

**Care Pathway for Women who have a family history of MCADD**

 **Disclosed at booking**

Medium-chain acyl-CoA dehydrogenase deficiency (MCADD) is a rare inherited disorder where the body cannot metabolise fat properly. If both parents are MCADD carriers, there is a one-in-four chance of their child being born with MCADD. These babies are especially vulnerable to sudden death in the first hours and days of life and so before the diagnosis can be confirmed or excluded a special feeding regimen needs to be established in addition to breastfeeding.

**BIMDG 2010**

 Seen by midwife for first contact appt at app 8-10 weeks, Midwife to ensure during booking woman is asked as per booking proforma page 4 woman is asked about family history of metabolic disorders, mentioning specifically MCADD

National standards

Family with a history of MCADD

No family history of MCADD, continue to routine newborn screening through 5-8 day blood spot

Contact the screening team on ext 7311 or the genetic counselling services on ext 4825 or complete a pregnancy genetic referral form from the community office or genetic screening office.

Screening team or Genetic specialist will contact the woman and discuss family history i.e. family tree with the woman

If Family history is confirmed , Consultant obstetrician informed and appointment made for consultation during the pregnancy with a consultant peadiatrician. The consultant paediatrician provides advice and agrees a birth and post-birth plan (including a special feeding regimen and early access to testing) with the services caring for the pregnant woman, and supply guidance and information to the parents/carers.

Declined ultrasound

The UKNSPC guidance emphasises the importance of taking a family history and recommends that:

“If a mother or her partner has a family history of MCADD you should make a referral to a paediatrician or genetic counsellor for advice. This advice should form the basis of a birth plan (making sure the birth plan is written in the mother’s notes). Depending on the risk of the baby having MCADD, the parents may be advised that their baby needs early screening for MCADD. A sample of blood should be collected 24-48 hours after birth on a blood spot card marked ‘Family history of MCADD’. The parents will also be given information about any special treatment required after the birth of their baby

BIMDG guidelines for care of the newborn with family history of MCADD

 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. It is essential to manage these patients appropriately at this time. Obviously, it is helpful to establish promptly whether the baby is affected since, if they are not, it allows everyone to relax.

**1. DIAGNOSIS**

When the mother is admitted in labour (or, failing this, when the baby is born) inform the consultant paediatrician responsible for patients with metabolic diseases. 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 spot 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)

Discuss urgent testing with the metabolic laboratory and write on the form - family history of MCADD.

**2. MANAGEMENT**

It is important to make sure that the baby has a good milk intake. A term baby should be fed every four hours from birth and a preterm baby every three hours. There is a particular risk of problems in the first 72 hours in *breast-fed* babies, since the supply

of breast milk is often poor at this stage. These babies may well need top-ups of formula milk. Because it is hard to know how much breast milk is being taken, there is a strong case for giving top-ups routinely for the first few days until a good milk supply is established.

**3. PROBLEMS**

If the baby is not taking adequate volumes orally, transfer to the neonatal unit and feed by naso-gastric tube.

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 to make this solution click here). If there is no oral intake, the volume should be increased over 3 days to 150 ml/kg/d.

If the baby seems drowsy or unwell in any other 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 to make this solution 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).

For more dietary information please refer to the dietary guidelines on the BIMDG website (click here)

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

**The British Inherited Metabolic Diseases Group (BIMDG).** This guidance can be accessed via http://www.bimdg.org.uk/guidelines.asp (select emergency protocols, then children).