

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 1n

CDG1N

CDG-1n

Niedobór flipazy Man5GlcNAc2-PP-Dol

Wrodzone zaburzenie glikozylacji typu 1n

Wrodzone zaburzenie glikozylacji typu 1n

Zespół CDG typu 1n

Zespół obniżonej glikozylacji glikoprotein typu 1n

CDG-1n

CDG1N

Carbohydrate deficient glycoprotein syndrome

type 1n

Congenital disorder of glycosylation type 1n

Congenital disorder of glycosylation type 1n

Man5GlcNAc2-PP-Dol flippase deficiency

Kod ORPHA

244310

Kod OMIM

612015

Kod ICD10

E77.8

Kod ICD11

5C54.0

[*Źródło](#)

orphonet