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 presentating with jaundice and vomiting, with subsequent normal development and growth, to more severe cases with manifestions 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

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*Źródło

orphanet