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

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*Źródło

orphanet