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

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*Źródło

orphanet