GLYCOGEN STORAGE DISEASE type 0, or glycogen synthase deficiency
GLYCOGEN STORAGE DISEASE type VI, or phosphorylase deficiency
GLYCOGEN STORAGE DISEASE type IX, or phosphorylase kinase deficiency

Priority patient: must not be kept waiting in A&E

Risk of hypoglycaemia if prolonged fasting, vomiting or diarrhoea

1 EMERGENCY WORKUP

Capillary and venous blood glucose (parameter guiding management).

Possibly also capillary blood ketones or urinary dipstick and lactate for the record (will not alter treatment path)

Any other tests arising from the clinical situation or the reason for coming to A&E.

2 IF THERE IS HYPOGLYCAEMIA < 60 mg/dl (3.3 mmol/l)

- Replenish with glucose with 1ml/kg of G30% (max 30 mL) orally or enterally if conscious, or 3mL/kg of G10% IV if unconscious (G30%can also be done by IV). Check capillary blood glucose (dextro) 5 minutes later.
- If still hypoglycaemic, replenish with glucose for a second time and check capillary blood glucose 5 minutes later.
- Set up a venous line **URGENTLY** without delaying glucose replenishment.
- Start an infusion straight away even if hypoglycaemia has been corrected: Infusion based on **G10%** glucose with addition of standard electrolytes* (not pure G10)

Age	0-24 mois	2-4 ans	4-14 ans	>14 ans - adulte	DEBIT MAX
Débit de perfusion	6ml/kg/h (10mg/kg/min)	5ml/kg/h (8mg/kg/min)	3,5ml/kg/h (6mg/kg/min)	2,5ml/kg/h (4mg/kg/min)	**120ml/h (3L/24h)**

e.g. Polyionique, Bionolyte, B45, Glucidion etc. If no pre-made solutions available, use G10% + 4g/L NaCl(70meq/L) and 2g/L KCl(27meq/L)

If impossible to give infusion => Nasogastric or gastrostomy tube: prepare the IV solutions listed above and pass them through the tube at the same rate.

- **Glucagon CONTRAINDICATED**
- If there are no gastrointestinal disorders and if the preparation is available: instead of the infusion, give enteral nutrition via the nasogastric tube using a standard enteral feeding product suited to the patient's age.

3 IN A SITUATION WITH A RISK OF HYPOGLYCAEMIA

- Any situation in which the patient is deprived of glucose intake for a period, depending on the patient's tolerance and age, e.g. where there is **vomiting**, **refusal to feed, diarrhoea, fasting** (**examinations, operations** etc.).

=>Infusion via a peripheral line

- **These patients do not present with any other specific issues regarding their treatment or in terms of risk of complications.**

4 MONITORING AFTER BLOOD GLUCOSE LEVELS ARE CORRECTED, OR WHILE ON INFUSION

●Check capillary blood glucose 1 hr after starting the infusion, then every 3 hrs.

- Risk of non-serious hyperglycaemia in cases of glycogen storage disease type 0 while receiving glucose infusion
- Risk of non-serious hyperlactataemia in cases of glycogen storage disease type 0, VI or IX while on glucose infusion

●Adjust the rate of infusion of G10% + electrolytes by +/-5mL/hr. Target: blood glucose between 60 and 120 mg/dL.

Look up the **emergency information** on the G2M

5 PATHOPHYSIOLOGY

Patients with **glycogen storage disease type 0 (glycogen synthase deficiency)** are unable to synthesise glycogen.

They do not present hepatomegaly or any characteristic clinical aspects. Gluconeogenesis and fatty acid oxidation remain functional, which reduces the risk of hypoglycaemia during a short fasting period.

If fasting is prolonged (> 4-6 hours) or in an equivalent situation (diarrhoea, vomiting, anorexia), they run the risk of hypoglycaemia accompanied by ketosis. For the same pathophysiological reasons, they may experience post-prandial hyperglycaemia, which should not be confused with diabetes, and post-prandial hyperlactataemia.

Apart from periods of fasting or the equivalent, these patients do not present any particular risks, and are not at risk of metabolic decompensation or coma.

Patients with **glycogen storage disease type VI or IX** cannot break down glycogen correctly when fasting, but their capacity for gluconeogenesis and fatty acid oxidation remains intact.

They present with hepatomegaly during childhood, which becomes more noticeable in adolescence, but the liver remains hyperechoic in ultrasound. There are no other characteristic clinical features (some exceptional forms may show muscle involvement)

Where fasting is extended (> 4-6 hours) or in an equivalent situation (diarrhoea, vomiting, anorexia), they run a risk of hypoglycaemia accompanied by ketosis. This risk applies mainly to newborns and young children, and subsequently goes away. The underlying biochemical pathophysiology means that, following excess sugar intake or in the post-prandial period, they can experience a moderate elevation in blood lactate without acidosis or symptoms that are characteristic of the disease.

6 GENERAL ADVICE

No particular drug contraindications apart from glucagon
All vaccines are recommended.

- **Never exceed the patient's usual fasting time: if admitted to hospital for another reason, keep to the normal diet and give cornflour at bedtime (or depending on the patient, night-time continuous enteral nutrition).**
- **If the patient has to fast (for example before surgery), give an infusion as described on the front page.**

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