Osteopetrosis

AKA “marble bone disease” (there is also some confusion with the term osteosclerosis; osteosclerosis fragilis generalisata is the obsolete term for osteopetrosis). A rare group of genetic disorders of defective osteoclastic resorption of bone resulting in increased bone density, may be transmitted either as autosomal dominant or recessive [1].

The dominant form is usually benign and is seen in adults and adolescents. The recessive (“malignant”) form is often associated with consanguinity and is similar to hyperostosis cranialis interna, but in addition to the proclivity for the skull, it also involves ribs, clavicles, long bones, and pelvis (long-bone involvement results in the destruction of marrow and subsequent anemia). Cranial nerves involved primarily include optic (optic atrophy and blindness are the most common neurologic manifestation), facial, and vestibulo-acoustic (with deafness); trigeminal nerve may also be involved. There may also be extensive intracranial calcifications, hydrocephalus, intracranial hemorrhage, and seizures. Bilateral optic nerve decompression via a supraorbital approach may improve or stabilize vision [2].

An Exceptional Neurosurgical Presentation of a Patient with Osteopetrosis [3].

[1], [2]
Al-Mefty O, Fox JL, Al-Rodhan N, et al. Optic Nerve Decompression in Osteopetrosis. J Neurosurg. 1988; 68:80–84

[3]
Isler C, Kayhan A, Ugurlar D, Hanimoglu H, Ulu MO, Uzan M, Erdincler P, Ozlen F. An Exceptional Neurosurgical Presentation of a Patient with Osteopetrosis. World Neurosurg. 2018 Jun 20. pii: S1878-8750(18)31297-X. doi: 10.1016/j.wneu.2018.06.081. [Epub ahead of print] PubMed PMID: 29935324.