

Tłuszczakowatość mózgowo-czaszkowo-skórna

Kod Orpha: 2396 Kod OMIM: 613001

Opis choroby *

Definicja

A rare, genetic skin disease characterized by the ocular, cutaneous, and central nervous system anomalies. Typical clinical features include a well-demarcated hairless fatty nevus on the scalp, benign ocular tumors, and central nervous system lipomas, leading sometimes to seizures, spasticity, and intellectual disability. Nevus psiloliparus, focal dermal hypo- or aplasia, eyelid skin tags, colobomas, abnormal intracranial vessels, hemispheric atrophy, porencephalic cyst, and hydrocephalus have also been associated.

Dane

Klasyfikacja

Choroba

Synonimy

Haberland syndrome
Zespół Haberlanda

Kod ORPHA

2396

Kod OMIM

613001

Kod ICD10

E88.2

Kod ICD11

EF02.1

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.