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