

Opis choroby *

Definicja

A new leukoencephalopathy, the CACH syndrome (Childhood Ataxia with Central nervous system Hypomyelination) or VWM (Vanishing White Matter) was identified on clinical and MRI criteria. Classically, this disease is characterized by (1) an onset between 2 and 5 years of age, with a cerebello-spastic syndrome exacerbated by episodes of fever or head trauma leading to death after 5 to 10 years of disease evolution, (2) a diffuse involvement of the white matter on cerebral MRI with a CSF-like signal intensity (cavitation), (3) a recessive autosomal mode of inheritance, (4) neuropathologic findings consistent with a cavitating orthochromatic leukodystrophy with increased number of oligodendrocytes with sometimes "foamy" aspect.

Dane

Klasyfikacja

Choroba

Synonimy

Childhood ataxia with diffuse central nervous system hypomyelination

Dziecięca ataksja z rozlaną hipomielinizacją centralnego układu nerwowego

Leukoencefalopatia z zanikającą istotą białą

Myelinosis centralis diffusa

Leukoencephalopathy with vanishing white matter

Myelinosis centralis diffusa

Kod ORPHA

135

Kod OMIM

615889

Kod ICD10

E75.2

Kod ICD11

8A44.3

*Źródło

orphanet