

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

Zespół wad wrodzonych

Synonimy

Hyperphosphatasemia tarda

Choroba Van Buchema

Hiperfosfatemia późna

Van Buchem disease

Kod ORPHA

3416

Kod OMIM

144750

Kod ICD10

M85.2

Kod ICD11

LD24.1Y

*Źródło

orphanet