British Inherited Metabolic Disease Group

This protocol has 3 pages

MEDIUM CHAIN ACYL CoA DEHYDROGENASE DEFICIENCY (MCADD) – ACUTE ILLNESS AND METABOLIC DECOMPENSATION

IMMEDIATE ACTION

- **Triage to high priority**
- **Hypoglycaemia occurs late – do not delay treatment even if blood glucose is normal**
- **Management should be based upon clinical status: '2. MANAGEMENT IN HOSPITAL'**
- **If the child is unwell and/or vomiting start intravenous fluids: '2B. INTRAVENOUS'**
- **Ongoing management should be guided by the child's specialist metabolic team. Contact them as soon as possible for advice.**

1. BACKGROUND AND SIGNS OF DECOMPENSATION

- MCAD deficiency is a disorder of fat catabolism. The clinical manifestations occur due to energy deficit during fasting, mainly during acute illnesses with reduced oral intake
- Infections, fasting, diarrhoea or vomiting can lead to serious illness with encephalopathy and even sudden death
- The early signs of decompensation may be subtle e.g. lethargy or 'floppiness', and results from the accumulation of toxic fatty acids
- Treatment aims to prevent catabolism by giving glucose either enterally or intravenousy l
- Hypoglycaemia only occurs at a late stage. Do not delay treatment even if blood glucose is normal.

2. MANAGEMENT IN HOSPITAL

- If the child is shocked or clearly very unwell, consider admission to PICU/HDU
- If admitted to a general/metabolic ward careful clinical assessment is essential including regular PEWS and neurological observations even if the patient does not appear encephalopathic
- 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
- The first decision about therapy is whether the child should be treated enterally (2A) or intravenously (2B). If there is any doubt, start IV fluids

- Intravenous fluids are indicated if the child is unable to tolerate oral fluids, is vomiting or has moderate or severe clinical dehydration

- Treat any infection.

2A. ENTERAL If the child is relatively well and not vomiting, try to give the emergency regimen (ER) feeds/drinks orally. If necessary, young children (typically under 2 years) can have a nasogastric tube inserted to administer the feeds. Emergency regimen feeds/drinks are based on glucose polymer solutions. The CHO concentration is age dependent and increases with age (see below). Fluid volumes to administer are calculated using the Holliday-Segar Formula for Maintenance Fluid Requirements by weight.

FULL ENTERAL EMERGENCY REGIMEN FEED/DRINKS

Use the patient's own ER recipe wherever possible
Use age-based ER recipes below if not available
If ER products are not available follow IV guidelines

NB: Feeds or supplements with added medium chain triglycerides are contraindicated in MCADD

Oral rehydration solutions are low in CHO and not suitable, unless given in addition to the ER feeds/drinks to help correct dehydration

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

EMERGENCY REGIMEN FEED/DRINK ADMINISTRATION

- Divide the maintenance volume over 24 hours: give 1-2hourly oral or by tube (bolus or continuous)
- If not tolerated follow IV therapy (2B)
- Introduce usual diet/feeds when child is ready to eat again
- Continue some ER feeds during normal diet reintroduction, particularly at night to reduce the fasting time.

Medications

- Antipyretics: as clinically indicated

Contact the child's specialist metabolic team and dietitian for further advice on the emergency regimen and re-introduction of usual diet plan

2B. INTRAVENOUS If the child is unwell and or vomiting, start IV fluids

IMMEDIATE FLUID RESUSCITATION:

- Give Glucose 200 mg/kg **if there is hypoglycaemia** (2ml/kg of 10% glucose or 1ml/kg of 20% glucose over a few minutes if blood glucose <3.0mM)

- Give 0.9% sodium chloride 20 ml/kg as a bolus **if the peripheral circulation is poor or the patient is in shock.** Repeat the sodium chloride bolus if poor circulation persists.

INITIAL FLUIDS AFTER RESUSCITATION:

- Run IV fluids of Glucose 10%/Sodium Chloride 0.45% at 5ml/kg/h ONLY until accurate fluid rates have been calculated **– do not leave on this high rate longer than necessary**. (for instructions to make this solution click here).

FURTHER FLUID MANAGEMENT IN FIRST 24 HOURS:

- Ongoing IV management is based upon administering maintenance fluids over 24 hours as Glucose 10%/Sodium Chloride 0.45% plus any calculated fluid deficit
- Deduct any fluid given during resuscitation from the total volume for the first 24 hours
- Potassium can be added once the plasma potassium concentration is known and the child is passing urine.

PROGRESS/MONITORING:

- Reassess after 4-6 hours or earlier if there is any deterioration or no improvement
- Clinical assessment should include PEWS and neurological observations
- If there is clinical deterioration seek specialist metabolic help.

Reassess hydration status and the need for ongoing IV fluids after 24 hours.

3. RE-INTRODUCTION OF ORAL FEEDS or FOOD:

- Intravenous fluids should not be stopped abruptly
- Introduce usual diet/feeds when child is ready to eat again
- Consider reducing IV fluid rate by 50% initially during the day when starting to re-introduce oral feeds or food. Emergency feeds may be given as the first feed but standard feeds or food given next.
- IV fluids should only be discontinued once enteral feeds are tolerated, and at least two successive feeds or meals.
- Continue some ER feeds during normal diet reintroduction, particularly at night to reduce the fasting time.

4. DISCHARGE

- Only discharge the child home if you and the family are entirely happy
- The child should have tolerated at least two successive oral feeds or meals before discharge
- The family must have a clear management plan for home of returning to the child's normal diet or feeds and when to stop any emergency feeds
- Be prepared to readmit if the child deteriorates.

For further information please refer to:

Merritt JL 2nd, Chang IJ. Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency. 2000 Apr 20 [Updated 2019 Jun 27]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2021. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1424/