

Opis choroby *

Definicja

A rare disorder of pyrimidine metabolism characterized by a variable phenotype ranging from absence of symptoms to severe neurological involvement with developmental delay, intellectual disability, and seizures. Additional signs and symptoms may include hypotonia, microcephaly, ocular abnormalities (such as microphthalmia, nystagmus, and strabismus), and autistic behavior, among others. Analysis of urine typically shows high levels of uracil and thymine. Patients are at risk of suffering from severe toxicity after the administration of the anti-neoplastic agent 5-fluorouracil.

Dane

Klasyfikacja

Choroba

Synonimy

Familial pyrimidinemia

Pirymidynemia rodzinna

Kod ORPHA

1675

Kod OMIM

274270

Kod ICD10

E79.8

Kod ICD11

5C55.1

*Źródło

orphanet