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Case report

A RARE CASE OF OSTEOGENESIS IMPERFECTA TYPE III

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ABSTRACT

Osteogenesis imperfecta (OI) the most common genetic cause of osteoporosis is a generalized disorder of connective tissue, characterized by increased bone fragility, low bone mass, recurrent fractures & numerous extra-osseous features with unusual presentations. We report a case of 7 year old female child presenting with respiratory distress with bowing of limb. This case is presented for its rarity.

Keywords: Osteogenesis imperfecta, Multiple malunited fractures, Recurrent respiratory infections, Chest wall deformity

INTRODUCTION

Osteogenesis imperfecta is also known as Brittle Bone Disease.[1] It is one of the most common causes of genetically the inherited osteoporosis.[2] It is a connective tissue disorder that classically demonstrates fragility of skeletal bones.[3] It has a broad spectrum ranging from perinatal lethal forms to milder forms in adult, though it is rare it should be considered in the differential diagnosis of children presenting with short stature and multiple deformities, apart from rickets and child abuse.

CASE REPORT

7years old girl born to non-consanguineous parents was admitted with respiratory distress & fever for three days in paediatrics department in Meenakshi medical college hospital and research institute. There was history of recurrent fractures and progressive deformity of all four extremities. She also suffered from recurrent respiratory tract infection requiring frequent hospitalization. Past history revealed progressive difficulty in walking, squatting after the age of 5 years. Her birth and neonatal period were uneventful. Development was normal till five years. No other family members were affected from similar illness.

On examination child was sick looking, pale, dyspenic, severely short statured and apparently macrocephalic. She had white sclera triangular facies with narrow thoracic cage, crowding and flaring of lower ribs. Abdomen was protuberant with marked kyphosis. Vision, hearing & dentition were normal. There was marked bowing of extremities. Respiratory examination showed Intercostal & subcostal retractions, B/L crepitation's & rhonchi were present. Other systems were normal.

Investigations revealed normal hemogram, renal function test and electrolytes were normal. Serum calcium was reduced and phosphorous was normal. Alkaline phosphatase was significantly raised (1448), Thyroid function test was normal. Chest x-ray revealed bilateral pneumonitic patches & haziness with lack of air spaces & callous formation.

Hip x-ray revealed protrusio acetabulum.

