NATIONAL CENTRE FOR INHERITED METABOLIC DISORDERS

EMERGENCY LETTER FOR HOSPITAL – Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCADD)

Addressograph

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

DIAGNOSIS

Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCADD) is an inherited disorder of fatty acid metabolism. The body is unable to complete the first step in beta-oxidation of long chain fatty acids in the mitochondria, resulting in impaired energy production.

Patients are managed with a diet low in long chain fat, with regular intake of carbohydrate and avoidance of prolonged fasting. Their diet is supplemented with medium chain triglycerides (MCT) and they may also be prescribed carnitine supplementation if indicated. Complications of VLCADD include hypoketotic hypoglycaemia, cardiomyopathy, arrhythmia and rhabdomyolysis.

When a patient with VLCADD is under metabolic stress e.g. they are unwell, fasting for a prolonged period of time, have surgery, or sustain a fracture or other trauma, they are at high risk of metabolic decompensation. They may present with **vomiting, myalgia, haematuria, weakness, shortness of breath, palpitations, decreased level of consciousness or lethargy**.

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 emergency diet plan with you.

Approved by NCIMD Consultant Group October 2023

EMERGENCY MANAGEMENT

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

- Stop fat exchanges (unless advised otherwise by metabolic team)
- Assess and resuscitate as necessary as per APLS protocols
- 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, lactate, VBG, renal/liver/bone profile, CK, acylcarnitine profile (dried bloodspot)
    - additional tests as dictated by presentation (e.g. CRP, urine culture, NPA, etc)
- Obtain a urine for dipstick (myoglobin will give a false positive for haemoglobin on the dipstick if there is rhabdomyolysis) +/- send urine for myoglobin to lab if available
- Consider performing an ECG and/or echocardiogram. Continuous cardiac monitoring should be commenced if there is rhabdomyolysis
- If blood glucose is <3mmol/l give a bolus of 2ml/kg 10% dextrose and recheck glucose in 15 minutes
- Commence patient on their unwell diet (see emergency diet plan for volumes and recipes) via enteral route (PO/NG/PEG)
- If the patient is not tolerating oral intake, or if CK is very high, commence IV fluids at 120%-150% of maintenance rate, using 0.9% NaCl + 10% dextrose, adjusted depending on electrolyte levels. **Caution**: fluid overload in cardiomyopathy
- KCl may be added to IV fluids once you have confirmed that urine has been passed and plasma potassium result is known – patients with rhabdomyolysis can develop hyperkalaemia so careful monitoring is required
- Give ondansetron for management of nausea and vomiting
- If patient is on regular carnitine supplement, continue their usual dose PO (do not give IV)
- If infection suspected, manage in line with the hospital sepsis pathway as patients can decompensate quickly. Consider discussion with Micro/ID especially in cases of sepsis or if the patient has multi-drug resistant organisms
- **Do not give IV lipids** to patients with VLCAD deficiency
- **Avoid anaesthetic agents high in long chain fats** (e.g. propofol)
- **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).