NATIONAL CENTRE FOR INHERITED METABOLIC DISORDERS (NCIMD)

EMERGENCY LETTER FOR HOSPITAL – MCAD Deficiency

Addressograph

AT RISK OF ACUTE METABOLIC CRISIS
Please attend to this patient without delay

DIAGNOSIS

Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD) is an inherited disorder of fatty acid metabolism. The body is unable to correctly remove medium chain carbon molecules from fatty acids in the mitochondria, resulting in impaired energy production.

When well, patients with MCADD have no specific presenting features and have no need for a specialised diet other than avoidance of medium chain triglycerides and foods high in coconut. They are provided with an age-appropriate maximum fasting time by the metabolic team. Some patients also take a medication called carnitine.

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 foods or drinks. **Always take vomiting, diarrhoea or poor feeding seriously in a child with MCADD.**

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

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

NOTES FOR PARENTS/CARERS

Contact the metabolic team if your child becomes unwell 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 medications and feeding supplies, and a copy of this letter and your emergency diet plan with you.

Approved by NCIMD Consultant Group June 2023

EMERGENCY MANAGEMENT

- Assess and resuscitate as necessary as per APLS protocols
- Assess for evidence of encephalopathy
- Check glucose and ketones 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, ketones, VBG, CK, ammonia, 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 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 obtained, until further assessment has been completed and the patient has been discussed with the metabolic team
    - If the patient is not tolerating oral intake, continue IV fluids at 120% of maintenance rate, using 0.9% NaCl + 10% dextrose + 2mmol/kg/day of KCl chloride (KCl can be added once you have confirmed that urine has been passed), adjusted depending on electrolyte levels
    - If the patient is relatively well, has a normal blood glucose, and is tolerating oral intake, you may follow their prescribed SOS 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 in 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 regular carnitine supplement, continue their usual dose PO (or IV if not tolerating oral intake)
- 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).