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

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*Źródło

orphanet