



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 III**

(also known as glycogen debrancher deficiency or Cori-Forbes disease)

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 III (GSDIII) is caused by a deficiency in glycogen debrancher enzyme leading to excess amounts of abnormal glycogen being deposited in the liver, muscles and in some cases the heart. Adult patients with GSDIII may become hypoglycaemic during a fast, the length of which varies widely between patients. Patients may have a greater tendency to become hypoglycaemic during intercurrent illness. The treatment aims to maintain normoglycaemia all the time either with frequent oral drinks, a continuous infusion via 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

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 given intravenously quickly then give Glucogel® into the buccal cavity. **NOTE: GSDIII is NOT similar to diabetes – Glucagon does NOT work for GSDs.**
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 GSDIII 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.

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

Cardiomyopathy, skeletal myopathy (including respiratory muscle involvement) and/or cirrhosis may occur in some patients.

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

Clinical assessment should include [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 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/>