destituet efforts of tracing the mother did not succeed.

Conclusion

In rural India the awareness of antenatal scanning is far from being acknowledged for its role in determining the congenital malformations rather known as a sex determining marvel. Meticulous evaluation of progressive viability of the foetus and diagnosis of the condition at an early foetal

gestation is pivotal determinants for the future of the foetus. Emphasizing the fact that if early diagnosis is not made during antenatal scanning the delivery may lead to the death of the mother due to the injury caused to the uterus by the angulated limbs and the sharp fractured bones and Educating the women of rural origin about the implications of regular antenatal evaluation using case reports such as the present case material is needed.

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