

Dihydropirimidynuria

Kod Orpha: 38874 Kod OMIM: 222748

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	Synonimy
Choroba	Dihydropyrimidinase deficiency Niedobór dihydropirymidyny

Kod ORPHA	Kod OMIM	Kod ICD10
38874	222748	E79.8

Kod ICD11
5C55.1

*[Źródło](#)

[orphanet](#)

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.