

# Niedobór dehydratazy NAD(P)HX

## Kod Orpha: 555402 Kod OMIM: 618321

### Opis choroby \*

#### Definicja

A rare neurometabolic disease characterized by infantile onset of repeated episodes of developmental regression and neurodegeneration, often triggered by febrile illnesses. Patients present with lethargy, hypotonia, irritability, gait ataxia, loss of speech, movement disorder, seizures, ophthalmoplegia, and hearing loss. Brain imaging shows generalized cerebral atrophy and bilateral basal ganglia abnormalities. Extensive skin lesions, cardiomyopathy, and pancytopenia have been reported in association. The condition is fatal in the first years of life.

#### Dane

#### Klasyfikacja

Choroba

#### Synonimy

CARKD deficiency

CARKD deficiency

#### Kod ORPHA

555402

#### Kod OMIM

618321

#### Kod ICD10

E88.8

#### Kod ICD11

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#### [\\*Źródło](#)

orphanet

### Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.