Opis choroby *

Definicja

A rare metabolic disorder belonging to the neutral aminoacidurias, mainly characterized by skin photosensitivity, ocular and neuropsychiatric features, due to abnormal renal and gastrointestinal transport of neutral amino acids (tryptophan, alanine, asparagine, glutamine, histidine, isoleucine, leucine, phenylalanine, serine, threonine, tyrosine and valine).

Dane

Klasyfikacja
Choroba

Synonimy
Aminoaciduria, Hartnup type
Aminoaciduria typu Hartnupów
Choroba Hartnupów
Hartnup disorder

Kod ORPHA
2116

Kod OMIM
234500

Kod ICD10
E72.0

Kod ICD11
5C60.Y

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*Źródło

orphanet