

Opis choroby *

Definicja

A rare disorder of folate metabolism and transport characterized, biochemically, by elevated formiminoglutamate in urine and plasma due to glutamate formiminotransferase deficiency, associated with a highly variable clinical phenotype, ranging from developmental delay, intellectual disability and anemia to normal development without anemia. Increased hydantoin-5-propionic acid and/or folate in plasma may also be associated.

Dane

Klasyfikacja

Choroba

Synonimy

FTCD deficiency

Glutamate formiminotransferase deficiency

Niedobór cyklodeaminazy formiminotransferazy

Niedobór FTCD

Formiminotransferase cyclodeaminase deficiency

Glutamate formiminotransferase deficiency

Kod ORPHA

51208

Kod OMIM

229100

Kod ICD10

E70.8

Kod ICD11

3A02.Y

[*Źródło](#)

orphanet