DISEASE:
COG7-CDG

NAME:	COG7-CDG
DESCRIPTION:	COG7-CDG is a congenital disorder of glycosylation characterised by dysmorphism, skeletal dysplasia, hypotonia, hepatosplenomegaly, jaundice, cardiac insufficiency, recurrent infections and epilepsy. To date, it has been described in two infants, both of whom died within the first three months of life. The syndrome is caused by a mutation in the gene encoding COG-7 (chromosome 16), a subunit of the oligomeric Golgi complex.
ORPHACODE:	79333
SYNONYMS:	CDG syndrome type IIe CDG-IIe CDG2E Carbohydrate deficient glycoprotein syndrome type IIe Congenital disorder of glycosylation type 2e Congenital disorder of glycosylation type IIe
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	COG7
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/3283

RELATED CONTENT

Related Genetic Tests

- Congenital disorders of glycosylation (79 genes)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- component of oligomeric golgi complex 7

Related Gene Panels

- Congenital disorders of glycosylation (79 genes) - KUL