British Inherited Metabolic Disease Group

PATIENT NAME: **HOSPITAL:**

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

ADULT EMERGENCY MANAGEMENT

GLYCOGEN STORAGE DISEASE TYPE 1

(also known as glucose-6-phosphatase (GSD1a) or glucose-6-phosphate translocase (GSD1b) deficiencies)

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

Glycogen storage disease type 1 (GSD1) is a disorder of glycogen breakdown and gluconeogenesis. Adult patients with GSD1 may become hypoglycaemic if they fast for longer than about 3-4 hours: and sometimes even after a much shorter time. During illness, patients have an even greater tendency to become hypoglycaemic. Emergency treatment aims to maintain normoglycaemia all the time either with frequent oral drinks, a continuous infusion via a nasogastric tube or gastrostomy or an intravenous infusion of glucose. This management should be started as soon as patients become unwell. The early signs of decompensation may be subtle and some patients have loss of hypoglycaemic awareness and may remain asymptomatic despite marked hypoglycaemia and metabolic acidosis. Others may present with a hypoglycaemic convulsion.

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
Lactate
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. If glucose cannot be quickly given intravenously then give Glucogel® into the buccal cavity. **NOTE: GSDI is NOT similar to diabetes – Glucagon does NOT work for GSDI.**

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 with GSD1 can quickly become hypoglycaemic if intravenous access fails / tissues – ensure intravenous 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.

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 consistently exceeds 10 mmol/L then reassess and either:

1) start an insulin infusion using 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/).
   or
2) if the patient is clinically improved and able to tolerate oral intake then reduce the rate of the glucose infusion and consider switching to their oral emergency regimen (click here).

COMPLICATIONS

Other potential complications of GSDI, include: platelet dysfunction with normal count (bleeding tendency), anaemia, gout, and renal tubular wasting. It is important to consider these issues when a patient presents to casualty and consider performing appropriate additional tests.

Patients with GSD 1b may also commonly have neutropenia with recurrent infections that may be life threatening, or a clinical presentation similar to inflammatory bowel disease.

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
Lactate

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 oral emergency regimen (click here) for more details.

MORE 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/