BIMDG

British Inherited Metabolic Disease Group

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

IMMEDIATE ACTION: TRIAGE TO HIGH PRIORITY

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1. Clinical Assessment (sections 1 & 2)

Please listen to parents' concerns carefully.
Hypoglycaemia occurs late – do not delay treatment, even if blood glucose is normal.
Other complications include encephalopathy, cardiac arrhythmias and sudden death.

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2. IV Access & Bloods (section 3)

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3. Contact Metabolic/ PICU/ Transport Team EARLY if critically unwell

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4. Management – key considerations (section 4):

- Is the patient tolerating emergency regimen/ feed or do they require intravenous therapy?
- Treat underlying cause of decompensation (e.g. intercurrent infection)
- Is symptomatic management required (e.g. antipyretics, antiemetics)?

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5. Ongoing management & monitoring (sections 5, 6 & 7)

These guidelines only cover the first 24 hours of management.

Contact the child's specialist Metabolic team early to inform them regarding the admission and discuss ongoing management.

This protocol has 7 pages

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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 intravenously.

- Hypoglycaemia only occurs at a late stage. Do not delay treatment even if blood glucose is normal.

2. Clinical assessment to guide decisions about management/ admission:

Clinical assessment should focus on the following:

- Vomiting - either as a cause or result of metabolic decompensation. This will guide further fluid management.

- Diarrhoea – this can be exacerbated by the emergency regimen glucose polymer drink, and may influence decisions about management with IV fluids.

- Intercurrent infection or other illness triggering metabolic decompensation.

- Neurological status – look for signs of encephalopathy

- Cardiovascular status – risk of cardiac arrhythmias in decompensated MCADD

- **Remember that hypoglycaemia in MCADD is a late sign and a normal blood glucose result is non-reassuring. Do not delay treatment in an unwell patient with normal blood glucose.**

Almost all patients who present to hospital will require admission (as they will likely have been self-managing at home with their emergency regimen prior to presenting).

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

If the child is shocked or clearly very ill, consider admission to ITU/ High Dependency.

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.

3. Investigations

- The following blood tests should be considered:

Blood:
Gas (pH, glucose)
Urea & electrolytes, Glucose
Full blood count

Other tests:
As clinically indicated (infection screen, CXR, etc.).

4. Management (fluids and medications)

Management decisions should be based primarily on the **clinical status**.

- The first decision about therapy is whether the child can be treated orally (section 4.1) or will need intravenous therapy (section 4.2).

- Factors that will influence the decision include:
  - How ill is the child?
  - Can they tolerate oral fluids/ medications?
  - Have they deteriorated suddenly in the past?

- Intravenous fluids are indicated if:

  1. The child is unable to tolerate oral fluids, or
  2. There is moderate or severe clinical dehydration

- If there is any doubt, start IV fluids.

4.1. Patient tolerating oral/ enteral feeds

- 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 carbohydrate 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 patient's own ER recipe where possible; use age-based ER recipes below if not available If ER products are not available, follow IV guidelines
NB: Feeds/ supplements with added medium chain triglycerides are contraindicated in MCADD **Oral rehydration solutions are low in carbohydrate and are therefore 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% carbohydrate) • Click Here for Emergency Regimen for Age 1- 2 years (15% carbohydrate) • Click Here for Emergency Regimen for Age 2-9 years (20% carbohydrate) • Click Here for Emergency Regimen for Age ≥ 10 years (25% carbohydrate)
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 (section 4.2) • 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/ antiemetics/ antibiotics: 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

4.2. Patient requires intravenous therapy:

If the child is unwell and not tolerating oral/ enteral feeds, start IV fluids.

4.2.1. Immediate fluid resuscitation:

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

- **If the peripheral circulation is poor or the patient is in shock,** give a 10 ml/kg bolus of plasmalyte/ balanced crystalloid as per Advanced Paediatric Life Support guidance (use 0.9% Sodium Chloride if plasmalyte not available). Repeat the bolus if the poor circulation persists, as for a shocked non-metabolic patient.

4.2.2. Initial fluids after resuscitation:

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

- For instructions on how to make this solution click here.

4.2.3. Further fluid management in first 24 hours:

- Ongoing IV management is based upon administering maintenance fluids over 24 hours as 10% Glucose/ 0.45% Sodium Chloride 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.

5. Monitoring progress:

- 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.**

6. Re-introduction of enteral feeds:

- 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.

7. Discharge planning

- 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/