## 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

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*Źródło

orphanet