Opis choroby *

Definicja

PMM2-CDG is the most frequent form of congenital disorder of N-glycosylation and is characterized by cerebellar dysfunction, abnormal fat distribution, inverted nipples, strabismus and hypotonia. 3 forms of PMM2-CDG can be distinguished: the infantile multisystem type, late-infantile and childhood ataxia-intellectual disability type (3-10 yrs old), and the adult stable disability type. Infants usually develop ataxia, psychomotor delay and extraneurological manifestations including failure to thrive, enteropathy, hepatic dysfunction, coagulation abnormalities and cardiac and renal involvement. The phenotype is however highly variable and ranges from infants who die in the first year of life to mildly involved adults.

Dane

Klasyfikacja
Choroba

Synonimy
CDG syndrome type Ia
CDG1A
CDG-Ia
Niedobór fosfomannomutazy 2
Wrodzone zaburzenie glikozylacji typu 1a
Wrodzone zaburzenie glikozylacji typu Ia
Zespół CDG typu Ia
Zespół obniżonej glikozylacji glikoprotein typu Ia
CDG-Ia
CDG1A
Carbohydrate deficient glycoprotein syndrome type Ia
Congenital disorder of glycosylation type 1a
Congenital disorder of glycosylation type Ia
Phosphomannomutase 2 deficiency

Kod ORPHA
79318

Kod OMIM
212065

Kod ICD10
E77.8

Kod ICD11
5C54.0

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*Źródło

orphanet