



British Inherited Metabolic Disease Group

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This protocol has 4 pages

FRUCTOSE-1,6-BISPHOSPHATASE DEFICIENCY- ACUTE DECOMPENSATION
(also called fructose 1,6 diphosphatase deficiency)
(standard version)

- **Please read carefully. Meticulous treatment is important as there is a high risk of complications.**
- **If the instructions do not make sense or a problem is not addressed you must discuss your concerns with the consultant on call.**

1. Background

Fructose-1,6-bisphosphatase deficiency is a disorder of gluconeogenesis. Most of the time patients are healthy & do not require a special diet. Infections, fasting, diarrhoea or vomiting can lead to serious illness, with drowsiness, tachypnoea, marked acidosis & hypoglycaemia. However hypoglycaemia occurs at a relatively late stage. Treatment aims to prevent metabolic decompensation by giving glucose - by mouth or intravenously. In young children, bicarbonate is often needed to correct acidosis.

During illness, patients must not be given fructose or sucrose.

The early signs of decompensation may be subtle but particularly look for tachypnoea. Always listen to parents carefully as they probably know much more than you do. As hypoglycaemia only occurs at a relatively late stage, intervention should take place whilst the blood glucose is normal.

2. Admission

Most patients who present to hospital will require admission as they are likely to have been having treatment already at home. Only allow the child home if you and the family are entirely happy and you have discussed the problems with the consultant on call. The family must have a clear management plan and be prepared to return if the child does not improve.

- **If there is any doubt, the child must be admitted, even if only necessary for a short period of observation.**

3. Initial plan and management in hospital

⇒ If the child is shocked or clearly very ill arrange for admission to ITU/High dependency.

⇒ If admitted to metabolic/general ward, make a careful clinical assessment including blood pressure and a [Glasgow coma score \(for details click here\)](#), even if the patient does not appear encephalopathic. This enables other staff to recognise if the child deteriorates, particularly around the time of a change of shifts.

The following blood tests should be done:

- pH and gases
- Glucose (laboratory and bedside strip test)
- Urea and electrolytes
- Full blood count
- Lactate
- Blood culture

4. Management

Management decisions should be based primarily on the **clinical** status. The first decision about therapy is whether the child can be treated orally or will need intravenous therapy.

- Can the child tolerate oral fluids?

If the child is relatively well - may be treated orally but assess very carefully.

If the child is obviously unwell - must be treated with intravenous fluids

- **If there is any doubt at all, put up an intravenous line.**

Treat any infection

A. ORAL.

If the child is relatively well, with no vomiting or no respiratory distress, oral feeds may be given. The emergency regimen should be used. This may be given as regular frequent drinks but if the patient is at risk of vomiting or is nauseated, fluid should be given either continuously or as small boluses more frequently. For more information about the [emergency oral management click here](#)

Age (years)	Glucose polymer concentration (g/100ml) *	Total daily volume**
0-1	10	150-200 ml/kg
1-2	15	100 ml/kg
2-6	20	1200-1500 ml
6-10	20	1500-2000 ml
>10	25	2000 ml

* If necessary, seek help from your local dietitian. In an emergency a heaped 5 ml medicine spoon holds approximately 7g of glucose polymer.

** For each drink the volume will generally be this figure divided by 12 and given 2 hourly but if the patient is nauseated or refuses try frequent smaller drinks or a continuous naso-gastric infusion.

Electrolytes should be added to the drinks if vomiting and/or diarrhoea is a problem using standard rehydration mixtures following manufacturer's instructions but substituting glucose polymer solution for water.

Warning: Fructose should not be given during acute illness. As oral medicines may contain sucrose and sorbitol, only sugar free medicines should be given but still check constituents.

B. INTRAVENOUS.

If the child is unwell

- Give Glucose 200 mg/kg **at once** (2 ml/kg of 10% glucose or 1ml/kg of 20% glucose) over a few minutes.
- Give normal saline 10 ml/kg as a bolus immediately after the glucose unless the peripheral circulation is poor or the patient is frankly shocked, give 20 ml/kg normal saline instead of the 10 ml/kg.. Repeat the saline bolus if the poor circulation persists as for a shocked non-metabolic patient.
- Continue with glucose 10% at 5 ml/kg/h **ONLY until next solution is ready– do not leave on this high rate longer than necessary.** – see below
- Quickly calculate the deficit and maintenance and prepare the intravenous fluids
 - Deficit: estimate from clinical signs if no recent weight available
 - Maintenance: Formula for calculating daily maintenance fluid volume (BNF for children) 100ml/kg for 1st 10kg then 50 ml/kg for next 10kg then 20ml/kg thereafter, using calculated rehydrated weight. Deduct the fluid already given from the total for the first 24 hours.
 - Give 0.45% saline/10% glucose ([for instructions to make this solution click here](#)).
- Having calculated the deficit and the maintenance, administer the appropriate rate of 0.45% saline/10% glucose to correct the deficit within 24 hours
- Recheck the electrolytes every 24 hours if still on IV fluids.

- Potassium can be added, if appropriate, once urine flow is normal and the plasma potassium concentration is known.

- Sodium bicarbonate is not given routinely as any acidosis usually corrects quickly, but sodium bicarbonate may be needed if, after the initial bolus of glucose, the pH is still <7.1 or the pH is deteriorating rapidly or the base deficit is still greater than 15 mmol/l.

Initially give a half correction [$0.15 \times \text{weight} \times \text{base deficit (mmol/l)}$] mmol sodium bicarbonate over at least 30 minutes. 1 ml of sodium bicarbonate 8.4% contains 1 mmol but this solution should be diluted *at least* 1ml to 5ml of 5% glucose. Then review and check U&E and pH & blood gases. As the acidosis normally corrects quickly, repeat doses of sodium bicarbonate are rarely needed. If further doses of sodium bicarbonate appear to be necessary, discuss with the consultant. Before doing so ask why? Is there another explanation such as sepsis? If further doses are given, reduce sodium chloride given in the other intravenous fluids.

- Hyperglycaemia can be a problem. If the blood glucose persistently exceeds the 8 mmol/l, start an insulin infusion using the local diabetes protocol rather than reducing the glucose intake.

Strict supervision is essential.

5. Progress:

Monitoring: Reassess after 4-6 hours or earlier if there is any deterioration or no improvement
Clinical assessment should include [Glasgow coma score \(for details click here\)](#) and blood pressure. Blood tests:

- Blood pH and gases
- Glucose (laboratory)
- Urea & electrolytes,

⇒ If improving, continue and for intravenous fluids after 24 hours, please refer to the previous section.

⇒ If deteriorating, seek specialist help without delay.

6. Re-introduction of oral feeds: Restart oral feeds as soon as possible; once the child is alert and has stopped vomiting. If necessary, consult your local dietitian for more details.

Sucrose, sorbitol and fructose should be avoided at this stage as much as possible. Beware oral medicines as these may contain sucrose and sorbitol!

7. Going Home: Only allow the child home if you and the family are entirely happy and you have discussed the problems with the consultant on call. The family must have a clear management plan and be prepared to return if the child deteriorates.

For further information please refer to:

Saudubray J-M, van den Berghe G, Walter JH. (editors) Inborn Metabolic Diseases. Diagnosis and treatment. 5th Edition. Springer 2012