



Hyperinsulinemic hypoglycaemia

Hypoglycaemia ¹



Gastrointestinal involvement due to protein-losing enteropathy

Abdominal pain ¹
Chronic diarrhoea ¹
Recurrent vomiting ¹
Signs of malnutrition
Growth retardation



Liver damage

Relatively firm ¹
hepatomegaly
Liver fibrosis ² 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

Additional tests

Hypoalbuminaemia
Intestinal villous atrophy
+/- Steatorrhoea

➤ Moderate
AST, ALT and GGT ¹
High liver elastometry ²
Liver fibrosis, monitor liver nodules
Screen for complications of portal hypertension

➤ Antithrombin III +FXI ¹
➤ Proteins C and S

- ¹ Early signs (birth to 2 years old)
- ² Late signs
- ★ Specialist medical opinion and reference laboratory

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

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 abnormal¹: Confirmatory genetic study +/- measure enzyme activity

Specialist advice from a Centre of Excellence:

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

<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 carbohydrate-deficient glycoprotein syndrome caused by phosphomannose isomerase deficiency \(has-sante.fr\)](#)

[Emergency protocols](#) for each symptom and/or disease;
<https://www.filiere-g2m.fr/urgences>