

Cobalamin C deficiency (cb1C)

Label

No risk of decompensation if treatments are taken compliantly.

1 PATHOPHYSIOLOGY

Patients with this disease are generally not at risk of coma or acute metabolic decompensation when receiving treatment.

cb1C disease is due to a defect in remethylation, with **hyperhomocysteinaemia** and urine excretion of methylmalonic acid (MMA). The disease can present with: deteriorating neurological function with impaired consciousness, visual deficits (macular disease), myelopathy, macrocytic anaemia +/- pancytopenia, and rarely metabolic acidosis. Due to hyperhomocysteinaemia, there is also a risk of thrombosis. Depending on the individual patient, this disease may require:

- Vitamin B12 in the form of hydroxocobalamin: by SC or IM injection multiple times per week.
Hydroxocobalamin: vials 2ml=10mg. IM or SC injections. IV injection possible (off-label), to dilute in 20 to 50 mL of normal saline.
- Oral treatments: Folinic acid, Cystadane, Levocarnil, Methionine
- No need for low protein diet.

2 IN CASE OF HOSPITALISATION / INTERCURRENT DISEASE

Manage the condition requiring emergency admission, or admission to hospital, as for any other patient; there is no need for expert metabolic advice.

- It is **essential to continue treatments**. If treatment is discontinued: Homocysteine levels may increase, with a **risk of thrombosis**.
- In situations where there is a **risk of dehydration**: increased risk of vascular thrombosis. Do not hesitate to start a glucose/electrolyte infusion (for example with bionolyte G5% or G10%): flow rate and electrolyte content to be adjusted according to the clinical situation.
- In **situations where there is a risk of thrombosis** (in plaster, prolonged bed rest, post-operatively): consider prophylactic anticoagulants.
- Exceptionally, where there is infection or severe dehydration, ketoacidosis +/- hypoglycaemia has been observed, with unexplained consciousness disorders, requiring appropriate management (infusion of bionolyte G10%, symptomatic treatment etc.).

3 DRUG CONTRAINDICATIONS / GENERAL ADVICE:



Contraindication: Entonox / Nitrous oxide (interaction with B12 metabolism with risk of increased homocysteine levels)

- All vaccinations are recommended

4 ACTION TO BE TAKEN BEFORE PLANNED GENERAL ANAESTHESIA

Preventing a risk of thrombosis:

1. Notify the specialist metabolic doctor and the biochemists about the date of surgery.
2. Discuss the use of prophylactic anticoagulation with the specialist team and a haemostasis specialist.
3. Tests to be carried out at the time of seeing the anaesthetist and the day before surgery (without waiting for the results):
 - Plasma amino acids (heparin tube)
 - Total plasma homocysteine (heparin or EDTA tube depending on the laboratory, see local requirements)
4. If homocysteine is elevated: Pre-operative glucose infusion, to increase hydration 2L/m² of body surface area/day.

NUMBERS AND MEDICAL SPECIALISTS

On-call telephone numbers for metabolic emergencies:

At night, only 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 nighttime.

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

Certificate issued on

Dr

Look up the emergency

