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 IL
CDG1L
CDG-IL
Zespół CDG typu IL
Niedobór mannozylotransferazy 7-9
Wrodzone zaburzenie glikozylacji typu 1L
Zespół obniżonej glikozylacji glikoprotein typu 1L
CDG-IL
CDG1L
Carbohydrate deficient glycoprotein syndrome type IL
Congenital disorder of glycosylation type 1L
Mannosyltransferase 7-9 deficiency

Kod ORPHA
79328

Kod OMIM
263210

Kod ICD10
E77.8

Kod ICD11
5C54.0

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*Źródło

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