

Opis choroby *

Definicja

Sclerosteosis is a very rare serious sclerosing hyperostosis syndrome characterized clinically by variable syndactyly and progressive skeletal overgrowth (particularly of the skull), resulting in distinctive facial features (mandibular overgrowth, frontal bossing, midfacial hypoplasia), cranial nerve entrapment causing facial palsy and deafness, and potentially lethal elevation of intracranial pressure.

Dane

Klasyfikacja

Zespół wad wrodzonych
Cortical hyperostosis-syndactyly syndrome
Hiperostoza korowa - syndaktylia

Kod ORPHA

3152

Kod OMIM

614305

Kod ICD10

M85.2

Kod ICD11

LD24.1Y

*Źródło

orphanet