

# 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

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#### [\\*Źródło](#)

orphanet

### Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.