DISEASE:
DPM3-CDG

NAME:	DPM3-CDG
DESCRIPTION:	DPM3-CDG is an extremely rare form of CDG syndrome (see this term) characterized clinically in the single reported case by muscle weakness, waddling gait and dilated cardiomyopathy (see this term).
ORPHACODE:	263494
SYNONYMS:	CDG syndrome type Io CDG-Io CDG1O Carbohydrate deficient glycoprotein syndrome type Io Congenital disorder of glycosylation type 1o Congenital disorder of glycosylation type Io
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	DPM3
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/1100

RELATED CONTENT

Related Genetic Tests

- Congenital disorders of glycosylation (79 genes)
- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- dolichyl-phosphate mannosyltransferase subunit 3, regulatory

Related Gene Panels

- Congenital disorders of glycosylation (79 genes) - KUL
- Neuromuscular disorders (166 genes) - VUB