COG7-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. COG7-CDG. ORPHA:79333

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.