

## 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