

CASE REPORT

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 imperfecta 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: OI is due to structural or quantitative defects in type 1 collagen. According to Silience 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

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heal at a normal rate; however they have a poor quality callus. Recurring fractures typically lead to progressive deformity, both shortening and angulation. This is due to the poor quality callus being easily deformed by weight-bearing forces. Skeletal survey in our case showed multiple fractures with angulation. The main cause of morbidity and mortality is cardiorespiratory. Recurrent pneumonias with declining lung function occur in childhood

Respiratory complications secondary to chest deformity and scoliosis lead to limitations in thoracic function and make it the principle cause of death in most patients with OI. Lung expansion and thoracic movements can be limited by deformities in the thoracic spine and abnormal positioned ribs⁴. The severity of these limitations will increase due to rib fractures and respiratory weakness. Other complications are hearing loss, kyphoscoliosis⁵. Combined medical and surgical modalities are feasible options for correction of deformities⁶⁻⁷. In our case fractures are seen right from the neonatal period. The child was neither on medical treatment nor could surgical correction be made due to financial constraints. As a result in our case the child was totally non-ambulant due to severe physical disability.

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



Fig.1: x-ray upper limbs showing fractures of radius and ulna



Fig.2: x-ray chest showing thin ribs



Fig.1: x-ray lower limbs showing fractures in femur and tibia and fibula



Fig. 4

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