DISEASE:
COG8-CDG

NAME:	COG8-CDG
DESCRIPTION:	The CDG (Congenital Disorders of Glycosylation) syndromes are a group of autosomal recessive disorders affecting glycoprotein synthesis. CDG syndrome type IIh is characterised by severe psychomotor retardation, failure to thrive and intolerance to wheat and dairy products.
ORPHACODE:	95428
SYNONYMS:	CDG syndrome type IIh CDG-IIh CDG2H Carbohydrate deficient glycoprotein syndrome type IIh Congenital disorder of glycosylation type 2h Congenital disorder of glycosylation type IIh
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	COG8
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