DISEASE:
ALG13-CDG

NAME:	ALG13-CDG
DESCRIPTION:	A form of congenital disorders of N-linked glycosylation characterized by microcephaly, hepatomegaly, edema of the extremities, intractable seizures, recurrent infections and increased bleeding tendency. The disease is caused by mutations in the gene ALG13 (Xq23).
ORPHACODE:	324422
SYNONYMS:	CDG syndrome type Is CDG-Is CDG1S Congenital disorder of glycosylation type 1s Congenital disorder of glycosylation type Is
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ALG13
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