

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

*Źródło

orphanet