

DISEASE:
SLC35A1-CDG

NAME:	SLC35A1-CDG
DESCRIPTION:	SLC35A1-CDG is an extremely rare form of CDG syndrome (see this term) characterized clinically in the single reported case by repeated hemorrhagic incidents, including severe pulmonary hemorrhage.
ORPHACODE:	238459
SYNOMYS:	CDG syndrome type IIf CDG-IIf CDG2F CMP-sialic acid transporter deficiency Carbohydrate deficient glycoprotein syndrome type IIf Congenital disorder of glycosylation type 2f Congenital disorder of glycosylation type IIf
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SLC35A1
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