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Case Report

A neonate with multiple skeletal deformities and pathological fractures: a case of osteogenesis imperfecta

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ABSTRACT

Full term male child born to a multigravida mother presenting with brachycephaly, widely patent anterior fontanelle, low-set ears, a high-arched palate and profound limb deformities marked by shortening, bowing, bilateral genu varum and congenital talipes equinovarus and multiple pathological fractures. Whole exome sequence was suggestive of COL1A2 mutation confirming our diagnosis of Osteogenesis Imperfecta. Child was managed with a multidisciplinary approach involving paediatrician, orthopaedics opinion, endocrinologist for further evaluation and management. Child was administered with Inj. Pamidronate and discharged on calcium supplements along on direct breast feeding.

Keywords: Limb deformities, Multiple fractures, Osteogenesis imperfecta, Pamidronate, WES

INTRODUCTION

Osteogenesis imperfecta (OI), or brittle bone disease, is a rare inherited connective tissue disorder characterized by reduced bone mass, increased bone fragility and recurrent fractures following minimal trauma. The condition is associated with variable skeletal deformities and extra skeletal manifestations such as blue sclerae, dentinogenesis imperfecta, joint hypermobility, and hearing loss.[1] OI is a multisystem disorder with potentially serious complications including basilar invagination, cardiovascular abnormalities such as aortic root dilatation and mitral valve prolapse, and restrictive lung disease secondary to scoliosis.[2]

The disorder exhibits marked genetic and clinical heterogeneity, most commonly resulting from mutations affecting type I collagen synthesis, with both autosomal dominant and recessive patterns of inheritance.[3] The global incidence of osteogenesis imperfecta is estimated to range between 1 in 15,000 and 1 in 20,000 live births.[11-13] The birth prevalence has been reported to be approximately 6-7 cases per 100,000 population worldwide.[11-14] The incidence appears largely consistent across different racial and ethnic groups, although certain populations may demonstrate a higher frequency of autosomal recessive forms, particularly in regions where consanguinity is common.[12-17]

Among the different subtypes, OI type I is the most common, accounting for the majority of cases, while more severe forms such as type II and type III occur less frequently worldwide.[11-13]

CASE REPORT

A male neonate, born to a 30-year-old multigravida mother, was delivered at term by normal vaginal delivery. The mother was G3P1L2 with no significant antenatal complications and no history of consanguinity.

The pregnancy was supervised, and antenatal records were available for review. Antenatal ultrasonography performed at 30 weeks of gestation revealed evidence of skeletal dysplasia. The estimated fetal weight was 1724 g, with femur length and humeral length markedly reduced and inconsistent with gestational age.

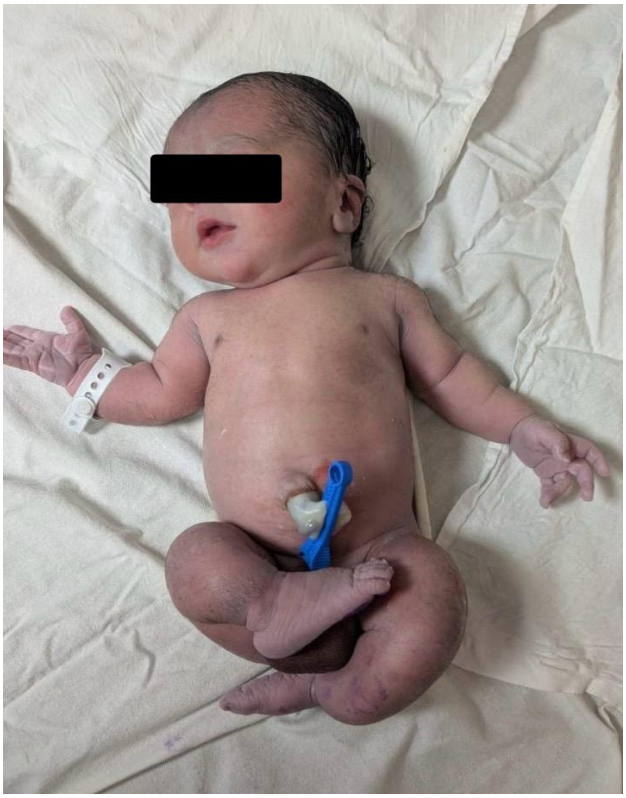

Figure 1: Showing baby on presentation post-delivery with skeletal dysplasia.

