BIMDG

British Inherited Metabolic Disease Group

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This protocol has 4 pages

LONG CHAIN FAT OXIDATION DISORDERS – ACUTE DECOMPENSATION

VLCAD DEFICIENCY
LCHAD DEFICIENCY
CPT II DEFICIENCY **(severe early onset variant),**
CACT (CARNITINE ACYLCARNITINE TRANSLOCASE) DEFICIENCY **(late onset),**
MULTIPLE ACYL COA DEHYDROGENASE DEFICIENCY (MADD)

(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.**

1. Background

These conditions are all disorders of the breakdown of fatty acids. The patients are treated with a low fat high carbohydrate diet with avoidance of fasting. (Even overnight fasting is avoided - patients either have a continuous tube feed or are woken and fed; some children take uncooked corn-starch before bed). Most of the time patients are healthy but infections, fasting, diarrhoea or vomiting can lead to serious illness with encephalopathy and even sudden death. The early signs of decompensation may be subtle, lethargy and /or 'floppiness'. Always listen to parents carefully as they probably know much more than you do.Hypoglycaemia only occurs at a relatively late stage so that bedside test for glucose 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. 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 even if the patient does not appear encephalopathic enter a Glasgow coma score (for details click here). This is very important since should the child deteriorate, particularly around the time of a change of shift, the new team will recognise any change.

The following tests should be done:

BLOOD	pH and gases
	Glucose (laboratory and bedside strip test)
	Urea and electrolytes
	Full blood count
	Lactate
	Blood spot acylcarnitines
	CK, Liver function tests
	Blood culture
Urine	myoglobin (use urine strip test for blood)

Complications

1. Cardiomyopathy. This may be problem, particularly at presentation. Arrange 2D Echocardiography if there are signs of cardio-respiratory problems

2. Rhabdomyolysis (muscle breakdown) is a common problem in severe VLCAD, LCHAD and CPT2 deficiency. The rhabdomyolysis may be exercise-induced, particularly in some older patients and may cause myoglobinuria and /or acute renal failure.

   If there is rhabdomyolysis – click here for guidance on management

3. 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.

- Factors that will influence the decision include, how ill is the child and whether they have deteriorated suddenly in the past?
- 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.**

A. ORAL.

If the child is relatively well and not vomiting 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 the patient has diarrhoea and vomiting using standard rehydration mixtures following manufacturer's instructions but substituting glucose polymer solution for water.

Medicines

- Some patients may be given carnitine orally- 100 mg/kg/24h in 4 divided doses
- Treat any infection

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). **Caution: fluid overload in those with cardiomyopathy**
    - Potassium can be added, if necessary, 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 IV fluids.

- Some patients may be given carnitine orally (**not** intravenously because long chain acyl carnitines may be arrhythmogenic) - 100 – 200 mg/kg/24h in 4 divided doses

- Treat any infection

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)
U&E,

If improving, continue and for intravenous fluids after 6 hours, please refer to the previous section.
If deteriorating, seek specialist help.

6. Re-introduction of oral feeds: As many more calories can be given orally safely oral feeds should be introduced as early as possible. It is usual to give soluble glucose polymer initially 10% and increase this both volume and concentration as tolerated. It is customary to delay the introduction of any protein or aminoacids but this will only prolong the period of catabolism. If necessary, consult your local dietitian for more details.

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, Baumgartner MR, Walter JH. (editors) Inborn Metabolic Diseases. Diagnosis and treatment. 6th Edition. Springer 2016