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 Ih
CDG1H
CDG-Ih
Niedobór glukozylotransferazy 2
Wrodzone zaburzenie glikozylacji typu 1h
Zespół CDG typu Ik
Zespół obniżonej glikozylacji glikoprotein typu 1h
CDG-Ih
CDG1H
Carbohydrate deficient glycoprotein syndrome type Ih
Congenital disorder of glycosylation type 1h
Congenital disorder of glycosylation type Ih
Glucosyltransferase 2 deficiency

Kod ORPHA
79325

Kod OMIM
608104

Kod ICD10
E77.8

Kod ICD11
5C54.0

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*Źródło

orphanet