



British Inherited Metabolic Disease Group

PATIENT NAME

HOSPITAL

DATE OF BIRTH

EMERGENCY CONTACT

**9-5pm Monday to Friday
Out of hours**

ADULT EMERGENCY MANAGEMENT

HMG CoA Lyase Deficiency

(also known as 3-hydroxy-3-methyl glutaryl CoA lyase 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

HMG CoA lyase deficiency is a disorder of ketone synthesis. This enzyme is also on the leucine breakdown pathway. For most of the time patients are healthy and do not require a special diet. However intercurrent infection, fasting, alcohol excess, diarrhoea or vomiting or morning sickness can lead to serious illness with encephalopathy and stroke-like episodes.

Early signs of decompensation may be subtle and non-specific eg lethargy. 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.**

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.

Careful clinical assessment is required including blood pressure and the [Glasgow Coma Scale \(click here\)](#), this is essential even if the patient does not appear encephalopathic.

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, ammonia)

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). Ensure normoglycaemia is maintained. **NOTE: Patients can quickly become hypoglycaemic if intravenous access fails / tissues – ensure iv access is secure.**
3. Treat any underlying infection or other clinical problem. Replace electrolytes as required.
4. Give analgesia, anti-pyretic or an anti-emetic as required.
5. Consider the possibility of refeeding syndrome in susceptible patients.

Acidosis can be marked but sodium bicarbonate is not given routinely. However, if acidosis persists after correction of blood glucose and perfusion, sodium bicarbonate may be needed if pH <7.1 or pH is deteriorating rapidly or base deficit is greater than 15mmol/L. If repeat doses of bicarbonate appear to be needed consider alternative explanations such as sepsis.

Hyperglycaemia can be a problem. If the blood glucose exceeds 10 mmol/L start an insulin infusion using the local diabetic protocol rather than reducing the glucose intake. **Strict supervision is essential.**

National guidelines are available at:

[\(http://www.diabetes.org.uk/About_us/Our_Views/Care_recommendations/The-Management-of-Diabetic-Ketoacidosis-in-Adults/\)](http://www.diabetes.org.uk/About_us/Our_Views/Care_recommendations/The-Management-of-Diabetic-Ketoacidosis-in-Adults/).

COMPLICATIONS

The major complications are hypoglycaemia, encephalopathy, stroke-like episodes and metabolic acidosis.

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

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

Clinical assessment should include the [Glasgow Coma Scale \(click here\)](#) and blood pressure.

Patients should remain on iv dextrose until tolerating oral food normally. See the BIMDG general [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.

Pubmed: <http://www.ncbi.nlm.nih.gov/pubmed/>