

Opis choroby *

Definicja

A form of congenital disorders of N-linked glycosylation characterized by severe neurological involvement, including hypotonia, developmental delay, intellectual disability, postnatal microcephaly, and progressive brain and cerebellar atrophy. Epilepsy with hypsarrythmia is frequently reported. Additional features that may be observed include failure to thrive, arthrogryposis multiplex congenita (AMC), vision impairment (optic atrophy, iris coloboma) and facial dysmorphism (hypertelorism with a broad nasal bridge, large and thick ears, thin lips, micrognathia). The disease is caused by loss of function mutations of the gene *ALG3* (3q27.3).

Dane

Klasyfikacja

Choroba

Synonimy

CDG syndrome type 1d

CDG1D

CDG-1d

Niedobór mannozylotransferazy 6

Wrodzone zaburzenie glikozylacji typu 1d

Wrodzone zaburzenie glikozylacji typu 1d

Zespół CDG typu 1d

Zespół obniżonej glikozylacji glikoprotein typu 1d

CDG-1d

CDG1D

Carbohydrate deficient glycoprotein syndrome type 1d

Congenital disorder of glycosylation type 1d

Congenital disorder of glycosylation type 1d

Mannosyltransferase 6 deficiency

Kod ORPHA

79321

Kod OMIM

601110

Kod ICD10

E77.8

Kod ICD11

5C54.0

[*Źródło](#)

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