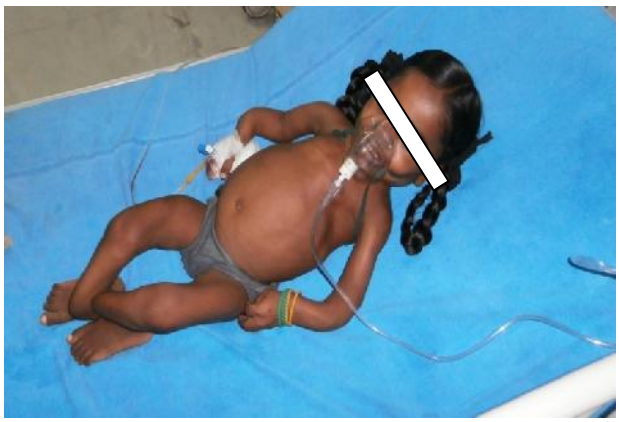

Fig 1: Child showing multiple deformities and respiratory distress

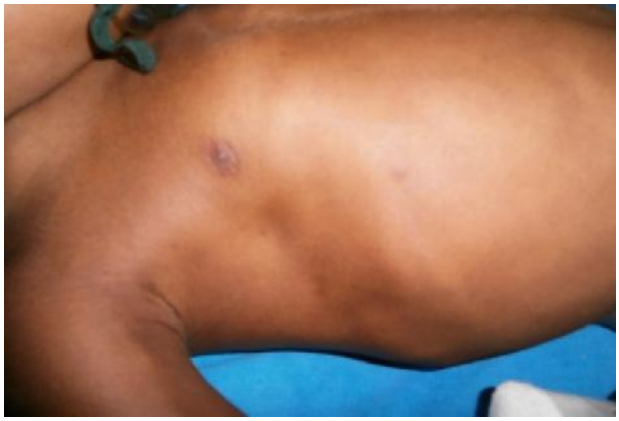

Fig 2: Thoracic wall deformity with flaring of ribs

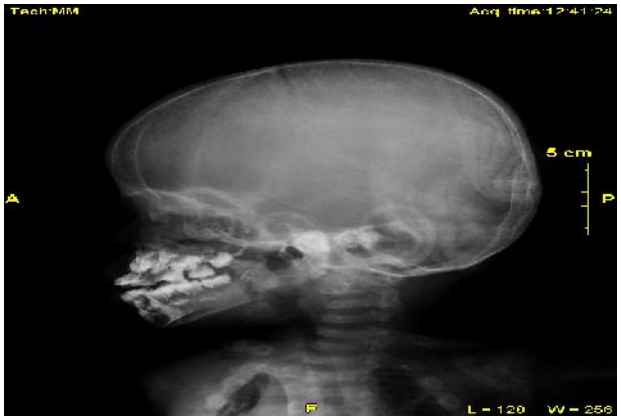

Fig 3: Skull x-ray

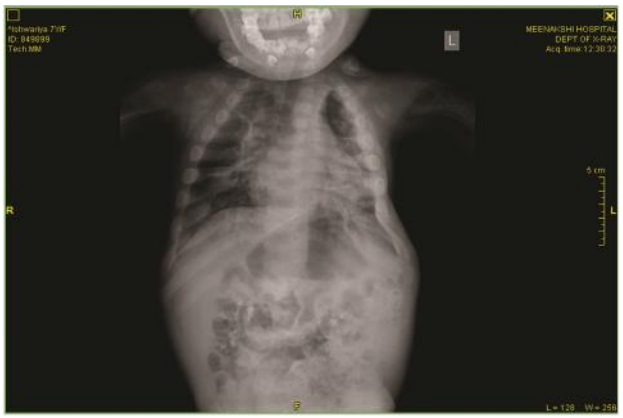

Fig 4: X-ray chest showing callous fornation in ribs and multiple pneumonitic patches

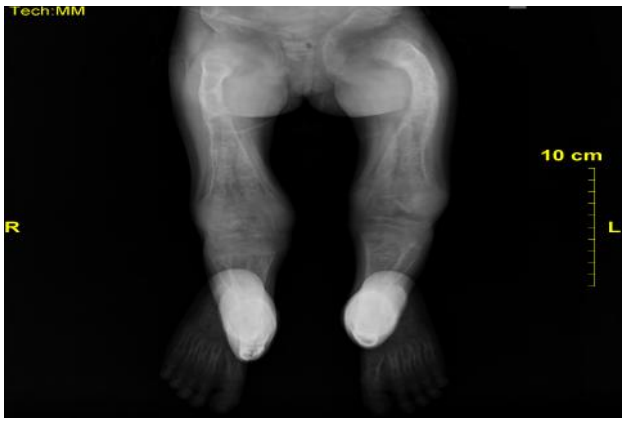

Fig 5: X-ray of the hip showing Protrusio acetabulum

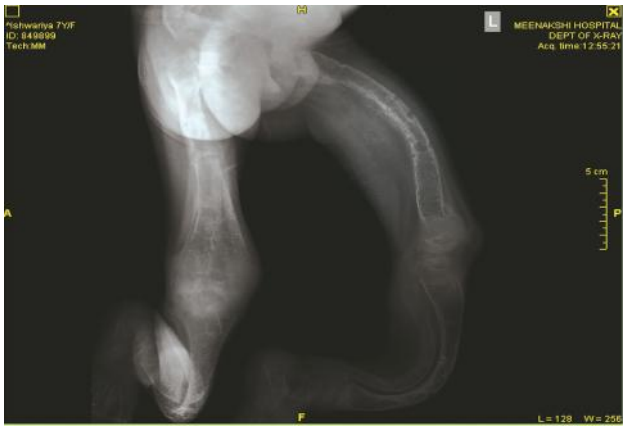

Fig 6: X-ray lateral view of vertebra showing kyphosis and platyspondly

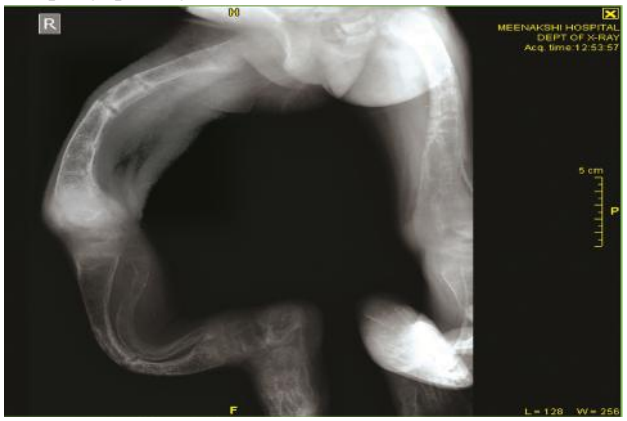

Fig 7: X-ray of long bones showing multiple healed and healing fractures with reduced trabacular pattren with thinning of the cortex

