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Aortopulmonary window in an infant with type i osteogenesis imperfecta: Case report

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Abstract

Some studies have shown an association between osteogenesis imperfecta and congenital heart diseases, but only those involving changes in the connective tissue of heart structures, such as heart valves, chordae tendineae, fibrous rings, ventricular septum and aortic root (dilatation). The concomitant presence of osteogenesis imperfecta and aortopulmonary window has not been reported in the specialized literature, rendering the present case report uncommon. We report the case of a male infant aged 2 months and 15 days, diagnosed with type I osteogenesis imperfecta and type I aortopulmonary window, submitted to surgery to completely repair his heart disease. In addition, we provide a literature review and discuss the clinical and surgical approaches to this infant, emphasizing that previous multidisciplinary planning is essential for a successful outcome.

Introduction : Osteogenesis imperfecta (OI) is a genetically heritable disease characterized by imperfect bone formation, which leads to bone fragility and bone mass loss1. This occurs because both the structure and function of type 1 collagen are affected by gene mutations, the most common being an autosomal dominant change in chromosomes 7 and 17, in the COL1A1 and COL1A2 loci, where glycine is replaced with another amino acid2. Osteogenesis imperfecta has an incidence of approximately 1 per 15,000-20,000 births, and a heterogeneous clinical presentation, ranging from asymptomatic to lethal3.

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