

Opis choroby *

Definicja

A form of congenital disorders of N-linked glycosylation that is characterized by gastrointestinal symptoms (diarrhea, vomiting, feeding problems with failure to thrive, protein-losing enteropathy), edema and ascites (including hydrops fetalis), hepatomegaly, renal tubulopathy, coagulation anomalies due to thrombocytopenia, brain involvement (psychomotor delay, seizures, ataxia), facial dysmorphism (low-set ears and retrognathia), pes equinovarus, and muscular hypotonia. Cataracts may also be observed. Prognosis is usually poor. The disease is caused by loss-of-function mutations in the gene *ALG8* (11q14.1), resulting in a block in the initial step of protein glycosylation.

Dane

Klasyfikacja

Choroba

Synonimy

CDG syndrome type 1h

CDG1H

CDG-1h

Niedobór glukozylotransferazy 2

Wrodzone zaburzenie glikozylacji typu 1h

Zespół CDG typu 1k

Zespół obniżonej glikozylacji glikoprotein typu 1h

CDG-1h

CDG1H

Carbohydrate deficient glycoprotein syndrome

type 1h

Congenital disorder of glycosylation type 1h

Congenital disorder of glycosylation type 1h

Glucosyltransferase 2 deficiency

Kod ORPHA

79325

Kod OMIM

608104

Kod ICD10

E77.8

Kod ICD11

5C54.0

[*Źródło](#)

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