

## Opis choroby \*

### Definicja

A rare inborn error of metabolism characterized by increased concentrations of sarcosine in plasma and urine due to sarcosine dehydrogenase deficiency. The condition is considered benign and not associated with any specific clinical phenotype. Mode of inheritance is autosomal recessive.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Sarcosine dehydrogenase complex deficiency

Niedobór kompleksu dehydrogenazy sarkozyny

#### Kod ORPHA

3129

#### Kod OMIM

268900

#### Kod ICD10

E72.5

#### Kod ICD11

5C50.71

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#### \*Źródło

orphanet