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Inborn Error of Metabolism Leads to Decreased Bone Quality

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DESCRIPTION

Children are prone to upper cervical spine injuries because of their relatively immature bones, lax ligaments, underdeveloped muscles, and large head-to-body mass ratio. Odontophyte fractures with chondrosis are one of the most common of these injuries. Many of these fractures can be treated conservatively with external splints, but fractures with significant displacement cannot be reduced and require surgical management. In these cases, most patients undergo posterior C1-2 fusion with arthrodesis, permanently limiting atlantoaxial Range of Motion (ROM). Here, we present a novel surgical approach to treat Densoidal Chondrotic fractures using temporary internal braces with a posterior C1-2 instrument without arthrodesis.

Hypophosphatasia (HPP) is an inherited musculoskeletal disorder caused by variant inactivation of the ALPL gene and subsequent decreased serum Tissue-Nonspecific Alkaline Phosphatase (TNSALP) activity. Increased incidence of fractures and prolonged bone healing are hallmarks of HPP. Available enzyme replacement therapy (asfotase alfa) has been reported to restore bone mineralization and bone quality in her adult HPP patients. Furthermore, asfotase alfa was shown to improve fracture healing from previous non-unions in two adult HPP patients. We hypothesize that the articular joints are partially filled with osteoids, offering great potential for treatment with asfotase alfa to promote bone healing. In this study, we report pediatric HPP and her three adult patients with her documented ALPL mutation with prolonged bone healing after arthrodesis, tibial stress fracture, and osteotomy. After initiation of treatment with asfotase alfa, biochemical analyzes quickly revealed increased levels of alkaline phosphatase (ALP) and bone-specific ALP and decreased levels of pyridoxal-5-phosphate (PLP). Importantly, even after up to 5 years without healing, progressive consolidation was demonstrated as assessed by custom three-dimensional analysis of repeated Cone-Beam Computed Tomography (CBCT) images. This is the bone volume per tissue volume (BV/TV) within the volume of interest (i.e., the area of non-healing bone). These radiographic findings were consistent with reported functional recovery, painless full-body weight bearing, and increases in neuromuscular parameters, such as improved muscle strength. Taken together, our results suggest that asfotase alfa improves non-union Osteosclerosis, possibly by re-mineralizing the anterior gap-filling osteoid tissue, and improves function in adult HPP patients characterized by increased BV/TV.

Osteogenesis Imperfecta (OI) is a genetic disorder characterized by frequent fractures and bone loss. Here, we describe a 5-year-old boy with clinical, radiological, and bone ultrastructural features typical of OI type 1. Determining the molecular genetic cause of his condition proved difficult as clinical exome and whole-exome analyzes were repeatedly reported negatively. Finally, manual analysis of exome. This indicates that it activates cryptic splice sites. *In vitro* studies of collagen expression confirmed cell accumulation and decreased COL1A2 secretion by 45%. This is the first report of potential splice sites within the COL1A2 coding region. It causes aberrant splicing and causes. Our experience with this case shows that routine diagnostic approaches may miss cryptic splice variants in causative genes due to the lack of universally applicable splice site prediction algorithms. For exome-negative cases, detailed analysis of common causative genes should be performed and trio-exome analysis is recommended.

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CONFLICT OF INTEREST

The authors declare that they have no conflict of interest.

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