

Opis choroby *

Definicja

A form of congenital disorders of N-linked glycosylation characterized by facial dysmorphism (prominent forehead, large ears, thin upper lip), generalized hypotonia, feeding difficulties, moderate to severe developmental delay, progressive microcephaly, frequent upper respiratory tract infections due to impaired immunity with decreased immunoglobulin levels, and decreased coagulation factors. Additional features include hypogonadism with or without hypospadias in males, skeletal anomalies, seizures and cardiac anomalies in some cases. The disease is caused by loss of function mutations of the gene *ALG12* (22q13.33).

Dane

Klasyfikacja

Choroba

Synonimy

CDG syndrome type Ig

CDG1G

CDG-Ig

Niedobór mannozylotransferazy 8

Wrodzone zaburzenie glikozylacji typu Ig

Zespół CDG typu Ig

Zespół obniżonej glikozylacji glikoprotein typu Ig

CDG-Ig

CDG1G

Carbohydrate deficient glycoprotein syndrome type Ig

Congenital disorder of glycosylation type 1g

Congenital disorder of glycosylation type Ig

Mannosyltransferase 8 deficiency

Kod ORPHA

79324

Kod OMIM

607143

Kod ICD10

E77.8

Kod ICD11

5C54.0

[*Źródło](#)

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