## Opis choroby *

Definicja

RFT1-CDG is a form of congenital disorders of N-linked glycosylation characterized by poorly coordinated suck resulting in difficulty feeding and failure to thrive; myoclonic jerks with hypotonia and brisk reflexes progressing to a seizure disorder; roving eyes; developmental delay; poor to absent visual contact; and sensorineural hearing loss. Additional features that may be observed include coagulation factor abnormalities, inverted nipples and microcephaly. The disease is caused by mutations in the gene RFT1 (3p21.1).

Dane

**Klasyfikacja**
Choroba

Synonimy
CDG syndrome type In
CDG1N
CDG-In
Niedobór flipazy Man5GlcNAc2-PP-Dol
Wrodzone zaburzenie glikozylacji typu 1n
Wrodzone zaburzenie glikozylacji typu In
Zespół CDG typu In
Zespół obniżonej glikozylacji glikoprotein typu In
CDG-In
CDG1N
Carbohydrate deficient glycoprotein syndrome type In
Congenital disorder of glycosylation type 1n
Congenital disorder of glycosylation type In
Man5GlcNAc2-PP-Dol flippase deficiency

**Kod ORPHA**
244310

**Kod OMIM**
612015

**Kod ICD10**
E77.8

**Kod ICD11**
5C54.0

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*Źródło

orphanet