DISEASE:
RFT1-CDG

NAME:	RFT1-CDG
DESCRIPTION:	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).
ORPHACODE:	244310
SYNONYMS:	CDG syndrome type 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
XREF(S):	Orphanet ICD-10 OMIM

ANALYTE(S):	RFT1
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