

Rapid Response Report

NPSA/2011/RRR002

From reporting to learning

26 October 2011

Keeping newborn babies with a family history of MCADD safe in the first hours and days of life

Issue

Medium-chain acyl-CoA dehydrogenase deficiency (MCADD) is a rare inherited disorder where the body cannot metabolise fat properly. With a regular intake of food, individuals can live a normal healthy life, but prolonged fasting or illnesses with vomiting can lead to encephalopathy, coma or sudden death. The disease affects about one in 10,000 babies born in the UK but if both parents are MCADD carriers, there is a one-in-four chance of their child being born with MCADD. In the first 2-3 days of life, when regular feeding is not fully established, new born babies are heavily dependent on fat metabolism for their energy needs and those with MCADD are especially vulnerable to early neonatal death.

Screening for MCADD is part of the UK new born screening programme, which is offered to all babies in England at 5-8 days of age. A baby with a family history of MCADD should have special rapid testing 24 to 48 hours after birth on a blood spot card marked 'Family history of MCADD' (see resources from the [UK Newborn Screening Programme Centre \(UKNSPC\)](#)) but must also be given a special feeding regimen from the moment of birth (see resources from the [British Inherited Metabolic Diseases Group \(BIMDG\)](#)). Breastfeeding should be encouraged, but needs to be supplemented for babies born to families with a history of MCADD to minimise the risk of early neonatal death.

Evidence of harm

Between 1 January 2006 and 30 June 2011, the National Reporting and Learning System received two reports of deaths of newborn babies from MCADD who were born to families with a history of the disease. It appeared that although the mothers had mentioned the family history to healthcare staff when they were pregnant, the staff were not aware of the significance of MCADD, and therefore did not arrange any specialist referrals, special feeding regimen or observation. Six additional 'no harm' incidents reported to the NRLS indicated similar omissions, fortunately without adverse effects.

For IMMEDIATE ACTION by all General Practitioners, NHS organisations providing obstetric, midwifery, neonatal or paediatric services, and specialist centres for inherited metabolic disease. The deadline for action complete is 26 April 2012.

1. Local organisations should ensure that midwives, general practitioners and medical staff working with pregnant women are reminded of the importance of taking an accurate family history at booking and acting on any mention of a family history of MCADD by a pregnant woman (in line with UKNSPC and BIMDG guidance). A clinical briefing sheet is supplied to help inform these staff.
2. Obstetric and midwife-led services should review their patient documentation to ensure there are appropriate triggers for asking new patients about family history of inherited diseases.
3. Obstetric, midwife-led, neonatal and paediatric services should ensure all relevant local and/or national guidance on MCADD is available to their staff.
4. Specialist services for inherited metabolic disorders (adult or paediatric) should ensure that when a baby, child, or adult is diagnosed with MCADD they have reliable systems for routinely informing the family that any future babies will need a special feeding regimen and observation in the first hours and days of life, so that family members are equipped to self-advocate. If informed about any at risk pregnancies, a notification must be sent to the relevant obstetric and neonatal units so that the appropriate guidance is followed after delivery.

Whilst this RRR is specific to MCADD, NHS organisations providing obstetric services may wish to use it as a trigger to review the effectiveness of their pre-birth care planning for all congenital conditions.



Further information

Supporting information for implementation of this RRR, (including links to the UKNSPC and BIMDG resources), and a clinical briefing sheet: www.nrls.npsa.nhs.uk/alerts. For further queries contact rrr@npsa.nhs.uk; Telephone 020 7927 9500.

* In Wales, routine newborn screening for MCADD does not currently take place, although diagnostic testing is carried out where clinically indicated, including babies born to families with a history of MCADD. Preparations are underway for routine screening to be introduced during 2012.