

# Niedobór ceramidazy alkalicznej 3

## Kod Orpha: 502444 Kod OMIM: 617762

### Opis choroby \*

#### Definicja

A rare genetic leukodystrophy characterized by infantile onset of stagnation and regression of motor and language development resulting in complete lack of communication and purposeful movement. Further neurological manifestations include truncal hypotonia, appendicular spasticity, dystonia, optic disc pallor, peripheral neuropathy, and neurogenic bladder. Patients also present multiple contractures, late-onset relative macrocephaly, short stature, and facial dysmorphism (including coarse facial features, sloping forehead, thick eyebrows, low-set ears, prominent nose, flat philtrum, and prominent lower lip). Brain imaging at advanced stages shows diffuse abnormal white matter signal and severe atrophy. Sural nerve biopsy reveals decreased myelination.

#### Dane

#### Klasyfikacja

Choroba

#### Synonimy

ACER3-related early childhood-onset progressive leukodystrophy  
Lekodystrofia z powodu niedoboru ceramidazy alkalinowej 3  
Leukodystrophy due to alkaline ceramidase 3 deficiency

#### Kod ORPHA

502444

#### Kod OMIM

617762

#### Kod ICD10

E75.2

#### Kod ICD11

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[\\*Źródło](#)

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## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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