NATIONAL CENTRE FOR INHERITED METABOLIC DISORDERS (NCIMD)

EMERGENCY LETTER FOR HOSPITAL – Maple Syrup Urine Disease (MSUD)

Addressograph

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

DIAGNOSIS

Maple Syrup Urine Disease (MSUD) is an inherited disorder of protein metabolism. The body is unable to correctly metabolise the branched chain amino acids (BCAAs) leucine, isoleucine and valine, resulting in build-up of these amino acids and their keto-acids. Leucine in particular is a neurotoxin. High levels of leucine in the brain lead to encephalopathy, cerebral oedema, and even death. People with MSUD are managed with a diet which is low in natural protein and supplemented with a BCAA-free synthetic protein. They also take supplements of isoleucine and valine, as these BCAAs can help to reduce leucine levels in several ways. Some patients may also take a thiamine supplement.

When a patient with MSUD 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. They may present with **irritability, lethargy, drowsiness, vomiting, confusion, ataxia (poor balance), slurred speech, or abnormal movements**. It is critical to react early to any signs of illness by using 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 medication and feeding supplies, and a copy of this letter and your emergency diet plan with you.

EMERGENCY MANAGEMENT

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

- **Stop natural protein** (unless advised otherwise by metabolic team)
- Assess and resuscitate as necessary as per Advanced Paediatric Life Support (APLS) protocols
- Obtain IV access and send bloods:
    - glucose and ketones (point of care)
    - FBC, renal/liver/bone profile, venous blood gas, serum and urine osmolality
    - **Branched chain amino acids (send urgently to the metabolic lab in Temple Street)**
    - 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), adjusted depending on electrolyte levels. NOTE: If a bolus has been given during resuscitation, subtract the bolus volume from the calculated 120% maintenance volume for 1st 24 hours due to risk of cerebral oedema
- Start IV 20% Lipid (Intralipid or SMOF) 2-3 g/kg/day to prevent catabolism
- Start the child's unwell synthetic protein (see emergency diet plan for volume and recipe) via enteral route (PO/NG/PEG) – **prioritise this over other oral intake**
- Aim for a plasma osmolality 285-295 mOsmol and sodium 140-145 mmol/L
- If child is nauseated or vomiting, give ondansetron
- **Manage fever proactively with paracetamol and ibuprofen**
- Strict monitoring/recording of fluid balance and assess regularly for fluid overload
- Keep **detailed and accurate** record of all diet and fluid intake
- High risk of cerebral oedema – monitor neurological status carefully (encephalopathy may be subtle, such as slight slurring of speech or subtle behavioural change and must be carefully assessed)
- 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
- Dosing of isoleucine and valine to be determined by metabolic consultant
- Patients who are very unwell +/- have very high leucine levels may require insulin infusion and/or transfer to PICU for specialist care and/or haemofiltration
- **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).