Navigating the challenges of severe osteogenesis imperfect in the perinatal period.

Rick Carrey*

Department of Pediatrics, Children's Hospital of Philadelphia, United States

Introduction

The perinatal period, spanning the time from conception to a few weeks after birth, is a critical phase in the life of both the newborn and their family. While this period is often marked by anticipation and joy, it can also bring unforeseen challenges, such as the diagnosis of severe osteogenesis imperfecta (OI). OI, a rare genetic disorder affecting the bones, can present profound medical and emotional complexities for families navigating the perinatal journey [1,2].

Osteogenesis imperfecta is a group of genetic disorders characterized by fragile bones that are prone to fractures. The severity of OI can vary, with some individuals experiencing milder symptoms and others facing significant challenges. In the perinatal period, the diagnosis of severe OI can be particularly challenging for parents as they grapple with the immediate medical needs of their newborn and the potential long-term implications for their child's quality of life. Severe OI is often detected through prenatal imaging, such as ultrasounds, which may reveal skeletal abnormalities indicative of the condition. The diagnosis can be emotionally overwhelming for parents, as they are confronted with the reality of a genetic disorder that affects the structural integrity of their child's bones. In these moments, healthcare professionals play a crucial role in providing accurate information, emotional support, and a pathway for understanding the complexities of severe OI [3,4].

The challenges associated with severe OI in the perinatal period extend beyond the physical aspects of the disorder. Parents may grapple with feelings of guilt, grief, and uncertainty about their child's future. The fragility of bones in infants with severe OI often requires careful handling, adding an extra layer of stress for parents who may feel a heightened sense of responsibility for their child's well-being. Medical interventions in the perinatal period for severe OI can be intricate, involving specialized neonatal care and collaboration between various healthcare professionals. The primary focus is on managing fractures, preventing deformities, and ensuring adequate respiratory function. This may include the use of supportive measures such as respiratory assistance and careful handling techniques to minimize the risk of fractures [5,6].

Beyond the immediate medical needs, families dealing with severe OI in the perinatal period may also face decisions about long-term care and quality of life. This involves discussions about mobility aids, orthopedic interventions, and ongoing medical monitoring to address the unique challenges associated with severe OI. Genetic counseling becomes an essential component, providing families with information about the inheritance pattern of OI and the potential risks in future pregnancies. Severe osteogenesis imperfecta in the perinatal period presents families with a unique set of challenges that extend beyond the immediate medical needs of the newborn. The emotional and psychological impact of the diagnosis on parents requires thoughtful and compassionate support from healthcare professionals, emphasizing open communication and access to resources that facilitate understanding [7,8].

In the face of severe OI, medical interventions in the perinatal period are crucial for managing fractures and preventing complications. The collaboration between healthcare providers, including neonatologists, orthopedic specialists, and genetic counselors, is paramount in ensuring a comprehensive and holistic approach to the care of infants with severe OI. As families embark on the journey of raising a child with severe OI, ongoing support and education are essential components of navigating the complexities of the disorder. This includes access to specialized medical care, information about adaptive technologies and interventions, and a network of support from healthcare professionals, support groups, and community organizations [9,10].

Conclusion

Severe osteogenesis imperfecta in the perinatal period serves as a poignant reminder of the resilience and strength of families facing unexpected challenges. By fostering a collaborative and supportive healthcare environment, we can empower parents to navigate the complexities of severe OI with confidence, ensuring that every child, regardless of their medical condition, has the opportunity to thrive and experience a fulfilling life surrounded by care and understanding.

References

1. Gatti D. Teriparatide Treatment in Adult Patients with Osteogenesis Imperfecta Type I. Calcif. Tissue Int. 2013;93:448–452.
2. Orwoll E.S. Evaluation of teriparatide treatment in adults with osteogenesis imperfecta. J. Clin. Investig. 2014;124:491–498.

***Correspondence to:** Rick Carrey, Department of Pediatrics, Children's Hospital of Philadelphia, United States, E-mail: carreyr@nemours.org

***Received:** 02-Dec-2023, Manuscript No. AAJCAH-23-122887; **Editor assigned:** 04-Dec-2023, Pre QC No. AAJCAH-23-122887(PQ); **Reviewed:** 18-Dec-2023, QC No. AAJCAH-23-122887; **Revised:** 22-Dec-2023, Manuscript No. AAJCAH-23-122887(R); **Published:** 30-Dec-2023, DOI: 10.35841/aajcah-7.6.185*

***Citation:** Carrey R. Navigating the challenges of severe osteogenesis imperfect in the perinatal period. J Child Adolesc Health. 2023;7(6):185*

3. Hoyer-Kuhn H. Two years' experience with denosumab for children with Osteogenesis imperfecta type VI. Orphanet J. Rare Dis. 2014;9:1–8.
4. Grafe I. Excessive transforming growth factor-β signaling is a common mechanism in osteogenesis imperfecta. Nat. Med. 2014;20:670–675.
5. Wang Q.. Alternative Splicing in COL1A1 mRNA Leads to a Partial Null Allele and Two In-frame Forms with Structural Defects in Non-lethal Osteogenesis Imperfecta. J. Biol. Chem. 1996;271:28617–28623.
6. Niyibizi C. Gene therapy approaches for osteogenesis imperfecta. Gene Ther. 2004;11:408–416.
7. Sagar R. Fetal Mesenchymal Stromal Cells: An Opportunity for Prenatal Cellular Therapy. Curr. Stem Cell Rep. 2018;4:61–68.
8. Niyibizi C.. Potential implications of cell therapy for osteogenesis imperfecta. Int. J. Clin. Rheumatol. 2009;4:57–66.
9. Götherström C. Stem Cell Therapy as a Treatment for Osteogenesis Imperfecta. Curr. Osteoporos. Rep. 2020;18:337–343.
10. Allyse M.. Non-invasive prenatal testing: A review of international implementation and challenges. Int. J. Women's Health. 2015;7:113–126.

Citation: *Carrey R. Navigating the challenges of severe osteogenesis imperfect in the perinatal period. J Child Adolesc Health. 2023;7(6):185*