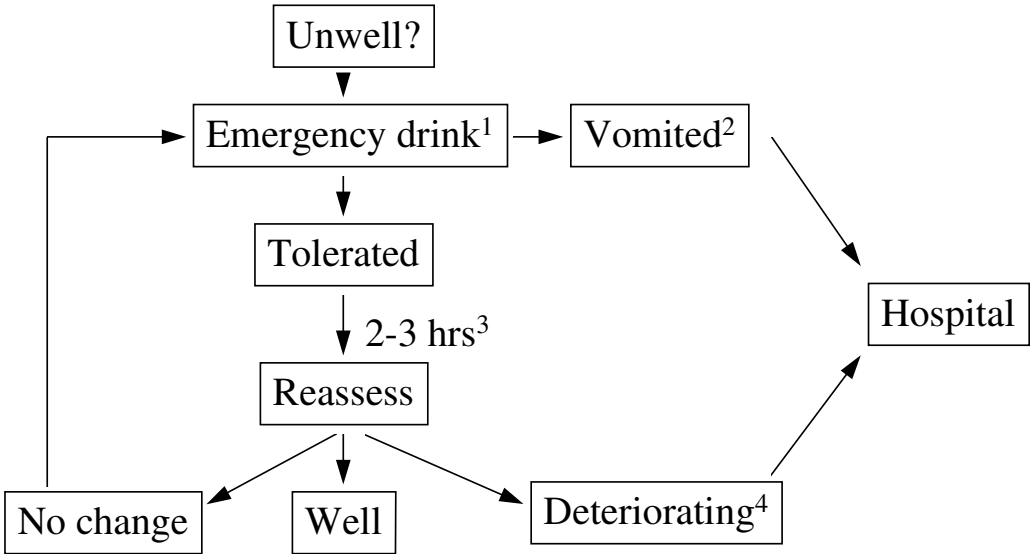
Guidelines for management of patients with MCAD deficiency during intercurrent illness

Background

MCAD deficiency is the commonest disorder of fat breakdown. Most of the time patients are healthy & do not require a special diet. Infections, fasting or vomiting can lead to serious illness, with encephalopathy and a risk of sudden death. This results from the accumulation of toxic fatty acids; hypoglycaemia also occurs, but only at a relatively late stage: it is dangerous to base one's treatment on monitoring of blood glucose, particularly BM stix. Treatment aims to inhibit mobilisation of fat by providing ample glucose - enterally or intravenously.

Management at home

Patients are generally instructed to follow this procedure:



Notes:

1. Volumes and concentrations of carbohydrate-containing drinks vary with age - each child will have their own instructions. Guidelines are presented below, derived from Dixon & Leonard, 1992, Arch Dis Child 67:1387-91. Glucose polymer preparations include maxijul, polycal etc.

Age (years)	Glucose polymer concentration (g/100ml)	Total daily volume*
0-1	10	150-200 ml/kg
1-2	15	95 ml/kg
2-6	20	1200-1500 ml
6-10	20	1500-2000 ml
>10	25	2000 ml

*For each drink the volume will be this figure divided by 12 or 8 (given 2 or 3 hrly).

2. If the parents are experienced & feel confident that their child is stable, they may try repeating the drink after a short interval but if this is still unsuccessful, admission is needed.

3. Patients should be reviewed & given carbohydrate-containing drinks every 2 hrs, day and night. May be increased to 3 hrs in older children.

4. Under these circumstances, admission is URGENT, particularly if the child has a glazed look.

Hospital management

1. Patients should always be admitted if the parent is sufficiently concerned to bring the child to hospital.

2. Find out why the child has been brought to hospital (vomiting, refusing drinks etc) & assess the patient quickly. If the problem is refusal of drinks or a single vomit, and there is no suggestion of incipient encephalopathy (such a glazed look or drowsiness), the child can be offered another drink orally or given it through a nasogastic tube.

3. If the child is unwell, check glucose, U&Es, blood gas, possibly NH_3 & other tests as appropriate.

4. Most children will require an intravenous infusion of glucose, which should be started without delay. Start with a small bolus (1 ml/kg of 25% glucose or 2.5 ml/kg of 10% glucose) and follow this with an infusion of 10% glucose, at the rates suggested below. Check electrolytes, but in the short term, it is usually unnecessary to add electrolytes to the infusion. Extra fluid should be given if the child is dehydrated or shocked, as for other patients.

Age (years)	**Weight (kg)**	**Glucose to be provided**	**10% glucose infusion rate**
0-2		10 mg/kg/min	150 ml/kg/day
2-6		8 mg/kg/min	120 ml/kg/day
>6	<30	6 mg/kg/min	90 ml/kg/day
>6	30-50	4.5 mg/kg/min	67 ml/kg/day
>6	>50	3 mg/kg/min	45 ml/kg/day

5. If there is any hint of incipient encephalopathy, start neurological observations - at least hourly.

6. Monitor BM 4 hourly if the child's condition is stable. If unstable, monitor BM hourly with blood glucose, U&Es & blood gas 6 hourly.

7. If the patient is not improving, seek specialist help. In particular, if there is concern about cerebral oedema or fluid overload, the concentration of the infusion should be increased, allowing the volumes to be reduced: this will require central venous access.

8. Allow the child to eat and drink when he wants to (unless further vomiting seems likely) and, once this is seen to be tolerated, discontinue the IVI.

9. Never discharge the patient until the parents are happy.

If you have any questions, please contact your local paediatrician or the familys metabolic consultant Dr. Allan Meldgaard Lund at Copenhagen University Hospital: +4535453887