Opis choroby *

Definicja

A rare primary bone dysplasia with increased bone density characterized by slowly progressive endosteal hyperostosis and osteosclerosis exclusively of the skull base and the calvaria, resulting in entrapment and dysfunction of cranial nerves I, II, V, VII, and VIII. First symptoms often appear during the second decade of life and include disturbances in smell, vision, facial sensation and expression, hearing, and balance, as well as headaches due to increased ocular and intracranial pressure. After the fourth decade, radiological progression is minimal, although decreased intracranial volume can lead to death in severe cases.

Dane

Klasyfikacja
Choroba

Kod ORPHA
443098

Kod OMIM
144755

Kod ICD10
M85.2

Kod ICD11
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*Źródło

orphanet