

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	Synonimy
Choroba	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

*Źródło

orphanet