Phenylketonuria

No acute risk

Label

1 IF CONSULTING AT A&E

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.

2 PATHOPHYSIOLOGY

This pathology DOES NOT present a risk of coma or acute metabolic decompensation.

It is a deficiency of the PAH enzyme that degrades phenylalanine. The diagnosis is made during neonatal screening (or later in childhood in the absence of screening, with cognitive consequences).

Depending on the patient, this disease requires:

- a strict low-protein diet with controlled intake of phenylalanine: strict diet + amino acid substitutes. This type of diet totally excludes meat, fish and eggs, and any other protein-rich foods.
- medicinal treatments: Kuvan® (BH4) in responsive patients.

It is important to follow this diet, including in cases of hospitalisation, but there is no risk of acute decompensation in the case of an isolated error. The patient's family knows about this diet.

3 DRUG CONTRAINDICATIONS / GENERAL ADVICE:

- No treatment is contraindicated (the aspartame contained in some treatments can constitute an intake of phenylalanine but this remains negligible).
- All vaccinations are recommended.
- In case of surgery: no particular precautions to be taken. No contraindication to anaesthetics.

4 ASSISTANCE WITH DIET

In all cases, the patient or parents must provide meals for the hospital stay (amino acid mixes, weighed protein-controlled diet).

If exceptionally a feeding bottle / meal is missed during a hospital stay: give an emergency, protein-free meal

-for patients with a diversified diet: : low-protein pasta, low-protein bread with butter and jam) provided by the family,

-for an infant : by bottle: 65 g malto-dextrin + 20 ml oil + 350 ml measured water (choose the volume you want to give); or PFD1® (1 scoop for 30 mL water (0.7 Kcal/ml) / Energivit® 1 scoop for 30 mL water (0.7 Kcal/ml) / or infant amino acid mix: 1 scoop for 30 mL water (0.7 Kcal/ml): (choose the volume you wish to provide).

In the event of hospitalization, the diet can be adapted with the help of the referral center during working hours (no risk of coma). The family is the expert for this diet. The amino acid mix is provided by the patient.

Contraindication to "light" or "zero" products containing aspartame, source of phenylalanine.

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