NATIONAL CENTRE FOR INHERITED METABOLIC DISORDERS (NCIMD)

EMERGENCY LETTER FOR HOSPITAL – Isovaleric Acidaemia (IVA)

Addressograph

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

DIAGNOSIS

Isovaleric acidaemia (IVA) is an inherited disorder of protein metabolism. The body is unable to correctly metabolise the amino acid leucine, resulting in a build-up of toxic metabolites. People with IVA are managed using a special diet which is lower in natural protein and may be supplemented with a synthetic protein that is free of leucine. They also take carnitine and glycine, medicines that help to clear the toxic metabolites from the body.

When a patient with IVA is under metabolic stress e.g. they are unwell, fasting for a prolonged period of time, have excessive protein intake, have surgery, or sustain a fracture or other trauma, they are at high risk of metabolic decompensation. Constipation should be avoided. They may present with **vomiting, poor appetite, irritability, confusion, abnormal movements or reduced level of consciousness**. It is critical to react early to any signs of illness by implementing their unwell plan.

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 regimen with you.

EMERGENCY MANAGEMENT

Always take illness in a patient with IVA seriously. Always discuss this patient with the metabolic consultant on call <u>without delay</u>.

- **Stop natural protein** (unless advised otherwise by metabolic team)
- Assess and resuscitate as necessary as per APLS protocols
- Obtain IV access and send bloods:
    - glucose and ketones (point of care)
    - FBC, renal/liver/bone profile, venous blood gas, glucose, lactate, ammonia, amylase/lipase (if appropriate)
    - Other tests as indicated by presentation (blood cultures, urine culture, NPA etc)
- Start 10% dextrose at 120% maintenance rate with 0.9% saline and 2 mmol/kg/day of potassium chloride (KCl can be added once you have confirmed that urine has been passed), adjust depending on electrolyte levels
- Consider starting IV 20% Lipid (Intralipid or SMOF) 2-3 g/kg/day to prevent catabolism (in discussion with the metabolic team)
- If indicated, start the child's unwell synthetic protein (see emergency diet plan for volume and recipe) via enteral route (PO/NG/PEG)
- Provide double the patient's usual dose of glycine enterally either PO or via NG (can be given as NG continuous infusion if not tolerating boluses)
- Provide double the patient's usual dose of carnitine either enterally or intravenously if not tolerated enterally
- If child is nauseated or vomiting, give ondansetron
- Manage fever proactively with paracetamol or ibuprofen
- Assess for and manage constipation
- Keep **detailed and accurate** record of all diet and fluid intake
- 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
- In the event of hyperammonaemia, the metabolic consultant on call may advise further medications such as carglumic acid (Carbaglu® - available from pharmacy in CHI at Temple Street, if needed), sodium benzoate or sodium phenylbutyrate
- **This plan is for the immediate management only and continued management <u>must be discussed</u> 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).