British Inherited Metabolic Disease Group

PATIENT NAME **HOSPITAL**

DATE OF BIRTH **EMERGENCY CONTACT**
9-5pm Monday to Friday
Out of hours

ADULT EMERGENCY MANAGEMENT
MAPLE SYRUP URINE DISEASE (MSUD)

IMMEDIATE ACTIONS
Triage to high priority

Maintain glucose 6-10 mmol/L.

These guidelines are intended for immediate emergency management only. Please contact your local metabolic team early for specific advice on individual patients.

BACKGROUND

MSUD is a disorder affecting the breakdown of branched chain amino acids (BCAA = Leucine, Isoleucine & Valine). In MSUD, encephalopathy occurs as a result of accumulation of the BCAA (particularly leucine), which are toxic at high concentrations. Plasma amino acids can seldom be measured urgently, so management has to be based on the clinical state.

Decompensation is often triggered by metabolic stress such as febrile illness, particularly gastro-enteritis or fasting but an obvious cause is not always apparent. The early signs of decompensation may be subtle, such as **lethargy** or **ataxia**. **Vomiting** is common and should always be taken seriously. However, the signs may be difficult to assess such as **irritability** or just **'not right'**. Always listen to patients and their carers carefully. Their assessment of the conscious level and precipitating factors should be taken into account. Always take illness seriously as there is a risk of death or permanent neurological damage.

Treatment aims to:

(a) Inhibit protein catabolism and promote anabolism by providing high calorie intake combined with the patient's usual MSUD BCAA-free amino acid formula.

(b) Lower BCAA levels by stopping or restricting 'natural protein' ('exchanges'). If this is insufficient, BCAA can be removed by haemofiltration / dialysis
(c) Ensure a balance is maintained between leucine, isoleucine and valine during decompensation by giving supplements of individual amino acids as required.

If there is any doubt at all, the patient should be admitted, even if only for a short period of observation.

INITIAL ASSESSMENT AND MANAGEMENT IN HOSPITAL

If the patient is shocked or clearly very ill arrange for admission to ITU / HDU.

Management decisions should be based primarily on the **clinical** status. It is particularly important to note any degree of encephalopathy as the major complication is cerebral oedema presenting with progressive encephalopathy. If the patient is relatively well – they may be treated orally using their oral emergency regimen (click here) (generally give 200ml of a 25% glucose polymer (eg. Maxijul) solution every 2 hours) but assess very carefully. If the patient is obviously unwell – they must be treated with intravenous fluids.

Record the Glasgow Coma Scale (click here). This will allow early identification of encephalopathy and deterioration.

INITIAL INVESTIGATIONS

Blood pH and gases
Urea & electrolytes
Glucose
Full blood count
Amino acids (quantitative - ask lab to measure or refer for measurement ASAP)
Urine ketones
Other tests as indicated by the clinical findings (eg. CRP, blood & urine cultures)

TREATMENT

If at all possible and the patient is able to tolerate even small amounts orally, then continue to give their BCAA-free amino acid mixture and some energy orally or enterally eg. via an NG tube.

Also:
1. Correct dehydration initially with 0.9% NaCl.

2. Start an intravenous 10% dextrose infusion as soon as possible at a rate of 2mls/kg/hr, (e.g. 140 mls/hr in a 70 kg person).

4. Treat any underlying infection or other clinical problem.

5. Give analgesia, anti-pyretic or an anti-emetic as required.

6. Consider the possibility of refeeding syndrome in susceptible patients.

MONITORING

Reassess regularly and if there is a change for the worse repeat the clinical assessment and blood tests:

Blood pH & gases
Urea & electrolytes
Plasma amino acids – requesting leucine, valine, and isoleucine levels.

Glucose: Hyperglycaemia can occur. If the blood glucose exceeds 10 mmol/L, start an insulin infusion according to the local diabetic protocol rather than reducing the glucose intake. **Strict supervision is essential.** National guidelines are available at: (http://www.diabetes.org.uk/About_us/Our_Views/Care_recommendations/The-Management-of-Diabetic-Ketoacidosis-in-Adults/).

Potassium: Potassium concentration should be monitored and corrected appropriately.

If signs of encephalopathy develop - seek specialist help. ITU support will be required. Haemofiltration (haemodialysis) may need to be considered urgently.

RE-INTRODUCTION OF ORAL OR ENTERAL FEEDING

As the patient improves, oral or enteral feeds should be introduced as early as possible. The BCAA-free amino acid mixture should be given throughout. Natural protein intake should be reintroduced / increased as tolerated. Anti-emetics may be needed. Consult your local metabolic dietitian or specialist centre for more details. See the BIMDG oral emergency regimen (click here) for MSUD for more details.

MORE USEFUL INFORMATION

http://www.bimdg.org.uk/ and click on the red tab for emergency guidelines.
Genereviews: http://www.ncbi.nlm.nih.gov/books/NBK1116/
Pubmed: http://www.ncbi.nlm.nih.gov/pubmed/