BIMDG
British Inherited Metabolic Disease Group

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This protocol has 3 pages

MCAD DEFICIENCY
MANAGEMENT OF NEWBORN BABIES WITH A FAMILY HISTORY.

• Please read this carefully as there is a risk of death and serious complications in the newborn period.

MCAD deficiency is an inherited disorder of fat breakdown and one of the commonest inborn errors of metabolism. Most of the time patients are healthy and do not require a special diet. Infections, fasting or vomiting can lead to serious illness, with encephalopathy (drowsiness, seizures etc) 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 so it is not safe to base the management on monitoring of blood glucose, particularly with bedside glucose strips. The aim of treatment is to provide an alternative energy substrate and inhibit mobilisation of fatty acids by providing ample glucose - enterally or intravenously.

Problems are common in the newborn period. These can be avoided if appropriate management is given. It is essential to manage these patients appropriately at this time. It is essential to establish promptly whether the baby is affected and needs continuing treatment.

1. DIAGNOSIS

When the mother is admitted in labour (or, failing this, when the baby is born) inform the consultant paediatrician responsible for metabolic disorders. Reliable results cannot be obtained using cord blood because of the risk of maternal contamination.

The following tests should be done when the baby is between 24 and 48 hours old:

- Blood acylcarnitines
- Urine organic acids
- DNA mutation analysis (with the introduction of newborn screening for MCADD, in most cases the genotype will be known for the index case). In the case of an affected sibling, urgent targeted mutation testing (for the known family mutation) should be requested and clearly marked on the

request. If the family MCADD proband is not a full sibling, mutation analysis should be requested only if the biochemical tests are suggestive of MCADD

2. MANAGEMENT

It is important to make sure that the baby has a good milk intake to prevent problems. A term baby should be fed at least every four hours and a preterm baby at least every three hours, from birth. If target feed volumes are not met by either a bottle or breast fed baby, then a nasogastric tube should be passed and feeds topped up to provide target volumes for age and weight. It is not necessary to monitor blood glucose, unless clinically indicated.

2a. Bottle fed babies

Bottle fed babies should be fed on demand, at least 4 hourly. Feed volumes should be gradually increased during the first week from around 20ml/kg on the first day to about 150ml/kg by 7 days of age.

2b. Breast fed babies

Breast fed babies are at particular risk of problems in the first 72 hours, because the initial supply of breast milk is low in volume and energy content , and only small volumes are consumed. Top-up bottle feeds of infant formula are therefore advised for the first three complete days (72hours). Some mothers may wish to try and express breast milk for top-ups.

Feeding guidelines as follows:

- the baby should be breast feed for at least 10 minutes (8 times/day), observed to check they have latched on well, and have a slow rhythmical suck and swallow (ie good technique).

- the baby should be given a top-up feed of infant formula
  Day 1 - 25ml/kg
  Day 2 - 40ml/kg
  Day 3 – 60ml/kg

- if top- up feeds are not tolerated the baby should be given these as continuous nasogastric feeds or IV fluids instead

Discharge plans

A judgment should be made on the baby's feeding ability before considering discharge, with the parents having clear instructions to return to hospital if feeding is poor.

3. PROBLEMS

If the baby seems drowsy or unwell *in any way*, transfer to the neonatal unit urgently and give an intravenous bolus of 2 ml/kg 10% glucose followed by an infusion of glucose 10% (or glucose 10%/saline 0.18% if available) at 100 ml/kg/day. Change to 10% glucose/0.18% once available – for instructions on how to make these fluids click here.

If there is no oral intake, the volume should be increased over 3 days to 150 ml/kg/d. Monitor blood glucose and plasma electrolytes but base treatment on the clinical state (since hypoglycaemia occurs at a late stage, see above).

If enteral feeds are not tolerated, start an intravenous infusion of 10% glucose (or glucose 10%/saline 0.18% if available) at 100 ml/kg/day. Change to 10% glucose/0.18% once available – for instructions on how to make these fluids click here.

If there is no oral intake, the volume should be increased over 3 days to 150 ml/kg/d. For more dietary information please refer to the dietary guidelines on the BIMDG website (http://www.bimdg.org.uk/guidelines).

Seek specialist help from metabolic centre if there are any problems or any points are not clear.

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