DISEASE:
PMM2-CDG

NAME:	PMM2-CDG
DESCRIPTION:	A rare congenital disorder of N-glycosylation and is characterized by cerebellar dysfunction, abnormal fat distribution, inverted nipples, strabismus and hypotonia. 3 forms of PMM2-CDG can be distinguished: the infantile multisystem type, late-infantile and childhood ataxia-intellectual disability type (3-10 yrs old), and the adult stable disability type. Infants usually develop ataxia, psychomotor delay and extraneurological manifestations including failure to thrive, enteropathy, hepatic dysfunction, coagulation abnormalities and cardiac and renal involvement. The phenotype is however highly variable and ranges from infants who die in the first year of life to mildly involved adults.
ORPHACODE:	79318
SYNONYMS:	CDG syndrome type Ia CDG-Ia CDG1A Carbohydrate deficient glycoprotein syndrome type Ia Congenital disorder of glycosylation type 1a Congenital disorder of glycosylation type Ia Phosphomannomutase 2 deficiency

XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	PMM2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- Congenital disorder of glycosylation (3 genes)
- Congenital disorders of glycosylation (79 genes)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- phosphomannomutase 2

Related Gene Panels

- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG
- Congenital disorder of glycosylation (3 genes) - KUL
- Congenital disorders of glycosylation (79 genes) - KUL