British Inherited Metabolic Disease Group

PATIENT NAME **HOSPITAL**

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

ADULT EMERGENCY MANAGEMENT
LONG CHAIN FATTY ACID OXIDATION DEFECTS

This protocol covers

- very-long-chain acyl-CoA dehydrogenase (VLCAD) deficiency,
- long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency,
- carnitine palmitoyltransferase (CPT 2) deficiency,
- carnitine-acylcarnitine translocase (CACT) deficiency,
- multiple acyl-CoA dehydrogenase (MAD) 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

These conditions are all disorders of the breakdown of fatty acids. The aim of treatment is to reduce the mobilisation and oxidation of fatty acids. Many, though not all, patients are treated with a low fat, high carbohydrate diet with avoidance of fasting. Some patients may also take regular medium chain triglyceride supplements in the form of MCT oil. Prolonged fasting is avoided – some patients have a continuous tube feed overnight or are woken and fed or take uncooked cornstarch before bed. Most of the time patients are healthy but intercurrent infections, prolonged fasting, alcohol excess, excessive exercise, vomiting or diarrhoea can lead to serious complications with rhabdomyolysis, encephalopathy and even sudden death.

SIGNS OF DECOMPENSATION

The early signs of decompensation may be subtle – lethargy, muscle aches and pain or feeling 'not right'. In adult patients muscle symptoms, including rhabdomyolysis may predominate. 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.

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

INITIAL INVESTIGATIONS

Blood pH and gases
Glucose
Urea, creatinine & electrolytes
Creatine kinase
Full blood count
Lactate
Blood spot or plasma acylcarnitine profile (contact your laboratory for advice)
Liver function tests
Urine myoglobin if available (use urine strip test for blood)
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 started as soon as possible at a rate of 2mls/kg/hr, (e.g. 140 mls/hr in a 70 kg person).

3. *(Carnitine may be given orally (NOT intravenously) - 100 – 200 mg/kg/24h in 4 divided doses. The role of carnitine in the emergency management of fatty acid oxidation disorders as listed above is controversial and current recommendations are that it should be avoided (J Inherit Metab Dis (2009) 32:498-505). Please discuss first with metabolic team on–call.)*

4. Treat any underlying infection or other clinical problem.

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

6. If significant renal impairment then consider dialysis.

COMPLICATIONS

Cardiomyopathy Arrange echocardiography and ECG if there are signs of cardio-respiratory problems or arrhythmia.

Rhabdomyolysis (muscle breakdown) is a common problem, particularly in VLCAD, CPT2 or LCHAD deficiency. Rhabdomyolysis may cause acute renal failure.

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
Creatine kinase

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. 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.

RE-INTRODUCTION OF ORAL OR ENTERAL FEEDING

As the patient improves, oral or enteral feeds should be introduced as early 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/