

Opis choroby *

Definicja

A rare inborn error of metabolism characterized by low serum carnosinase activity, persistent carnosinuria, and carnosinemia. The clinical phenotype is highly variable, with some patients remaining asymptomatic, while others have been reported to show severe developmental delay, intellectual disability, hypotonia, seizures, and other neurological signs and symptoms.

Dane

Klasyfikacja

Wada biologiczna

Synonimy

Deficyt karnozydazy

Kod ORPHA

1361

Kod OMIM

212200

Kod ICD10

E70.8

Kod ICD11

5C50.F1

*Źródło

orphanet