

Opis choroby *

Definicja

A form of congenital disorders of N-linked glycosylation characterized by progressive microcephaly, hypotonia, developmental delay, drug-resistant infantile epilepsy, and hepatomegaly. Additional features that may be observed include failure to thrive, pericardial effusion, renal cysts, skeletal dysplasia, facial dysmorphism (frontal bossing, hypertelorism, depressed nasal bridge, low-seated ears, large mouth) and hydrops fetalis. The disease is caused by loss-of-function mutations in the gene *ALG9* (11q23).

Dane

Klasyfikacja

Choroba

Synonimy

CDG syndrome type 1L

CDG1L

CDG-1L

Zespół CDG typu 1L

Niedobór mannozylotransferazy 7-9

Wrodzone zaburzenie glikozylacji typu 1L

Zespół obniżonej glikozylacji glikoprotein typu 1L

CDG-1L

CDG1L

Carbohydrate deficient glycoprotein syndrome type 1L

Congenital disorder of glycosylation type 1L

Mannosyltransferase 7-9 deficiency

Kod ORPHA

79328

Kod OMIM

263210

Kod ICD10

E77.8

Kod ICD11

5C54.0

[*Źródło](#)

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