Opis choroby *

Definicja

A rare genetic disorder of pyrimidine metabolism characterized by early onset of megaloblastic anemia, global developmental delay, and failure to thrive, associated with massive urinary overexcretion of orotic acid (sometimes with orotic acid crystalluria). Patients without megaloblastic anemia, but with additional manifestations such as epilepsy, have also been reported.

Dane

Klasyfikacja
Choroba

Synonimy
Orotidylic decarboxylase deficiency
Kwasica orotowa
Niedobór dekarboksylazy orotydyny
Niedobór syntetazy monofosforanu urydyny
Uridine monophosphate synthetase deficiency

Kod ORPHA
30

Kod OMIM
258900

Kod ICD10
E79.8

Kod ICD11
3A03.0

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*Źródło

orphanet