Dihydropyrimidinuria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Dihydropyrimidinuria. ORPHA:38874

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.