

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

*Źródło

orphanet