

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

*Źródło

orphanet