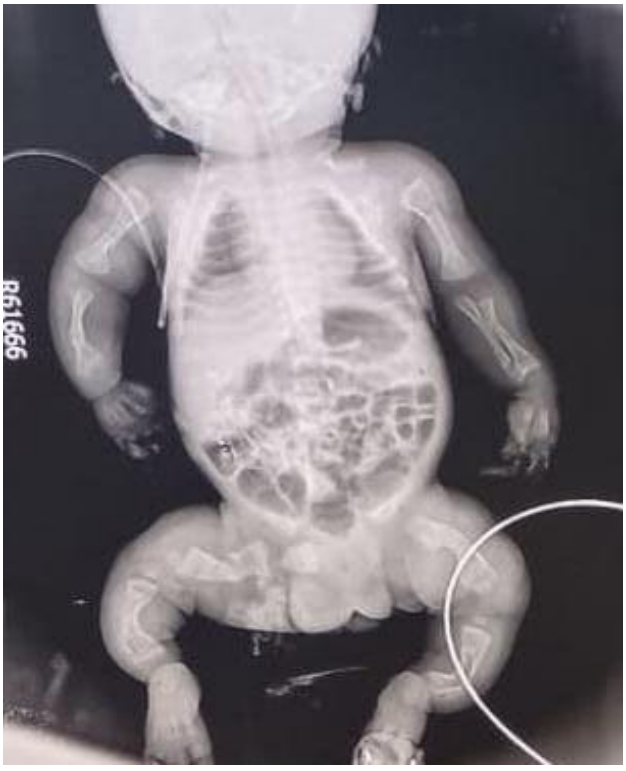

Figure 2: Infantogram suggestive of multiple fractures.

Long bones appeared shortened and abnormal, raising suspicion of an underlying skeletal disorder. The tibial length measured 46.8 mm (corresponding to approximately 23 weeks' gestation), and humeral length measured 31.2 mm (corresponding to approximately 20 weeks' gestation). Based on these findings, a provisional diagnosis of skeletal dysplasia was considered and the mother was referred to a tertiary care centre for further evaluation and management. The neonate was admitted to the Neonatal intensive care unit immediately after birth for further assessment. On examination, the baby had features suggestive of osteogenesis imperfecta, including skeletal fragility and abnormal limb proportions. There was no evidence of birth asphyxia. Systemic examination did not reveal any major visceral anomalies.

General examination

On examination of the head and face, the child had brachycephaly with widely open anterior fontanelles. The ears were low-set, and the oral cavity showed a high-arched palate. The neck appeared short.

Systemic examination

On respiratory system examination, the chest showed widely spaced nipples, and air entry was bilaterally equal. On cardiovascular examination, S1 and S2 were normal with no audible murmur. The abdomen was soft and non-tender, with no organomegaly detected.

Musculoskeletal examination

The right upper limb showed swelling over the upper one-third of the forearm, with a suspected fracture of the upper one-third of the humerus. The left upper limb appeared visually normal, and the digits were non-overlapping. Examination of the lower limbs revealed shortened lower limbs, bowing of the legs at the knee joints, bilateral knee flexion deformity, and bilateral genu varum. Additionally, bilateral congenital talipes equinovarus (CTEV) was noted.

Radiological findings

An infantogram revealed multiple skeletal injuries, including a fracture involving the upper one-third of the right humerus, fractures of the right radius and ulna and fractures of the upper one-third of the left radius and ulna. In addition, a midshaft fracture of the right femur was identified. Bilateral bowing of the tibia and fibula was also noted suggesting underlying skeletal fragility or deformity.

Laboratory findings

Laboratory evaluation revealed a normal serum calcium level, while the serum 25-hydroxyvitamin D level was reduced at 17.8 ng/ml. Renal and liver function tests were within normal limits. Haematological investigations showed a haemoglobin level of 15.8 g/dl and a haematocrit of 46.6%. The total leukocyte count was

elevated at 19,600/mm³, whereas the platelet count was within the normal range at 300,000/mm³. In view of the antenatal findings, clinical features, and biochemical profile, whole genome sequencing was performed, which revealed a mutation suggestive of a collagen-related disorder and a diagnosis of osteogenesis imperfecta was made.

