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