NATIONAL CENTRE FOR INHERITED METABOLIC DISORDERS

EMERGENCY LETTER FOR HOSPITAL – Infantile Liver Failure Syndrome Type 1 (ILFS1) – a.k.a. LARS deficiency

Addressograph

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

DIAGNOSIS

Infantile liver failure syndrome type 1 (ILFS1), also known as LARS deficiency, is an inherited disorder due to pathogenic variants in the *LARS1* gene. Patients with LARS deficiency present typically in the first year of life with failure to thrive, developmental delay, liver impairment, or encephalopathy. The disorder is characterised by episodes of liver dysfunction or liver failure and/or encephalopathy in the context of illness. **The risk is particularly high with episodes of fever.** Patients with LARS deficiency frequently have anaemia (resistant to iron supplementation), and may have seizures, particularly with illness. Some patients have recurrent hypoglycaemia.

Children with LARS deficiency may become disproportionately unwell with mild or minor illness, therefore any illness must be taken seriously and managed appropriately. Maintaining protein intake, particularly when unwell, may be helpful for this group of patients, especially with febrile illness. Early initiation of enteral or parenteral nutrition during this period is indicated. They should avoid prolonged periods of fasting and maintain adequate calorie intake.

NOTES FOR PARENTS/CARERS

Contact the metabolic team at the first signs of illness to discuss your child's care. Do not delay giving paracetamol if your child has a fever. 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 diet plan with you.

EMERGENCY MANAGEMENT

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

- Assess and resuscitate as necessary as per APLS protocols
- Check glucose and ketones at point of care on presentation
- **Manage fever proactively and aggressively with regular paracetamol (ibuprofen second line)**
- Obtain IV access and send the following bloods to the lab:
    - full blood count, glucose, lactate, renal/liver/bone profile, venous blood gas, coagulation profile
    - additional tests as dictated by presentation (e.g. CRP, urine culture, NPA, etc)
- Commence patient on their unwell feeding plan (see emergency diet plan for volumes and recipes) via enteral route (PO/NG/PEG)
- If the patient is not tolerating oral intake, commence IV fluids with 0.9% NaCl + 10% dextrose + 2mmol/kg/day KCl at 100-120% of maintenance fluid requirements as an interim measure to provide calories
- Following discussion with the metabolic team, consider commencing stock/standard PN and/or IV 20% lipid infusion in patients who are not tolerating oral intake
- Give ondansetron for management of nausea and vomiting
- 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
- If there is derangement of the coagulation profile, give a stat dose of Vitamin K 300 micrograms/kg PO/IV (max dose 10mg). Further management may be discussed as needed with the metabolic and haematology teams
- Some patients will present with acute liver failure and require discussion with the paediatric liver service in CHI at Crumlin and with PICU
- **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).