Homocystinuria (CBS deficiency)

Label

In case of surgery / anaesthesia

= Risk of thrombosis

1 PATHOPHYSIOLOGY

<u>This pathology DOES NOT present a risk of coma nor acute metabolic decompensation.</u>

It is a deficiency in the CBS enzyme that converts homocysteine to cystathionine. This pathology induces psychomotor retardation, collagen anomalies mimicking Marfan syndrome (hyperlaxity, excess height, eye lens dislocation, etc.) and **hyperhomocysteinaemia, which is a risk factor for vascular thrombosis (either venous or arterial).**

Depending on the patient, this disease requires:

- a low-protein diet with controlled intake of methionine in order to limit hyperhomocysteinaemia: strict diet + amino acid substitutes. This type of diet totally excludes meat, fish and eggs, and any other protein-rich foods.
- medicinal treatments: Cystadane®, folic acid, +/- aspirin at platelet aggregation inhibitor dose, +/- anticoagulant.
- in B6-responsive patients: Treatment by B6 (pyridoxine) only.

2 IN CASE OF HOSPITALISATION OR INTERCURRENT PATHOLOGY

Provide treatment for the pathology that caused the patient to be admitted to A&E or hospital, as for all other patients, with no need for metabolic expertise.

It is **essential to continue the treatment and the diet (following the patient's usual treatment).**

Should treatment be stopped: Risk of increase in homocysteine and **risk of thrombosis**

<u>ASSISTANCE WITH DIET:</u>

If exceptionally a feeding bottle / meal is missed during a hospital stay: give an emergency, protein-free meal (low-protein pasta, low-protein bread with butter and jam) provided by the family, or, if by bottle: 65 g malto-dextrin + 20 ml oil + 350 ml measured water (choose the volume you want to give); or PFD1® / Energivit® / Duocal ®: 1 measuring spoon per 30 mL of water (0.7 Kcal/ml)

3 DRUG CONTRAINDICATIONS / GENERAL ADVICE:

- Exercise caution with repeated use of MEOPA (interaction with the B12 metabolism with risk of increase in homocysteine)
- Contraceptives containing oestrogen should be avoided due to increased risk of thrombosis in vitamin B6 non-responsive patients.

- All vaccinations are recommended.
- Anaesthesia: see overleaf

This emergency protocol is proposed by the G2M network working group. The protocol may be modified under the responsibility of the referring doctor. In no circumstances does it replace the responsibility of the doctor treating the patient in A&E.

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4 PROCEDURE WHEN PREPARING FOR ANAESTHESIA

A. Planned anaesthesia: Preventing the risk of thrombosis

- Inform the referring metabolic doctor and biochemists of the date of surgery.
- Optimise metabolic control: emergency diet (no proteins, with amino acid mixture without methionine), 7 to 10 days before surgery (duration and method to be discussed depending on the metabolic balance: orally, CEF, IV), with **total homocysteine control a few days before the operation (Objective <50 µM)**.
- Discuss prophylactic anticoagulation, depending on the risk of haemorrhage and metabolic balance.
- Work-up to be performed during the anaesthesia consultation and the day before surgery:
  - Plasma amino acid chromatography (heparin tube)
  - Total plasma homocysteine (heparin tube or EDTA, check with the biochemistry laboratory)
- The day before surgery:
  - Despite induced fasting, CONTINUE THE AMINO ACID MIXTURE ORALLY for as long as possible.
  - Infusion using **10% serum glucose** with standard electrolyte additions* (not pure 10% glucose) with 2 to 2.5 L//m²/d (hyperhydration to limit the risk of thrombosis)

***e.g.:** Polyionic, Bionolyte, B45, Glucidion, etc. if no solutes available, 10% glucose + 4g/L of NaCl (70 meq/L) and 2g/L of KCl (27 meq/L)

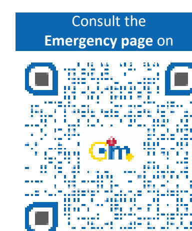
B. Emergency procedures

- Emergency surgery should not be delayed.
- Apply the infusion and instructions below from the start of treatment.
- Measure total homocysteine without waiting for the result.
- Plan for prophylactic anticoagulation whatever the age of the patient.

C. Outpatient surgery: not recommended (depending on the metabolic balance)

D. Local and local-regional anaesthesia: possible. Avoid MEOPA.

Consult the **Emergency page** on



REFERENCE DOCTORS AND CONTACT DETAILS

On-call telephone numbers for metabolic emergencies of:

At night, only the medical teams can call in emergency situations and only if the emergency certificate has not been understood or if the clinical state or test results are worrying. As far as possible make calls before night time.

Secretarial issues must be dealt with via the medical secretariat during the week or by email addressed to the patient's referring metabolic doctor.

Certificate issued on Dr