Hypophosphatasia

Hypophosphatasia (HPT) and cleidocranial dysplasia (CCD) are rare genetic disorders characterized by both defective ossification and bone mineralization. Patients usually present with craniosynostosis and cranial defects which in many cases require surgical repair. There is only 1 reported case of combined HPT and CCD in the literature.

The reported case of Blionas et al. involves a 3.5-year-old girl with concomitant homozygous CCD and heterozygous HPT. The child had an extended cranial defect since birth which improved with the administration of Strensiq and was followed until preschool age. Bone defects were relatively minor on revaluation. Due to the limited final defect, we decided not to intervene. In HPT-CCD patients, bone defects are overestimated due to osteomalacia, and thus, management strategy should be less aggressive. They should undergo surgical repair with cranioplasty with the use of cement and/or titanium meshes in case of extended final defects [1].

1)
Blionas A, Friehs GM, Zerris VA. Hypophosphatasia and cleidocranial dysplasia-a case report and review of the literature: the role of the neurosurgeon. Childs Nerv Syst. 2021 Jun 15. doi: 10.1007/s00381-021-05261-1. Epub ahead of print. PMID: 34131769.

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