Osteogenesis imperfecta (OI)

Osteogenesis imperfecta (OI) represents a rare heterogeneous group of inherited disorders characterized by low bone mass, increased bone fragility, and other connective tissue manifestations. This condition can contribute to dramatic complications after a seemingly insignificant injury.

Typically includes fractures; bowed, irregular thickened bones; short/normal size extremities; soft, thin skull.

The finding of premature osteoporosis in persons with osteogenesis imperfecta suggests a possible role for the type I collagen genes in bone integrity. However, the relative importance of any one gene, particularly that of specific VDR genotypes, has been controversial.

Cranial base pathology is a serious complication of osteogenesis imperfecta (OI), and may develop despite bisphosphonate treatment. Early initiation of bisphosphonate treatment may delay development of craniocervical junction pathology. Careful followup of cranial base morphology is warranted, particularly in patients with severe OI [1].

see Intracranial epidural hematoma in osteogenesis imperfecta.

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Arponen H, Vuorimies I, Haukka J, Valta H, Waltimo-Sirén J, Mäkitie O. Cranial base pathology in pediatric osteogenesis imperfecta patients treated with bisphosphonates. J Neurosurg Pediatr. 2015 Mar;15(3):313-20. doi: 10.3171/2014.11.PEDS14113. Epub 2015 Jan 10. PubMed PMID: 25559924.