

## 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