BIMDG

British Inherited Metabolic Disease Group

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This protocol has 4 pages

CARNITINE TRANSPORTER DEFICIENCY (CTD) – ACUTE DECOMPENSATION.

(standard version)

- **Please read carefully. Meticulous treatment is important as there is a high risk of serious complications.**

- **If the instructions do not make sense or a problem is not addressed you must discuss your concerns with the consultant on call.**

- **Intervene whilst the blood glucose is still normal.**

1. Background

Carnitine transporter deficiency usually presents with cardiomyopathy and muscle weakness. It responds well to carnitine supplementation and for most of the time patients are healthy. However infections, fasting and particularly vomiting and diarrhoea can lead to serious illness with encephalopathy, hypoglycaemia and potentially worsening cardiomyopathy.

The early signs of decompensation may be subtle e.g. lethargy or 'floppiness'. Always listen to parents carefully as they probably know much more than you do.Hypoglycaemia only occurs at a relatively late stage (or very late) 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 restore carnitine concentrations and reduce mobilisation of fat by providing ample glucose - enterally or intravenously.

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 at all, 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 (click here), even if the patient does not appear encephalopathic. This allows 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 blood gases
Glucose (laboratory and bedside strip)
Urea & electrolytes
Full blood count
Blood culture

If acute cardiomyopathy is suspected, arrange an urgent Chest X-Ray, ECG and/or echocardiogram.

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 and not vomiting, medication and feeds may be given orally. Give carnitine 100 mg/kg at once. If there is a risk of vomiting, give the dose slowly over 30 minutes.

For feeds the emergency regimen should be used. Do not delay. 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

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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.
- Give carnitine at the same time 100 mg/kg over 30 minutes
- 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.

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

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

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. In the presence of cardiomyopathy, continuous ECG monitoring is advised.

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

⇨ If improving, continue and for intravenous fluids after 6 hours, please refer to the previous section. Ensure carnitine treatment continues at 100mg/kg/day (usually in three divided doses)

⇨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, seek specialist advice.

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