

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

Kod ORPHA	Kod OMIM	Kod ICD10
2116	234500	E72.0

Kod ICD11

5C60.Y

*Źródło

orphanet