DISEASE:
ALG6-CDG

NAME:	ALG6-CDG
DESCRIPTION:	A form of congenital disorders of N-linked glycosylation characterized by feeding problems, mild-to-moderate neurologic involvement with hypotonia, poor head control, developmental delay, ataxia, strabismus, and seizures, ranging from febrile convulsions to epilepsy. Retinal degeneration has also been reported. A minority of patients show other manifestations, particularly intestinal (such as protein-losing enteropathy) and liver involvement. The disease is caused by loss of function mutations of the gene ALG6 (1p31.3).
ORPHACODE:	79320
SYNONYMS:	CDG syndrome type Ic CDG-Ic CDG1C Carbohydrate deficient glycoprotein syndrome type Ic Congenital disorder of glycosylation type 1c Congenital disorder of glycosylation type Ic Glucosyltransferase 1 deficiency
XREF(S):	Orphanet OMIM ICD-10

ANALYTE(S):	ALG6
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