DISEASE:
ALG11-CDG

NAME:	ALG11-CDG
DESCRIPTION:	A form of congenital disorders of N-linked glycosylation characterized by facial dysmorphism (microcephaly, high forehead, low posterior hairline, strabismus), hypotonia, failure to thrive, intractable seizures, developmental delay, persistent vomiting and gastric bleeding. Additional features that may be observed include fat pads anomalies, inverted nipples, and body temperature oscillation. The disease is caused by mutations in the gene ALG11 (13q14.3).
ORPHACODE:	280071
SYNONYMS:	CDG syndrome type Ip CDG-Ip CDG1P Carbohydrate deficient glycoprotein syndrome type Ip Congenital disorder of glycosylation type 1p Congenital disorder of glycosylation type Ip
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ALG11
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