DISEASE:
ALG3-CDG

NAME:	ALG3-CDG
DESCRIPTION:	A form of congenital disorders of N-linked glycosylation characterized by severe neurological involvement, including hypotonia, developmental delay, intellectual disability, postnatal microcephaly, and progressive brain and cerebellar atrophy. Epilepsy with hypsarrythmia is frequently reported. Additional features that may be observed include failure to thrive, arthrogryposis multiplex congenita (AMC), vision impairment (optic atrophy, iris coloboma) and facial dysmorphism (hypertelorism with a broad nasal bridge, large and thick ears, thin lips, micrognathia). The disease is caused by loss of function mutations of the gene ALG3 (3q27.3).
ORPHACODE:	79321
SYNONYMS:	CDG syndrome type Id CDG-Id CDG1D Carbohydrate deficient glycoprotein syndrome type Id Congenital disorder of glycosylation type 1d Congenital disorder of glycosylation type Id Mannosyltransferase 6 deficiency

XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	ALG3
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