The neonate was managed conservatively with gentle handling, adequate nutrition, vitamin D supplementation, calcium supplementation and multivitamin support. The neonate was administered intravenous Pamidronate therapy. Parents were counselled regarding the nature of the disease, need for long-term follow-up, fracture prevention strategies and multidisciplinary care. Currently, the child is on regular follow-up, has received a second dose of pamidronate and has failure to thrive with persistent baseline respiratory distress secondary to chest wall involvement due to skeletal dysplasia.

DISCUSSION

Systemic involvement and complications

Beyond the musculoskeletal system, OI may involve multiple organ systems, contributing significantly to morbidity and mortality. Neurological complications such as basilar invagination can result in potentially fatal brainstem compression. Cardiovascular abnormalities including aortic root dilatation and mitral valve prolapse have been reported. Progressive scoliosis may lead to restrictive lung disease, further compromising respiratory function.[2]

Historical classification

The earliest classification of OI was proposed by Looser in 1906, dividing the condition into two forms: osteogenesis imperfecta congenita (Vrolik disease), presenting at birth with severe manifestations, and osteogenesis imperfecta tarda (Ekman–Lobstein disease), characterized by later onset and milder disease.[3]

Clinical variability and disease types

OI demonstrates remarkable clinical heterogeneity, largely determined by disease subtype. Classical forms (Types I-IV) are inherited in an autosomal dominant manner, whereas more recently described types (VI-XIII) follow autosomal recessive inheritance.[5] Disease severity ranges from mild forms with infrequent fractures to perinatal lethal variants with profound skeletal dysplasia.

Molecular genetics and pathophysiology

Most cases of OI arise from mutations affecting type I collagen synthesis, structure, or processing. Glycine substitutions in COL1A1 or COL1A2 chains disrupt collagen triple helix formation and are commonly implicated in severe and lethal forms.[7] Additionally, mutations in genes such as WNT1, SERPINF1, P3H1, CREB3L1, and CRTAP have been identified, particularly in autosomal recessive OI, highlighting the genetic heterogeneity of the disorder.[7]

Advances in medical and surgical management

Although no definitive cure exists, significant advances have improved outcomes in OI. Bisphosphonate therapy has emerged as a cornerstone of management, reducing bone pain, increasing bone mineral density and decreasing fracture rates.[7] Pamidronate and zoledronate are the most commonly used agents, with zoledronate demonstrating higher potency and clinical benefit in both paediatric and adult populations.[7] Bisphosphonates also exert a valuable analgesic effect, improving quality of life.[8] Orthopaedic interventions, including intramedullary rodding, are employed to correct deformities and prevent recurrent fractures, particularly in moderate to severe disease.[4]

Emerging therapeutic strategies

Recent experimental studies have explored anabolic therapies targeting bone formation pathways. Antisclerostin antibodies and anti–transforming growth factor-beta (anti-TGFβ) antibodies have demonstrated improvements in bone mass and strength in animal models, offering promising future therapeutic avenues for patients with OI.[6]

Genetic counselling and prenatal diagnosis

Genetic counselling is of paramount importance in OI. Affected individuals have a 50% risk of transmitting the condition to offspring, while unaffected parents carry a 5-7% recurrence risk. Antenatal diagnosis is feasible between 14 and 16 weeks of gestation using ultrasonography, with findings such as severe long-bone shortening, femur length–abdominal circumference ratio <0.16, hypoplastic thorax, and intrauterine fractures. Chorionic villous sampling allows molecular confirmation in recurrent or high-risk cases.[9]

Differential diagnosis

Multiple fractures in the newborn may result from birth trauma, metabolic bone disease or other hereditary skeletal dysplasias, necessitating careful clinical, radiological and genetic evaluation to establish an accurate diagnosis.[9]

CONCLUSION

Osteogenesis imperfecta represents a complex, genetically diverse disorder with multisystem involvement. While curative therapy remains elusive, advances in pharmacological treatment, surgical techniques and genetic diagnostics have significantly improved patient outcomes. Ongoing research into novel

anabolic agents holds promise for further therapeutic breakthroughs in the future.

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