DISEASE:
MPI-CDG

NAME:	MPI-CDG
DESCRIPTION:	MPI-CDG is a form of congenital disorders of N-linked glycosylation, characterized by cyclic vomiting, profound hypoglycemia, failure to thrive, liver fibrosis, gastrointestinal complications (protein-losing enteropathy with hypoalbuminaemia, life-threatening intestinal bleeding of diffuse origin), and thrombotic events (protein C and S deficiency, low anti-thrombine III levels), whereas neurological development and cognitive capacity is usually normal. The clinical course is variable even within families. The disease is caused by loss of function of the gene MPI (15q24.1).
ORPHACODE:	79319
SYNONYMS:	CDG syndrome type Ib CDG-Ib CDG1B Carbohydrate deficient glycoprotein syndrome type Ib Congenital disorder of glycosylation type 1b Congenital disorder of glycosylation type Ib Phosphomannose isomerase deficiency
XREF(S):	Orphanet OMIM ICD-10

ANALYTE(S):	MPI
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/3296

RELATED CONTENT

Related Genetic Tests

- Congenital disorder of glycosylation (3 genes)
- Congenital disorders of glycosylation (79 genes)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- mannose phosphate isomerase

Related Gene Panels

- Congenital disorder of glycosylation (3 genes) - KUL
- Congenital disorders of glycosylation (79 genes) - KUL