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CASE REPORT

A Neonate With Multiple Fractures

RAMESH BHAT. Y*, K. PRAKASHINI**

ABSTRACT

Fractures in a neonate commonly result from a genetic predisposition and birth trauma. Osteogenesis imperfecta, a rare heterogeneous group of disorders, mainly affecting the bones, remains the commonest cause of genetic osteoporosis. Osteogenesis imperfecta congenita (OIC) is the most severe form among them, with an incidence of 1 in 60000 live births. Presented here, is a neonate with OIC, having multiple bone fractures and deformities.

Key Words: Multiple fractures, Neonate, Osteogenesis imperfecta

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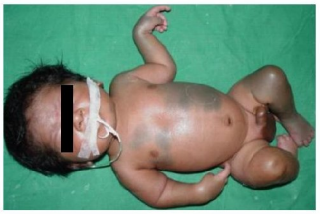
Introduction

Multiple fractures in a neonate are usually caused by genetic predisposition and birth trauma. Osteogenesis imperfecta is a rare group of disorders with genetic osteoporosis, predisposing the neonate to multiple fractures. Osteogenesis imperfecta congenita (OIC) is the most severe form among them[1],[2],[3]. The affected infant may be stillborn or may have multiple bone fractures and deformities at birth. We describe here, a neonate having severe bone manifestations.

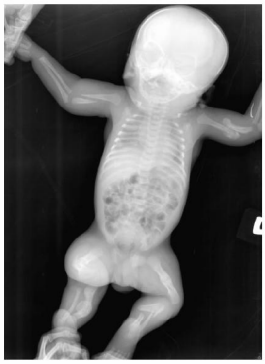
Case Report

A sixteen days old male neonate was brought with a history of multiple bruises and limb deformities from birth. He was born to a primi mother, vaginally, at home. At admission, his vitals were stable, his cry was good and he weighed 2840 gms. His head circumference was 34.5cm. His skull was soft, with wide sutures. His anterior fontanelle measured 4×4 cm and the posterior, 1cm. The sclera was white. Multiple bruises over the chest, abdomen and limbs were noticed. There were multiple deformities in the limbs, with restricted movements. The legs were bent and bowed [Table/Fig 1]. The systemic examination was normal. The radiograph showed multiple bone fractures [Table/Fig 2] involving the long bones of the upper and lower limbs and ribs. These fractures were at different stages of healing. Excessive callus at places, were also noticed. The unfractured right fibula and forearm bones were bent. The skull vault was thin, with wormian bones, in the parieto-occipital region. Multiple rib fractures showed a beaded appearance. Generalized osteopenia was noted. The neurosonogram and abdominal ultrasound were normal. A diagnosis of osteogenesis imperfecta congenita (OIC) was considered. The

conservative treatment of fractures was undertaken.



(Table/Fig 1)Clinical photograph of a neonate with multiple deformities of limbs and multiple bruises.



Table/Fig 2) Radiograph showing multiple, old and new bone fractures.

Discussion

Osteogenesis imperfecta (OI) is a heterogeneous group of genetic disorders that mainly affect the bones. Abnormalities of type I collagen due to mutations in genes encoding collagen proteins or enzymes involved in collagen biosynthesis form the pathogenetical basis of the disease. Typical manifestations are fragile bones with multiple bone fractures and bone deformities[1]. OIC or osteogenesis imperfecta type II is the most severe form, with an incidence of 1 in 60000 live births. Approximately 60 % are stillborn and most others die shortly after birth, usually from respiratory failure. Infants with OIC have bones that appear bent or crumpled. Poor foetal ossification may lead to multiple intrauterine fractures as well. They have soft and thin skull bones, a narrow chest with a small rib cage and underdeveloped lungs, blue sclerae and thin and fragile skin. Roentgenograms show the crumpled appearance of the long bones, especially of the femora and multiple fractures of the ribs. Our case had most of these manifestations. Diffuse osteopenia was observed. The inheritance pattern of OIC is said to be autosomal recessive, but a dominant new mutation may also be responsible. Ultrasonography is useful for prenatal diagnosis [2]. Polymerase chain reaction based assays are suggested for preimplantation genetic diagnosis of milder varieties.

For an affected infant, an aggressive multidisciplinary rehabilitative approach is indicated to optimize functional ability and walking [3],[4],[5] The fractures continue to occur due to the extreme bone fragility and deformities. The number of fractures and deformities and the age at which they begin, greatly influence the prognosis, the achievement of walking and autonomy. Three types of treatment are available: nonsurgical management (physical therapy, rehabilitation, bracing and splinting), appropriately timed surgery (intramedullary rodding, spinal and basilar impression surgery) and drugs to increase the strength of the bone and to decrease the number of fractures. The conservative treatment of fractures includes a foam mould for the torso and limbs and additional support for specific unstable painful fractures. Physical and occupational therapy for increased stability of bone, improved mobility, prevention of contractures, prevention of head and spinal deformity and muscle strengthening remain an integral part of the treatment protocol.

Surgical correction of deformities and internal splinting of the long bones with intramedullary rods may be required. The combination of external and internal fixation and electric wheel chairs, greatly reduce the frequency of fractures and facilitate the general care and development [3],[4]. Surgery may also be required in patients with progressive spinal deformity and in those with symptomatic basilar impression. In recent years, growth hormone (GH) and bisphosphonate agents have been used in OI therapy. The beneficial effect of GH therapy

(a positive effect on bone turnover, bone mineral density and height velocity rate) has been reported in patients with moderate forms of OI.

Bisphosphonates have proved beneficial and have become the standard of care in children with severe OI [5]. Therapy has also reduced the need of surgical procedures. Bisphosphonate treatment increases bone mineral density and the level of ambulation and reduces the fracture rate and bone pain with good short term safety. Effects on bone include increase in size of vertebral bodies and thickening of cortical bone. These results allow for more efficacious corrective surgery using intramedullary rodding of the long bones and paravertebral instrumentation. Cyclical intravenous pamidronate administration includes 3-day cycles, every 2 to 4 months depending on the age of the patients (the younger the patient, the shorter the interval between cycles), with an annual dose of 9 mg/kg per year. Within 1 to 2 weeks after the first infusion cycle, considerable decrease in bone pain and often, complete disappearance have been reported [5]. Their patients also felt more energetic. A newer bisphosphonate, Zoledronic acid, with a longer half life, greater potency, and more convenient dosing, is being studied in children with OI. Gene-based therapy may be the answer in future.

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