NATIONAL CENTRE FOR INHERITED METABOLIC DISORDERS

EMERGENCY LETTER FOR HOSPITAL – Glycogen Storage Disorder Type 1a/1b

Addressograph

AT RISK OF HYPOGLYCAEMIA
Please attend to this patient without delay
Always contact the NCIMD for further advice

DIAGNOSIS

Glycogen storage disorders (GSDs) are a group of metabolic disorders affecting either the synthesis or breakdown of glycogen in the body, which is crucial for blood glucose homeostasis. There are two forms of GSD Type 1: GSD 1a, which is caused by deficiency in glucose-6-phosphatase and GSD 1b, caused by deficiency in the glucose-6-phosphate transporter. Both types of GSD 1 cause severe fasting hypoglycaemia, elevated lactate and hepatomegaly. Patients with GSD 1b also have neutrophil dysfunction which can lead to severe infections.

Complications of GSD 1a and 1b include anaemia, hyperlipidaemia, renal complications, and increased bleeding risk. Specifically in GSD 1b, complications also include severe infection and inflammatory bowel disease. GSD 1 is managed with strict adherence to fasting times and a diet with regular intake of complex carbohydrate and low in simple sugars. They may also need a gastrostomy tube to provide feeds. Uncooked cornstarch or a modified cornstarch (Glycosade®) are often used to improve tolerance of fasting. Children with GSD 1b will also be taking G-CSF and/or empaglifozin for management of neutropaenia/neutrophil dysfunction.

Patients with GSD 1a and 1b are **extremely vulnerable to hypoglycaemia** with a very short period of fasting. Triggers may include reduced oral intake, intercurrent illness (especially if vomiting/diarrhoea), or severe exertion.

Maximum fasting time (well) - _____ **hours (overnight)**

Maximum fasting time (unwell) - _____ **hours (day and night)**

NOTES FOR PARENTS/CARERS

Contact the metabolic team at the first signs of illness to discuss your child's care. Metabolic Clinical Nurse Specialist contact: (01) 878 4409 (during working hours).

If you are advised to attend the emergency department, remember to bring your medication, feeding supplies, a copy of this letter and your feeding plan with you.

EMERGENCY MANAGEMENT

Always take illness in a patient with GSD Type 1 seriously. Always discuss this patient with the metabolic consultant on call without delay.

- Assess and resuscitate as necessary as per APLS protocols
- Check glucose at point of care immediately on presentation
- Obtain IV access and send the following bloods to the lab:
    - full blood count, glucose, lactate, VBG, renal/liver/bone profile, blood culture
    - additional tests as dictated by presentation (e.g. CRP, urine culture, NPA, etc)
- If blood glucose <3mmol/l and tolerating oral intake, consider giving a snack or the rescue prescribed in the patient's diet plan (typically 5-10g carbohydrate) and recheck glucose in 10 minutes. Repeat if still <3mmol/l. Follow with next due snack/meal/cornstarch
- If blood glucose <3mmol/l and not tolerating oral/enteral intake, consider a bolus of 10% dextrose 2ml/kg IV and recheck glucose in 10 minutes. Repeat if still <3mmol/l. Always follow a bolus of dextrose with IV maintenance fluids as described below
- If unable to obtain IV access, Glucogel or equivalent can be given to treat the hypoglycaemia
- Use of glucagon is contraindicated
- If not tolerating oral intake, commence IV fluids at 100% maintenance rate, using 0.9% NaCl + 10% dextrose + 2mmol/kg/day of potassium chloride (KCl can be added once you have confirmed urine has been passed), adjusted depending on glucose and electrolyte levels
- In consultation with the metabolic team, consider providing a small volume of glucose polymer solution (e.g. SOS) via NG or PEG alongside the IV fluid infusion to reduce risk of hypoglycaemia in the event of failure of the IV cannula – or encourage small amounts of oral intake if possible
- If the patient is tolerating oral intake, you must follow their prescribed unwell diet plan orally/NG/PEG, with particular attention to fasting intervals (often continuous feed)
- Monitor glucose at point of care 2 hourly and adjust glucose infusion rate (GIR) to maintain blood glucose between 4mmol/l and 6mmol/l
- Give ondansetron for management of nausea and vomiting
- If restarting feeds, do not stop IV fluids abruptly – rate can be weaned gradually once the child has demonstrated that they are tolerating oral intake in a satisfactory quantity
- If infection suspected, manage in line with the hospital sepsis pathway. Patients with GSD 1b are at higher risk of serious bacterial infection and may be neutropenic at presentation. Consider discussion with Micro/ID especially in cases of sepsis or if the patient has multi-drug resistant organisms. Co-amoxiclav should be avoided as clavulanic acid is not recommended in GSDs – amoxicillin is safe to use if appropriate
- **This plan is for the immediate management only and continued management must be discussed with the metabolic team**

The on call service for the National Centre for Inherited Metabolic Disorders is available to discuss this patient at +353 1 878 4200 via the CHI at Temple Street switchboard (available 24 hours a day). The metabolic ward nurse is also available to discuss at +353 1 878 4200, ask for bleep 836 (available 24 hours a day).