

**Hyperinsulinemic** hypoglycaemia

Hypoglycaemia 0



**Gastrointestinal involvement** due to protein-losing enteropathy

Chronic diarrhoea 1 Recurrent vomiting Signs of malnutrition Growth retardation



Liver damage

Hepatomegaly 0

relatively firm

Liver fibrosis @leading to: Portal hypertension (PHT): **Splenomegaly** 

Complications of PHT:

- Digestive haemorrhage (oesophageal varices)
- Protein-losing enteropathy
- Intrapulmonary shunt
- Pulmonary arterial hypertension



Haematological diseases and haemostasis disorders

Arterial and/or venous thromboses (irrespective of area)

Bleeding disorders:

More rarely, frequently digestive, related to portal hypertension

**Other** 

Hypothyroidism (rare)

No generalised neurological damage

Hypoalbuminaemia Intestinal villous atrophy

+/- Steatorrhoea

Moderate

AST, ALT and GGT 0

High liver elastometry 2

Liver fibrosis, monitor liver nodules

Screen for complications of portal hypertension

▲ Antithrombin III +FXI •

> Proteins C and S

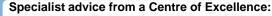
- Early signs (birth to 2 years) old)
- Late signs
- Specialist medical opinion and reference laboratory

Protein glycosylation defect type MPI-CDG (=CDG lb)?

**Specialist workup in collaboration with Centre of Excellence** at the same time as looking for other potential differential diagnoses

Plasma transferrin glycosylation study on serum (dry tube)

If abnormal<sup>1</sup>: Confirmatory genetic study +/- measure enzyme activity



G2M network: Rare Disease Centre of Reference / Competence : https://www.filiere-g2m.fr/annuaire/

+/- other network(s) depending on clinical symptoms: MHEMO / Firendo / Filfoie

Initial assessment and specialist treatment coordinated by the Centre of Excellence, specific treatment to be rapidly implemented Genetic counselling, family screening in a specialist centre

## For more information:

PNDS: French National Authority for Health -- MPI-CDG carbohydratedeficient glycoprotein syndrome caused by phosphomannose isomerase deficiency (has-sante.fr)

Emergency protocols for each symptom and/or disease; https://www.filiere-g2m.fr/urgences







