Opis choroby *

Definicja

Dihydropyrimidinase (DPD) deficiency is a very rare pyrimidine metabolism disorder with a variable clinical presentation including gastrointestinal manifestations (feeding problems, cyclic vomiting, gastroesophageal reflux, malabsorption with villous atrophy), hypotonia, intellectual deficit, seizures, and less frequently growth retardation, failure to thrive, microcephaly and autism. Asymptomatic cases are also reported. DPD deficiency increases the risk of 5-FU toxicity.

Dane

Klasyfikacja
Choroba

Synonimy
Dihydropyrimidinase deficiency
Niedobór dihydropirymidynazy

Kod ORPHA
38874

Kod OMIM
222748

Kod ICD10
E79.8

Kod ICD11
5C55.1

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*Źródło

orphanet