

Opis choroby *

Definicja

A rare, genetic disorder of tryptophan metabolism characterized by massive urinary excretion of xanthurenic acid (XA), 3-hydroxykynurenine and kynurenine and increased XA concentration in plasma. The clinical phenotype is highly variable, ranging from asymptomatic or mild cases presenting with jaundice and vomiting, with subsequent normal development and growth, to more severe cases with manifestations which include intellectual disability, cerebellar ataxia, pellagra, progressive encephalopathy with muscular hypotonia, global developmental delay, stereotyped gestures and/or congenital deafness.

Dane

Klasyfikacja

Choroba

Synonimy

Kynureninase deficiency
Acyduria ksanturenowa
Niedobór kinureninazy
Xanthurenic aciduria

Kod ORPHA

79155

Kod OMIM

236800

Kod ICD10

E70.8

Kod ICD11

5C50.3

*Źródło

orphanet