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 Id
CDG1D
CDG-Id
Niedobór mannozylotransferazy 6
Wrodzone zaburzenie glikozylacji typu 1d
Wrodzone zaburzenie glikozylacji typu Id
Zespół CDG typu Id
Zespół obniżonej glikozylacji glikoprotein typu Id
CDG-Id
CDG1D
Carbohydrate deficient glycoprotein syndrome type Id
Congenital disorder of glycosylation type 1d
Congenital disorder of glycosylation type Id
Mannosyltransferase 6 deficiency

Kod ORPHA
79321

Kod OMIM
601110

Kod ICD10
E77.8

Kod ICD11
5C54.0

---

*Źródło

orphanet