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Osteogenesis Imperfecta in a Neonate: Case Report

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Abstract

Osteogenesis imperfecta (OI) is a rare group of inherited connective tissue disorders characterized by increased bone fragility. Diagnosis of OI is mainly based on the clinical features of the disorder. We report, the case of a male neonate delivered via cesarean section at 38 weeks of gestation to a 20-year-old mother who has no family history suggestive of OI. He had clinical features of a type II OI and had a multidisciplinary approach to care. He was discharged for follow-up with pediatric and orthopedic specialists.

Keywords: Osteogenesis imperfect; Brittle bone disease and fragile bone disease; Term; Neonate

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Introduction:

Osteogenesis imperfecta (OI) is a rare group of inherited connective tissue disorders characterized by increased bone fragility (Smith R, 1984). The condition occurs in approximately 1 in 20,000 live births and results from mutations in the collagen, type I, alpha 1 and collagen type I alpha 2 genes, which encode the alpha 1 and the alpha 2 chain of type I procollagen, respectively (Burton BK et al. 2005; Marini JC et al. 2011). OI is usually inherited in an autosomal dominant manner, although autosomal recessive variants have also been reported (Ward LM et al. 2002; Glorieux FH et al. 2000).

The clinical spectrum of OI is broad, ranging from perinatally lethal forms to mild presentations that may go undiagnosed until adulthood (Marini JC et al. 2011). Four main types have been traditionally described: Type I is the mildest and most common, while Type II is the most severe (Marini JC et al. 2011). We report a case of a male neonate with osteogenesis imperfecta due to its rarity and full-term presentation.

Case presentation

A male neonate was delivered via cesarean section at 38 weeks of gestation with a birth weight of 2,560 grams. His transition to extrauterine life was uneventful. His mother attended regular antenatal visits, received only prescribed medications, and antenatal ultrasonography revealed no evidence of fractures or fetal anomalies. There was no family history of recurrent fractures or bone fragility.

On physical examination, the neonate had blue-grey sclera, a large anterior fontanelle, an open posterior fontanelle, and widely separated cranial sutures. Deformities of his limbs were noted, with limited spontaneous movement and palpable crepitus over the left humerus and both femora (Figure 1).

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A diagnosis of osteogenesis imperfecta type II was made. Radiographic evaluation showed fractures of both humeral and femoral long bones and the left tibia, with generalized osteopenia (figure 2). The orthopedic team applied splints to the affected limbs, and the infant was discharged for follow-up with pediatric and orthopedic specialists.

Figure 1: Neonate with Osteogenesis imperfect

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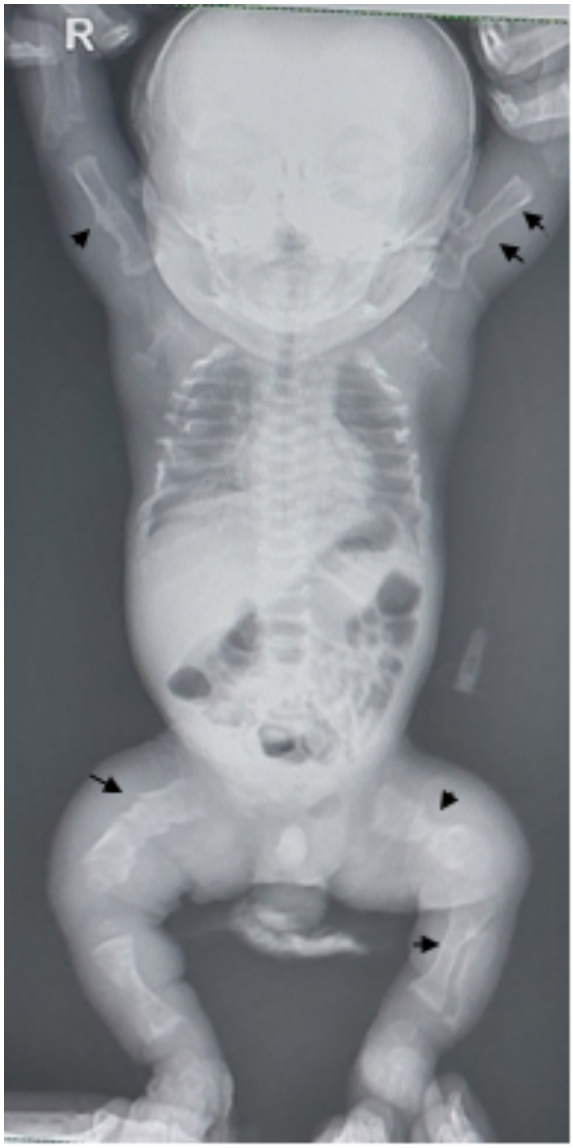

Figure 2: Multiple fractures with generalized osteopenia

Discussion

Osteogenesis imperfecta, or brittle bone disease, encompasses a group of genetic connective tissue disorders characterized by bone fragility and fractures occurring with minimal or no trauma (Smith R, 1984). The clinical presentation varies widely—from

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perinatal lethality to mild adult-onset forms (Marini JC et al. 2011).

Type I OI is the mildest and most prevalent form, typically presenting with fractures during early childhood when the child begins to walk. Fractures during the neonatal period are uncommon, and their frequency usually decreases after puberty (Marini JC et al. 2011). In contrast, Type II OI represents the most severe form, often resulting in perinatal death. Affected neonates may present with multiple fractures at birth, short limbs, a large fontanelle, bluish sclera, fragile skin, and hypotonia (Marini JC et al. 2011).

Types III and IV represent intermediate phenotypes with normal sclera. Type III is the most severe non-lethal form, associated with significant bone deformities due to multiple congenital fractures. Type IV has subtypes A and B, distinguished by the presence or absence of dentinogenesis imperfect (Sillence DO et al. 1979).

Diagnosis of OI is primarily clinical, supported by radiographic findings. Molecular genetic testing or collagen analysis of cultured skin fibroblasts can confirm the diagnosis and help distinguish OI from conditions such as child abuse (Rauch F et al. 2004; Marini JC et al. 2011). Prenatal diagnosis is possible by ultrasonography at 14–16 weeks of gestation, identifying features such as markedly short-long bones, a femur length-to-abdominal circumference ratio below 0.16, a hypoplastic thorax, and evidence of fractures or bowing (Parilla BV et al; 2003; Teng SW et al. 2003).

There is currently no definitive cure for OI. Management focuses on minimizing fractures and deformities while optimizing functional independence. A multidisciplinary approach involving orthopedic surgery, physiotherapy, counseling, and rehabilitation is essential in improving quality of life. In

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addition, pharmacological therapy with bisphosphonates has shown promise in increasing bone density, reducing fracture rates, and improving growth outcomes, but prognosis still varies depending on the severity and number of clinical features present (Rauch F et al. 2004; Polousky JD et al. 2009; Marini JC et al. 2011).

Conclusion

Osteogenesis imperfecta is a rare hereditary disorder requiring lifelong, multidisciplinary management to minimize fractures, prevent deformities, and improve overall functional outcomes.

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