

## Opis choroby \*

### Definicja

MPI-CDG is a form of congenital disorders of N-linked glycosylation, characterized by cyclic vomiting, profound hypoglycemia, failure to thrive, liver fibrosis, gastrointestinal complications (protein-losing enteropathy with hypoalbuminaemia, life-threatening intestinal bleeding of diffuse origin), and thrombotic events (protein C and S deficiency, low anti-thrombin III levels), whereas neurological development and cognitive capacity is usually normal. The clinical course is variable even within families. The disease is caused by loss of function of the gene *MPI* (15q24.1).

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

CDG syndrome type 1b

CDG1B

CDG-1b

Niedobór izomerazy fosfomannozy

Wrodzone zaburzenie glikozylacji typu 1b

Wrodzone zaburzenie glikozylacji typu 1b

Zespół CDG typu 1b

Zespół obniżonej glikozylacji glikoprotein typu 1b

CDG-1b

CDG1B

Carbohydrate deficient glycoprotein syndrome type 1b

Congenital disorder of glycosylation type 1b

Congenital disorder of glycosylation type 1b

Phosphomannose isomerase deficiency

#### Kod ORPHA

79319

#### Kod OMIM

602579

#### Kod ICD10

E77.8

#### Kod ICD11

5C54.0

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