

Opis choroby *

Definicja

Hyperostosis corticalis generalisata, also known as van Buchem disease, is a rare craniotubular hyperostosis characterized by hyperostosis of the skull, mandible, clavicles, ribs and diaphyses of the long bones, as well as the tubular bones of the hands and feet. Clinical manifestations include increased skull thickness with cranial nerve entrapment causing inconsistent cranial nerve palsies.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Hyperphosphatasemia tarda Choroba Van Buchema Hiperfosfatemia późna Van Buchem disease

Kod ORPHA	Kod OMIM	Kod ICD10
3416	144750	M85.2

Kod ICD11
LD24.1Y

*Źródło

orphanet