British Inherited Metabolic Disease Group

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This protocol has 4 pages

CARNITINE PALMITOYL TRANSFERASE 1 (CPT1) DEFICIENCY AND HMG COA SYNTHASE DEFICIENCY

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.**
- **Intervention should occur whilst the blood glucose is still normal.**

1. Background

The treatment for these disorders is similar to that for MCAD Deficiency. For most of the time patients are healthy & do not require a special diet. However infections, fasting, diarrhoea or vomiting can lead to serious illness, with encephalopathy and even sudden death. This results from the accumulation of toxic fatty acids.

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 prevent 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.

- **Inform Metabolic Team on admission**
- **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 (for details 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 shift.

The following blood tests should be considered:
pH and gases
Glucose (laboratory and bedside strip test)
Urea and electrolytes
Full blood count

Consider other tests as clinically indicated.

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?
- Is the child dehydrated? – Note this can be difficult to assess. The best guide is the difference between the current weight and a recent one when well.

Mild: up to 5% weight loss - may be treated orally but assess carefully.
Moderate or severe: >5% - must be treated with intravenous fluids

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

Treat any infection

MANAGEMENT CONTINUED ON NEXT PAGE

A. ORAL.

If the child is relatively well and not vomiting, oral feeds may be given.
The emergency regimen should be used.
Do not delay.

FULL ENTERAL EMERGENCY FEED – glucose polymer solution
Use patient's own ER recipe.
Use age-based ER recipes below if not available.
If ER products not available use IV guidelines.

NB: MCT feeds and supplements contraindicated in HMG CoA Synthase Deficiency
Oral rehydration solutions are low in CHO and not suitable

- Click Here for Emergency Regimen for Age ≤ 1 year (10%)
- Click Here for Emergency Regimen for Age 1- 2 years (15%)
- Click Here for Emergency Regimen for Age 2-9 years (20%)
- Click Here for Emergency Regimen for Age ≥ 10 years (25%)

EMERGENCY FEED ADMINISTRATION

- give feeding volume for body weight (see recipe)
- feed orally: 2 hourly day and night
- if not tolerated or fluid requirements not met, administer, continuously by tube, without delay
- administer bolus or continuously by tube feed, without delay for a maximum of 24-36hours
- introduce usual diet/feeds as soon as clinically stable

Medications

- antipyretics: as clinically indicated

Contact the child's specialist metabolic team and dietitian for further advice on the ER and introduction of usual diet/feeds

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 0.9% 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 0.9% 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).
    - Potassium can be added once the plasma potassium concentration is known and the child is passing urine.
- 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 intravenous fluids.
- If hyperglycaemia is a problem, it is preferable to reduce the glucose concentration to 5% rather than using insulin.

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.

- If still obviously encephalopathic continue intravenous fluids but if able to take oral fluids safely, switch to drinks by mouth.

- If deteriorating, seek specialist help without delay.

6. Re-introduction of oral feeds: Intravenous fluids should not be decreased too quickly. Once the child is alert and has stopped vomiting oral feeding can be restarted and intravenous fluids reduced. Only allow the child home if you and the family are entirely happy. **It must be clearly demonstrated that the child can tolerate at least two successive feeds / meals before discharge.** 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, Baumgartner MR, Walter JH. (editors) Inborn Metabolic Diseases. Diagnosis and treatment. 6th Edition. Springer 2016