British Inherited Metabolic Disease Group

PATIENT NAME **HOSPITAL**

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

ADULT EMERGENCY MANAGEMENT
MEDIUM CHAIN FAT OXIDATION DISORDERS

This protocol covers
- medium-chain acyl-CoA dehydrogenase (MCAD) deficiency,
- 3-hydroxy-3-methyl-glutaryl-CoA (HMG) synthase deficiency,
- carnitine palmitoyltransferase-1 (CPT1) deficiency.

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
MCAD deficiency is the most common disorder of fat breakdown. The treatment for other disorders such as HMG-CoA synthase deficiency and CPT1 deficiency is similar. Most of the time patients are healthy and do not require a special diet. However, intercurrent infection, prolonged fasting, excessive alcohol intake, vomiting or diarrhoea can lead to serious illness, with encephalopathy and even sudden death. This results from the accumulation of toxic fatty acids.

SIGNS OF DECOMPENSATION
The early signs of decompensation may be subtle – lethargy, nausea, vomiting or feeling 'not right'. Hypoglycaemia only occurs at a relatively late stage so that blood glucose / BMstix should not be relied

on. Do not delay treatment just because the blood glucose is not low. **The aim should always be to intervene whilst the blood glucose is normal**. Treatment aims to prevent mobilisation of fat by providing ample glucose - enterally or intravenously.

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. 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 solution every 2 hours) but assess very carefully. If the patient is obviously unwell – they must be treated with intravenous fluids.

INITIAL INVESTIGATIONS

Blood pH and gases
Glucose
Urea & electrolytes
Full blood count
Liver function tests
Other tests as clinically indicated (eg. CRP, Blood & urine cultures)

TREATMENT

1. Correct dehydration initially with 0.9% NaCl. Correct hypoglycaemia initially with 50ml of 50% dextrose over 30 minutes.

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

3. Treat any underlying infection or other clinical problem.

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

MONITORING

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

Blood pH and gases
Glucose
Urea & electrolytes

Please note that in disorders of fatty acid oxidation associated with an inappropriately low ketone body response neither blood nor urine ketone levels are reliable and should not be used to make treatment decisions.

Potassium: Hypokalaemia may occur so plasma potassium concentration should be monitored and corrected appropriately.

Patients should be monitored closely for deterioration. Clinical assessment should include the Glasgow Coma Scale (click here) and blood pressure.

RE-INTRODUCTION OF ORAL OR ENTERAL FEEDING

As the patient improves – restart oral or enteral feeding as soon as possible. See the BIMDG oral emergency regimen (click here) 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/