DISEASE:
DDOST-CDG

NAME:	DDOST-CDG
DESCRIPTION:	DDOST-CDG is a form of congenital disorders of N-linked glycosylation characterized by failure to thrive, developmental delay, hypotonia, strabismus and hepatic dysfunction. The disease is caused by mutations in the gene DDOST (1p36.1).
ORPHACODE:	300536
SYNONYMS:	CDG syndrome type Ir CDG-Ir CDG1R Carbohydrate deficient glycoprotein syndrome type Ir Congenital disorder of glycosylation type 1r Congenital disorder of glycosylation type Ir
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	DDOST
CREATED:	13 May 2019 - 01:02
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