NATIONAL CENTRE FOR INHERITED METABOLIC DISORDERS

EMERGENCY LETTER FOR HOSPITAL – 3-Hydroxy-3-methylglutaryl-CoA (HMG-CoA) synthase deficiency

Addressograph

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

DIAGNOSIS

3-Hydroxy-3-methylglutaryl-CoA (HMG-CoA) synthase deficiency is an inherited disorder of ketone body synthesis. Ketones are an important source of energy for the brain, heart and kidneys during prolonged periods of fasting.

When well, patients with HMG-CoA synthase deficiency have no specific presenting features and have no need for a specialised diet. They are provided with an age-appropriate maximum fasting time by the metabolic team.

When unwell, patients are required to restrict their fasting times in order to avoid metabolic crisis. They have an emergency plan with age-specific carbohydrate drinks (e.g. SOS) and volumes for use during illness. Catabolism (e.g. intercurrent illness, vomiting, diarrhoea, prolonged fasting) can lead to serious illness, with encephalopathy and even sudden death. However, if the correct emergency management is followed, outcomes are excellent. Intravenous dextrose may be required if patients are vomiting, have diarrhoea or are unable to manage sufficient carbohydrate-containing foods or drinks. **Always take vomiting, diarrhoea or poor feeding seriously in a child with HMG-CoA synthase deficiency.**

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 a copy of your emergency regimen with you

Approved by NCIMD Consultant Group October 2023

EMERGENCY MANAGEMENT

- Assess and resuscitate as necessary as per APLS protocols
- Assess for evidence of encephalopathy (e.g. confusion, behavioural changes etc)
- Check glucose at point of care on presentation – however, **do not be falsely reassured by a normal blood glucose, as hypoglycaemia is a late sign of decompensation**
- Obtain IV access and send the following bloods to the lab:
    - glucose, VBG, renal/liver/bone profile
    - additional tests as dictated by presentation (e.g. CRP, urine culture, NPA, etc)
- If blood glucose is <3mmol/l give a bolus of 2ml/kg 10% dextrose and recheck glucose in 10-15 minutes
- If unable to obtain IV access, Glucogel or equivalent can be given to treat the hypoglycaemia
- Always commence IV maintenance fluids as described below once IV access has been established, until further assessment has been completed and the patient has been discussed with the metabolic team:
    - If the patient <u>is not tolerating</u> oral intake, provide IV fluids at 120% of maintenance rate, using 0.9% NaCl + 10% dextrose + 2mmol/kg/day of potassium chloride (KCl can be added once you have confirmed that urine has been passed), adjusted depending on electrolyte levels
    - If the patient is relatively <u>well</u>, has a normal blood glucose, and <u>is tolerating</u> oral intake, you may follow their prescribed emergency diet plan orally/via NG tube or ensure regular intake of complex carbohydrate foods (e.g. bread, cereal, pasta, rice, pancakes, crackers etc.), and discontinue the IV fluids following discussion with the metabolic team
- **Do not use Dioralyte/oral rehydration solution alone** as this contains insufficient glucose for requirements
- Give ondansetron for management of nausea and vomiting
- If patient is on carnitine supplementation, continue usual dose
- If restarting feeds, do not stop IV fluids abruptly – the rate can be weaned gradually once the child has demonstrated that they are tolerating oral intake in a satisfactory quantity

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).