DISEASE:
ALG12-CDG

NAME:	ALG12-CDG
DESCRIPTION:	A form of congenital disorders of N-linked glycosylation characterized by facial dysmorphism (prominent forehead, large ears, thin upper lip), generalized hypotonia, feeding difficulties, moderate to severe developmental delay, progressive microcephaly, frequent upper respiratory tract infections due to impaired immunity with decreased immunoglobulin levels, and decreased coagulation factors. Additional features include hypogonadism with or without hypospadias in males, skeletal anomalies, seizures and cardiac anomalies in some cases. The disease is caused by loss of function mutations of the gene ALG12 (22q13.33).
ORPHACODE:	79324
SYNONYMS:	CDG syndrome type Ig CDG-Ig CDG1G Carbohydrate deficient glycoprotein syndrome type Ig Congenital disorder of glycosylation type 1g Congenital disorder of glycosylation type Ig Mannosyltransferase 8 deficiency

XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	ALG12
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