DISEASE:
COG6-CGD

NAME:	COG6-CGD
DESCRIPTION:	A rare congenital disorder of glycosylation characterized by neonatal onset of global developmental delay, hypotonia, failure to thrive, hematological/immunological abnormalities, recurrent infections, liver involvement (with hepatosplenomegaly, cholestasis, fibrosis, or cirrhosis), and enteropathy. Additional reported manifestations include dysmorphic craniofacial features (such as microcephaly, broad palpebral fissures, and retrognathia), hypohidrosis, hyperkeratosis, and cardiac and musculoskeletal anomalies. Brain imaging may show hypoplastic corpus callosum, cerebral and cerebellar atrophy, and enlarged ventricles.
ORPHACODE:	464443
SYNONYMS:	CDG syndrome type IIL CDG-IIL CDG2L Congenital disorder of glycosylation type 2l Congenital disorder of glycosylation type IIL
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	COG6
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/2155

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