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-thrombine 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 Ib
CDG1B
CDG-Ib
Niedobór izomerazy fosfomannozy
Wrodzone zaburzenie glikozylacji typu 1b
Wrodzone zaburzenie glikozylacji typu Ib
Zespół CDG typu Ib
Zespół obniżonej glikozylacji glikoprotein typu Ib
CDG-Ib
CDG1B
Carbohydrate deficient glycoprotein syndrome type Ib
Congenital disorder of glycosylation type 1b
Congenital disorder of glycosylation type Ib
Phosphomannose isomerase deficiency

Kod ORPHA
79319

Kod OMIM
602579

Kod ICD10
E77.8

Kod ICD11
5C54.0

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*Źródło

orphanet