A CASE OF OSTEOGENESIS IMPERFECTA WITH SIGNIFICANT DISABILITY
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ABSTRACT: Osteogenesis imperfecta (OI) is a rare genetic disorder characterized by structural and quantitative defects in type 1 collagen resulting in susceptibility to fractures of long bones or vertebral compressions from mild or inconsequential trauma¹. There are different types that range in severity from mild form to perinatal lethal form. We present a case of type 3 osteogenesis imperfect with multiple fractures, severe short stature and severe disability who survived till 5 years of age.

KEY WORDS: osteogenesis imperfecta, multiple fractures, disability

INTRODUCTION: Osteogenesis Imperfecta (OI) is an autosomal dominant disorder that occurs in all racial and ethnic groups. OI means imperfect bone formation. Individuals with OI have brittle bones most often as a result of mutations affecting collagen type I, which is the most prevalent protein in bone, skin and other connective tissues. These mutations can lead to different levels of skeletal deformities and in some cases frequent multiple fractures¹. The incidence of OI that is detectable at infancy is 1 in 20000². Type 2 is the lethal form. Type 3 is the most severe non-lethal form. The most common variety is type 1. We present a case of type 3 osteogenesis imperfecta with severe physical disability.

CASE REPORT: A 5 year old male child, born to second degree consanguineous married couple with a known history of OI came with complaints of fever, cough and tachypnea. He weighed 7 kgs and his length was 66.5 cms. Mother noticed fractures in the thigh in the newborn period on day 8. On examination child had multiple deformities of limbs. He had relative macrocephaly, triangular facies, flat face, short neck, deformed limbs and extreme short stature. Scleral hue was white. He had severe tachypnea with chest retractions. On auscultation crepitations were heard throughout the lung fields. He had past history of repeated respiratory tract infections for which he was admitted and treated with intravenous antibiotics. He had no hearing impairment. Various x-rays were obtained including images of his chest, pelvis, upper and lower extremities.

X-ray chest showed bilateral non-homogenous patches, thin ribs. X- Rays of long bones demonstrated gross demineralization with thin cortex. Multiple fractures were seen in the radius, ulna, humerus, tibia and fibula at various stages of healing. The bone survey showed bowing of the proximal femora bilaterally. It also showed an irregularity of the proximal femora bilaterally consistent with prior fractures. The child was started on intravenous antibiotics for severe pneumonia. His respiratory distress worsened and he succumbed to the disease.

DISCUSSION: OIs is due to structural or quantitative defects in type 1 collagen. According to Sillence classification, type 1 is milder form. Type 2 is severe perinatal lethal form. Type 3 OI is the severe nonlethal form with typical characteristics of relative macrocephaly, multiple fractures, normal sclera, progressive long bones, spine deformities and short stature. Fractures in individuals with OI
CASE REPORT

Limb deformities in OI prevail as a therapeutic challenge. Multidisciplinary management would improve function and quality of life.

CONCLUSION: Limb deformities in OI prevail as a therapeutic challenge. Multidisciplinary management would improve function and quality of life.
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