

# Niedobó hydrolazy S-adenozylhomocysteiny

## Kod Orpha: 88618 Kod OMIM: 613752

### Opis choroby \*

#### Definicja

A rare, multisystemic inherited metabolic diseases characterized clinically, by a variable spectrum of severity, primarily comprised of psychomotor delay, myopathy and liver dysfunction. Most patients present in infancy, but the onset can be already *in utero* or in adult age. Hypermethioninemia is frequent, but often absent in infancy. Creatine kinase is elevated in most patients.

#### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Hipermetioninemia spowodowana niedoborem hydrolazy S-adenozylhomocysteiny

#### Kod ORPHA

88618

#### Kod OMIM

613752

#### Kod ICD10

E72.1

#### Kod ICD11

5C50.B

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#### [\\*Źródło](#)

orphanet

### Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.