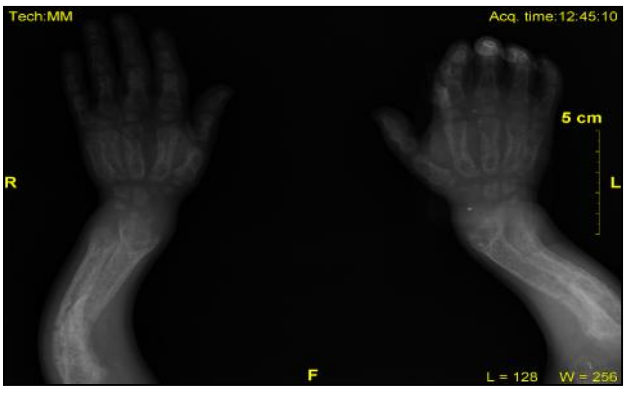

Fig 8: POPCORN appearance in the metaphysis

X-ray of extremities showed thinned cortices. Osteoporosis and loss of transverse trabeculations. POPCORN appearance was seen in metaphysis of long bones. X –ray spine shows platyspondly

DISCUSSION

Osteogenesis imperfecta (OI) the most common genetic cause of osteoporosis is a generalized disorder of connective tissue, characterized by increased bone fragility[4], low bone mass, recurrent fractures & numerous extra-osseous features with unusual presentations.

Pathogenesis : It is a group of phenotypically related disorder caused by the deficiencies in the synthesis of type I collagen .The genetic defect is due to the mutation in the genes that code for the alpha 1 & alpha 2 chains of the collagen molecule(type I collagen)[6]. Life expectancy may be shortened[7]. This case is presented for its rarity and unusual presentation.

Table 1: Types of Osteogenesis imperfecta[8]

Type	Inheritance	Severity	Fractures	Bone Deformity	Stature
I	AD	Mild	Few to 100	Uncommon	Normal or slightly short for family
II	AD	Perinatal lethal	Multiple fracture of ribs, minimal calvarial mineralization, platyspondyly, marked compression of long bones	Severe	Severely short stature
III	AD Rare recessive	Severe	Thin ribs, platyspondyly, thin gracile bones with many fractures, "popcorn" epiphyses common	Moderate to severe	Very short
IV	AD	Moderate to mild	Multiple	Mild to moderate	Variably short stature
V	AD	Moderate	Multiple with hypertrophic callus	Moderate	Variable
VI	Uncertain	Moderate	Multiple	Rhizomelic shortening	Mild short stature
VII	AR	Moderate	Multiple	Yes	Mild short stature

AD: Autosomal dominant, AR: Autosomal Recessive

CONCLUSION

Clinical features and imaging studies are suggestive of Osteogenesis imperfecta type-III and this case is presented for its rarity.

REFERENCES

1. Thompson EM, Young ID, Hall CM, Pembrey ME. Osteogenesis imperfect type-III. Delineation of the phenotype with reference to genectic heterogeneity. American Journal of Medical genectics. 1986;23(3);821-32
2. Falvo KA, Klain DB, Krauss AN, Root L, Auld PA. Pulmonary function studies in osteogenesis imperfecta. Am Rev Respir Dis. 1973;108:1258-60.
3. Goldman AB, Davidson D, Pavlov H. Popcorn calcifications: a prognostic sign in osteogenesis imperfecta. Radiology. 1980;136 (2): 351-8.
4. Ian Bickle, Drsaqba farooq. Osteogenesis imperfect,http:/radiopedia.org/articles/ Osteogenesis imperfecta-1
5. Chris Souder, Osteogenesis imperfect. Osteogensis imperfecta. http:/www.orthobullets. com/paediatrics/412/ Osteogenesis-imperfecta
6. Plotkin H. Syndromes with congenital brittle bones. BMC Pediatr. 2004;4:16.
7. Paterson CR, Ogston SA, Henry RM. Life expectancy in osteogenesis imperfecta. BMJ. 1996;312:351.
8. Robert Ds, Melanie GP, Peter Hb. Osteogenesis Imperfecta (Brittle Bone Disease, OI). http://www.ncbi.nlm.nih.gov/books/NBK1295/